

**Neurofibromatosis Type 1 (NF1): Family Experiences and  
Healthcare Management of a Genetic Syndrome Characterised by  
a Highly Uncertain Phenotype**

Submitted by Daniele Carrieri, to the University of Exeter as a thesis for the degree of Doctor of Philosophy in Sociology, September 2011.

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## Abstract

Neurofibromatosis Type 1 (NF1) is a dominantly inherited disorder (births incidence: 1/3000) with a high spontaneous mutation rate. NF1 has been described as a *condition without parameters*; physical features, cognitive symptomatology, and complications such as malignancy, are highly variable, both within and between families, and over the lifetime of affected individuals. This thesis explores the significance of the recent classifications of NF1 as a 'genetic syndrome', in terms of the subjecthood of affected individuals, their family experiences and the way it is managed within the healthcare system. The research is based on qualitative semi-structured interviews of NF1 individuals, their families (n=30) and healthcare professionals who work with NF1 (n=11) and employs Grounded Theory and Narrative Analysis inspired methods of analysis. As such, it provides an empirical investigation of many of the sociological theories which have been developed in response to genetic disease, particularly genetic responsibility, biocitizenship and the medicalization of the family.

NF1 was still experienced by patients and treated by the healthcare system, as a condition without parameters i.e., as a disparate set of symptoms with uncertain meaning, rather than as a 'whole'. The majority of the respondents - regardless of the severity of NF1 - rejected NF1 genetic identities, employing diverse downplaying strategies to normalise it. NF1 was salient at certain critical junctures in individuals' lifecourses, especially in relation to reproductive choices, disclosure and management of pressing symptoms. Family experiences with genetic conditions, the relations of family or kinship, health behaviours, familial surveillance and disclosure did not necessarily follow the lines of biomedical knowledge and genetic inheritance. The analysis also revealed a degree of mirroring between the structure of the healthcare provision for NF1 and patients' constructions of the condition, for example the lack of illness identity. The example of NF1 shows that the identification of the genetic basis of a condition does not necessarily provide patients and healthcare professionals with more parameters to manage it.

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# **1. Introduction**

## **1.1 Scope of the Thesis**

This thesis explores the place of genetics in patients' experiences with neurofibromatosis type 1 (NF1) - a relatively unknown, but common genetic condition (average birth incidence: 1/3000 worldwide). NF1 is a very complex disorder: physical features, cognitive symptomatology and complications such as malignancy are highly variable and unpredictable, both within and between families, and over the lifetime of affected individuals. In its extreme forms, NF1 can lead to diverse typologies of tumours which can be life-threatening and perceived as disfiguring. NF1 can also cause learning difficulties and other neurological symptoms. The disorder has been strongly associated with 'monstrosity' images which in mildly or seriously affected individuals can lead to stigmatization and/or psychological distress (Huson and Hughes 1994; Ablon 1995; Legendre, Charpentier-Cote et al. 2011).

This issue has been explored by Ablon, who has conducted the most extensive qualitative-sociological research on NF1 patients to date (Ablon 1992; Ablon 1995; Ablon 1996; Ablon 1999; Ablon 2000; Ablon 2002). From her analysis of illness narratives, she found that alongside feelings of shame and stigma, the unpredictability of NF1 was one of the most damaging psychological aspects of the disorder (Ablon 1992; Ablon 1999). The extreme variability and unpredictability of the condition, that is the lack of a clear prognostic model, was a very common cause of anxiety in patients. While many conditions like achondroplasia, Huntington's disease or Duchenne muscular dystrophy have relatively constant prognostic outcomes, the highly unpredictable and variable nature of NF1 leaves patients with no clear ideas about the development of their disorder.

Patients with achondroplasia may not know how high they will grow, and individuals with Huntington's disease may not know when and how they will develop the disease, but NF1 is variable and uncertain beyond this. Individuals diagnosed with NF1 may not know whether they will develop life-threatening and/or disfiguring tumours, cognitive impairments or simply a few moles. To capture this feature, Ablon (1999) called NF1 a *condition without parameters*.

In the 1990s, NF1 became a genetic syndrome (Riccardi 1999; Ruggieri and Huson 1999) - i.e., a genetic basis (the 17q11.2 gene on chromosome 17) was attributed to the cluster of variable symptoms and features that are identified as characteristic of the syndrome (Latimer, Featherstone et al. 2006). This thesis is a sociological examination of the impact of this genetic categorization on patients' and families' experiences and their and professionals' experiences of healthcare service provision. That is, does categorising NF1 as a genetic syndrome bring coherence to 'a condition without parameters' in the experience of individuals, families and the healthcare system?

In recent years, there have been growing expectations of the expansion of genetic practices in healthcare (e.g. genetic testing, screening, diagnosis). Genetic medicine has been represented within the public health sector, the media and healthcare professionals - to cite only a few - as being capable of delivering important knowledge about diseases, predispositions, behaviours and other individual traits. 'Genetic innovation' has been likened to promises of improving control over our biological makeup, minimizing disease and risk and ultimately increasing our future wellbeing.

Stemming from this context of 'geno-hype' (Lock 2006), an increasing number of social scientists have become interested in exploring the potential and actual impact of genetics on patients, healthcare and society. This relatively young, yet prolific, research

enterprise has engaged with pressing questions related to the extent to which genetics may alter the way individuals experience their health, illness and, ultimately, their understanding of their selves. Several scholars have advocated the idea that as genetic practices are significantly entering the medical - and by extension social - domain, they are redefining disease and health and transforming individuals' subjectivity (Lippman 1992; ten Have 2006; Vailly 2006). Individuals have been described as incorporating genetic information into their everyday lives and increasingly understanding their risk or disease - and potentially their personhood - in genetic terms (Lippman 1998; Kerr and Shakespeare 2002; Petersen and Bunton 2002; Rose 2006). These claims are encapsulated in the notion of genetic responsibility - the idea that genetic information and technologies place individuals in a web of relations of duty and responsibility to their family members, society and themselves, influencing life plans such as reproductive decisions and career aspirations (Hallowell 1999; Novas and Rose 2000; Petersen and Bunton 2002). Given the familial implication of genetic knowledge, a number of scholars have also specifically analysed the impact of genetic medicine on the construction of the family (Cox and McKellin 1999; Finkler 2000; Featherstone, Atkinson et al. 2006). Some have argued that - alongside individuals' subjectivity - genetic practices are also changing ('medicalizing') family bonds and relations, strengthening the ties of biological relatedness (Finkler 2000). On a broader social scale, genetic responsibility is linked to the notions of biocitizenship and biosociality (Rabinow 1996; Rose and Novas 2004), that is, the idea that individuals are increasingly seeking social support based on shared biological identities and activism.

These theoretical constructs dovetail with broader public health discourses which - even before the advent of the 'genetic revolution' in healthcare - promote a model of 'responsible citizens' (and patients /families) who autonomously draw on scientific and

technological innovation in order to increase the knowledge about their health and to engage in risk-minimization behaviours (Petersen and Lupton 1996; Petersen and Lupton 1997; Petersen and Bunton 2002). Moreover, similar to the public health discourses, many of these sociological constructs - whilst acknowledging the complexity of genetic information - rest nonetheless on the assumption of a link between genetic information and a sense of certainty and identity (a 'genetic knowledge about the self'). Genetic testing and diagnosis are regarded as providing significant information on one's present and future health and, thereby, leading to a significant improvement in prevention, treatment and intervention capacities.

However, it is important to stress that the application of genetic technologies in medicine also produces uncertainty. Genetic tests provide probabilistic estimates which can be complex for healthcare professionals and patients to interpret. Often genetic diagnoses do not coincide with clear prognoses; conditions which are characterised as being partly or wholly dependent on genetic factors can still be variable and unpredictable. It can also be difficult to predict genes-genes, genes-environment interactions both in cases of complex multigenic disorders or predispositions and in cases of monogenic disorders with clearer prognostic pathways (Kitcher 2003).

Drawing on these fundamental considerations, I examine the complex, variable and unpredictable condition of NF1 in order to explore the views and experiences of patients, families and professionals. I investigate what it means for affected individuals, both those who have been diagnosed with this condition and their families, that NF1 is considered a 'genetic syndrome' and what are the implications for the healthcare system. Thus, I provide an empirical investigation of the notions of genetic

responsibility, biocitizenship and medicalization of the family in the context of a highly unpredictable genetic condition.

These theoretical constructs often use examples such as Huntington Disease (HD), or Hereditary Breast and Ovarian Cancer (HBOC), whose profiles are less complex and variable than those of multisystem genetic syndromes. I provide, instead, an empirical investigation of genetic responsibility, biocitizenship and medicalization of the family based on - in Ablon's vivid words - a *condition without parameters* (Ablon 1999).

There is undoubtedly a certain degree of uncertainty in HD and HBOC. Although HD is highly penetrant, individuals may not know the onset and severity of their disease.

HBOC can cast a spectre over individuals' lives for decades as they may not know whether or not they will get cancer.

However, NF1 is more complex condition with much more variable and unpredictable manifestations. It can cause different physical and cognitive symptoms (e.g. bone lesions, benign tumours, malignant life-threatening tumours, visual impairment, learning difficulties etc.). Moreover, it can be life-threatening to a tiny minority, highly disfiguring and stigmatising to others, it can develop in unpredictable ways during patients' life courses, but it can also be very mild.

I investigate whether the transformation of patients' subjecthood, contained within ideas of genetic responsibility, biocitizenship and medicalization of the family occurs when a multisystem, highly variable and unpredictable syndrome like NF1 is identified as being 'genetic'. This research is sociologically relevant, as it poses important questions on the pervasiveness of these notions. The uncertainty and variability of NF1, that is, the lack of a 'role model', or constant prognostic parameters, may prevent patients and families from understanding their syndrome as a 'whole' and thereby endorsing a NF1-specific disease identity. The neurologic and cognitive symptomatology could also represent

another important mitigating factor, as it may hinder patients' capability to understand the complexity of their syndrome.

At the heart of my research lies an important tension between the complexity and uncertainty of genetic information and the promises of genetic engineering to increase our knowledge of - and the possibility to intervene on - disease, health and other human traits (behaviours, psychiatric disorders etc.) and to 'know ourselves better' through knowing our genome. As I aim to contribute to the clinical management of complex genetic disorders, I also consider NF1 because it is a relatively common condition, hence an important disorder in terms of health policy.

Overall, this thesis investigates:

- (1) The relevance of the notions of genetic responsibility, medicalization of the family, biocitizenship and biosociality to NF1, that is, the ways in which knowledge about the genetic basis of NF1 influences individual and familial experiences with the syndrome, the definition of the self, the management of the condition and the use of structural support such as support groups.
- (2) The interrelation of individual and familial experiences of disease, particularly individual and familial meaning making practices around uncertainty.
- (3) How patients and families are managed by the healthcare system, how they experience it and how genetic classifications play (or do not play) into this.

This thesis aims to contribute to: (i) sociological literature on the meaning and impact of genetic medicine at individual, familial and social level; (ii) the available qualitative psychosocial research on NF1; (iii) policy making and clinical practice about the needs of service users with this and similar genetic syndromes.

The research is based on semi-structured qualitative interviews with individuals with NF1, their family members and healthcare professionals involved in the treatment of the condition. The experiences of family networks' with NF1 have been accessed through the sampling and interviewing of many individuals from the same family.

Individuals are recruited both from the NHS sector and from other sources in order to collect a variety of experiences (from individuals and families with different manifestation of the syndrome). This allows a more representative sample of individuals and family with NF1. The research combines Narrative and Grounded Theory inspired methods of analysis.

To my knowledge, this thesis represents the first in-depth study of the impact of the genetic categorization of NF1 on patients' and families' illness identities and on the healthcare management of this condition. Previous sociological qualitative work on NF1 (mainly Ablon's research) took place before the recent stress on the genetic causality and heritability of the syndrome and before the explosion of literature on the implications of genetic diagnosis and uncertainty.

Alongside the investigation of the genetic aspects of NF1, another key point of interest and originality of this thesis is the focus on the familial implications of genetic knowledge. These two points of originality are closely tied together as genetic information has familial implications. Genetic information and risk are distributed among blood relatives and family networks, potentially having significant repercussions on family relations and bonds. Tapping the familial experiences of genetics and disease offers a precious insight into how family dynamics and processes can influence the way patients face genetic conditions. Thus, the study of the experience of genetics and family patterns of disease within families can produce a more nuanced understanding of

the psychosocial impact of medical genetics. Furthermore, investigating how individuals and their family networks experience genetic syndromes has significant clinical implications. It can highlight aspects of patients and families' experiences which may not be always visible in clinical settings, ultimately contributing to more patient-oriented services.

Although the importance of studying the social implication of genetic practices within the familial context has been recognized (Cox and McKellin 1999; Fanos and Puck 2001; Downing 2005; Hallowell, Arden-Jones et al. 2005; Featherstone, Atkinson et al. 2006; Williams, Skirton et al. 2009), research still tends to focus primarily, or mainly, on individuals' accounts about their families and the psychological implications of receiving an informative test result. Few studies examine how affected patients and their families jointly construct the meaning of genetic risk within their everyday lives and how, in turn, such pre-existing constructions shape the experience of genetic risk (Strauss and Corbin 1990). Moreover, the empirical research on experiences with genetic disorders which has focused on familial dynamics around genetic knowledge is normally based on interviews of one or at most two individuals within the same family (parents or partners) and often analyses individuals in the family as separate units. This research project aims for an extensive exploration of family experiences and dynamics, achieved by interviewing as many family members as possible within the same network and by triangulating individual and familial accounts.

## **1.2 Structure of the Thesis**

The next two chapters set the medical and theoretical background of this project.

**Chapter 2** introduces NF1. After delineating some of the most salient moments in the history of this complex disorder and its link to images of 'monstrosity', it summarises the relevant biomedical studies on this condition, providing an essential scientific and



clinical backdrop to understanding the accounts of the patients, families and healthcare professionals interviewed. The chapter continues with an analysis of the existing psychosocial and qualitative studies on NF1 and will highlight the differences and the contributions of this dissertation.

**Chapter 3** reviews relevant literature on the sociology of genetic medicine, with a particular focus on research that addresses the impact of genetic medicine at the individual, social and familial levels. This chapter also set the research questions and provide a rationale for undertaking sociological research on individual and familial understandings of NF1.

**Chapter 4** presents the study methodology. First, it discusses the suitability of qualitative semi-structured interviews for addressing the research questions. Second, it explains the significant point of novelty of the methodology: the systematic analysis of family networks. The family level analysis is an original, or, at least, underused technique. The strengths and weaknesses of this approach are discussed. The chapter concludes with a description of the research process.

**Chapter 5** takes a narrative approach to the analysis of one specific family from the sample and illustrates the links between individual and familial experiences with genetic disorders. **Chapter 6** presents findings related mainly to the impact of genetic knowledge at the individual and social level, whilst **Chapter 7** tackles the familial dimension of understanding and negotiating living with NF1. These two chapters draw on the interviews with patients and families only. Finally, **Chapter 8** combines healthcare professionals and patients and families' interviews to address the issue of the healthcare system management of this disorder and its impact on illness experiences. Findings are summarised in the concluding **Chapter 9**. The chapter also presents a discussion of the theoretical and policy implications of this thesis, final reflections on its weaknesses and suggestion for future research.

## **2. Neurofibromatosis Type 1 (NF1)**

### **2.1 Introduction**

The following chapter aims to provide the reader with background knowledge on NF1. It collects and discusses information which is very important to the understanding of the nature and rationale of this research project. The chapter begins with a brief sketch of the history of NF1 (§ 2.2), to proceed with a review of the medical (§2.3), scientific (§2.4), cognitive, psychological and qualitative research (§2.5) on this syndrome. The chapter will also start to delineate the research gap that this thesis seeks to redress (§ 2.6).

### **2.2 Historical Notes**

#### **“Prodigies of Nature”**

Reported cases of individuals who probably have NF1 or variants of NF1 have been found over the centuries, dating as far back as the 2<sup>nd</sup> century AD (Huson and Hughes 1994; Ruggieri and Huson 1999; Ruggeri and Tenconi 2001). All these testimonies attest extreme cases of physical disfigurement and belong to recordings of “prodigies of nature” and/or “monsters” (Zanca and Zanca 1980). This depiction has traversed history and, as will be shown, is still present nowadays.

Several illustrations and descriptions of both historical and medical importance have been discovered in manuscripts dating from the 10<sup>th</sup> to 16<sup>th</sup> century (Zanca and Zanca 1980). The first clinical description of NF1 is currently attributed to the Italian physician, philosopher and naturalist Ulisse Aldovrandi, who in 1592 recorded a case known as the ‘Homuncio’. The ‘Homuncio’ was described as a man of Indian origin of short stature with soft fleshy masses hanging from the left side of his head and chest.

This report appeared in 1642 in Aldovrandi's book called 'Monstrorum Historia' (Aldovrandi 1642).

In the 18<sup>th</sup> century, it is possible to find in Buffon's 'Histoire Naturelle' drawings of a child, known as 'Buffon's girl', affected by skin tumours and pigmented lesions (Hecht 1989). Cruveilhier's 'Anatomie Pathologique du Corp Human' (Cruveilhier 1829) also presents technical illustrations of pathological studies of NF1.

There also seems to be a potentially interesting (and to my knowledge unexplored) link between the facial appearance of certain extreme cases of NF1 and cubist portraits.<sup>1</sup>

Looking in particular at many of Picasso's portraits, it is possible to hypothesise that among his influences, there may also be the possibility that the artist encountered one or more individuals with NF1. Many artists have been interested in diseases and in portraying individuals affected by different form of ailments (see for example Velasquez and Goya who were important points of reference for Picasso (Warncke 2006)).

The first clinical study in the English language is that of Akenside in 1768 (Akenside 1768) who was also the first to report that NF1 could occur in more than one family member. The first review of the disease was published in 1849 by the Dublin professor of surgery Smith (Smith 1849).

The term neurofibroma was coined for the first time by von Recklinghausen in 1882 in his report titled 'On Multiple Cutaneous Fibromas and their Relationship to Multiple Neuromas' (Recklinghausen 1882). This is why neurofibromatosis is also known as von Recklinghausen's disease.

In 1900, Thomson undertook the first systematic genetic survey on 77 individuals reporting that NF1 was familial in 30 cases. But the question of the mode of inheritance was solved only in 1950 by the studies of Borberg (Borberg 1951) and Crowe, Schull

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<sup>1</sup> I have to thank one of my interviewees with NF1 for raising this point.

and Neel (Crowe, Schull et al. 1956); both studies concluded that there was an autosomal dominant pattern of inheritance of NF1 with sporadic cases of new gene mutations.

NF1 has been essentially unknown to the general public until it became identified as ‘The Elephant Man Disease’ through the publication in 1971 of Montagu’s ‘The Elephant Man: a Study on Human Dignity’ (Montagu 1971). Montagu’s book was centred on the major essay of the prominent 19<sup>th</sup> century surgeon Sir F. Treves ‘The Elephant Man and Other Reminiscences’ published in 1923 (Treves 1923). Joseph Carey Merrick was a young English man, so seriously disfigured that he lived a miserable life as a carnival attraction in the famous “freak shows”. In 1884 Treves brought Merrick to a London hospital and let him live there (in a sheltered place) for the next four years until he died at age 25. Merrick’s condition was diagnosed as NF1 early in the 20<sup>th</sup> century. In the 2<sup>nd</sup> half of the 20<sup>th</sup> century, alongside Montagu’s and other books (Howell and Ford 1980; Drimmer 1985), several plays and a film<sup>2</sup> drew attention to Merrick’s case, leading some of the public to consider the Elephant Man to be the symbol and the stereotype for NF1 (Ablon 1995).<sup>3</sup> According to some scholars, the expression ‘The Elephant Man Disease’ became a metaphor for the “grimmest extreme of ugliness” (Ablon 1995; Ablon 2000).

However, current clinical studies claim that Joseph Merrick was affected by Proteus syndrome and not by NF1 (Tibbles and Cohen 1986; Cohen 1988). For example, in examining Merrick, Weber, one of the most famous dermatologist of the time, did not find any sign of the pigmentary lesions typical of NF1 (Ruggeri and Tenconi 2001).

Moreover, it has to be noted that Merrick’s case was particularly extreme, both with

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<sup>2</sup> D. Lynch ‘The Elephant Man’ 1981

<sup>3</sup> There are other famous historical and literary cases of possible NF1. Among the literary ones we can quote Quasimodo, the Hunchback of Notre Dame, who was depicted by Victor Hugo as having neurofibromatosis. Again this famous case represented a serious manifestation of NF (Ruggeri and Tenconi 2001; Hugo ed. 1999) .

respect to Proteus syndrome and NF1. Nevertheless, the consequences of the initial diagnosis have left a vivid and symbolic image in the mind of the general public (especially two decades ago), bolstering the association between the Elephant Man and NF1.

Some clinicians have claimed that the enormous publicity of Joseph Merrick's story generated by books, films, plays and other media has had (at least in the US) consequences for the research on NF1, generating interest in scientists and funding related research (Ablon 1995; Ruggeri and Tenconi 2001). Equally, the link between NF1 and the Elephant Man had a considerable negative impact on the image associated with the syndrome. For example, Cohen found it increased the distress of patients diagnosed with NF1 and their family members (Cohen 1988). In attempting to put a halt to the widespread and alarming misuse of the image of the Elephant Man both within the media and the medical environments, Michael Cohen Jr., one of the clinical geneticists who re-diagnosed Merrinck's disorder as Proteus syndrome, coined the vivid expression "Elephant Fever"(Cohen 1988).

There are also numerous media representations of NF1 in current circulation. In 2004, there was the case of a Romanian woman with an 80kg tumour shown on a programme entitled 'Megatumour'; she was treated in the US. Probably even more sensational is the recent case in 2007 of a Chinese man with a 15Kg tumour on his face operated on in China.

In 2011 a programme called 'Beauty and the Beast' was aired on Channel 4 in the UK. The programme matched people with 'superficial' beauty issues with those with facial disfigurements. One of the episodes featured a person with NF1 with disfiguring facial neurofibromas, presenting, once again, an extreme image of NF1.

These depictions of NF1 clearly suggest some links with the past representation of extreme cases evoked above. Media representations of less extreme and much more common NF1 cases are more rare and difficult to find.

Despite media examples, it seems overall that NF1 is not a very well-known condition. The historical appearances of NF1 have at most provided famous ‘shock’ examples, without making the syndrome a known and debated condition among the general public.

### **Support Groups**

In the 1970s, the first support organizations for NF1 were established. The stated mission of these organizations was to improve the quality of life of individuals with NF1 and their family networks by connecting different families in order to share experiences, circulating accessible and correct information and, above all, promoting the study of and research into the syndrome. The first organization in the world was the National NF Foundation (NNFF, from 2005 known as the Children’s Tumour Foundation <http://www.ctf.org/>). It was founded in 1978 by Lynn Countermanche (a nurse with NF1), her physician Dr. Rubenstein and the attorney Hirschritt. The UK association (founded by Clare Webb and Trish Green) <http://www.nfauk.org/> followed in 1982. This association has changed its name several times, moving from ‘Let’s Increase Neurofibromatosis Knowledge (LINK)’ to ‘The Neurofibromatosis Association’ and very recently, in the year 2010, it became ‘The Neuro Foundation’. Thanks also to the activity of the USA and UK associations, the first NF research program in the world started in 1984. In 1988 an International Symposium, the National Institute of Health Consensus Development, was held and the diagnostic criteria still in use for NF1 (see table 1) were first developed. In the same year NF1 and NF2 were distinguished and their genes were mapped to the respective chromosomes. In 1990 the NF1 gene was cloned, opening the possibility for more studies to look at genotype/phenotype correlations. In 2000 molecular testing for NF1 was made

available; a test can identify mutations in more than 95% of individuals meeting the diagnostic criteria (Radtke, Sebold et al. 2007).

On the internet, apart from the health and support organizations sites, there are few forums or blogs in which NF1 is discussed. An exception in the UK is represented by the blog of a young girl, Kirsty Ashton, who tries to publicly share her daily life living with NF1, as well as providing empathic advice to those affected with the same condition. Kirsty Ashton's involvement in children charity activities has earned her the Manchester Pride prize in 2005 <http://www.kirstysstory.co.uk/index.htm>. Furthermore, very recently (in 2011) the Neuro Foundation launched its Facebook page which may serve as an additional opportunity to link individuals with NF1 (who have access to and are able to use computers and internet).

The US equivalent to Kirsty Ashton's blog is that of Reggie Bibbs, called 'Just Ask: it's NF' <http://www.reggiebibbs.com/>. It is also worth mentioning the charitable and fundraising activities of Gillian Anderson (X-Files actress) in spreading the awareness of NFI <http://www.gilliananderson.ws/charities/nf.shtml>.

As will be explored in Chapter 3, the anthropologist Paul Rabinow in his famous essay 'Artificiality and Enlightenment: From Socio-Biology to Biosociality' (Rabinow 1996) specifically refers to the NF associations as an existing example of what he calls 'biosociality' i.e. the formation of new social and political groups related to a specific genetic profile where people align themselves in terms of genetic narratives and practices:

There already are, for example, neurofibromatosis groups whose members meet to share their experiences, lobby for their disease, educate their children, redo their home environment and so on. That is what I mean by biosociality (Rabinow 1996:102).

According to Rabinow, the NF support groups and similar organizations have also the aim of resisting social prejudice and pressure of normalization, sensitizing the public opinion.



**FIGURE 1**

A Juxtaposition I made of Picasso's 'Woman with a Red Beret' (on the left) and Reggie Bibb's self-portrait/branding of his foundation (on the right)





FIGURE 2

1642: an illustration of the Homuncio from Aldrovandi 'Monstrum Historia'



FIGURE 3

1749: a painted illustration of Buffon's girl from 'Histoire Naturelle'



**FIGURE 4**

1862-1890: a picture of Joseph Merrick



**FIGURE 5**

2007: a picture of Huang Chuncai, the Chinese man with a 15Kg facial tumour

## 2.3 NF1 Medical Background

At present, the major classified forms of neurofibromatoses are neurofibromatosis Type 1 (NF1) and Neurofibromatosis Type 2 (NF2) (NIH 1988; Ferner 2007; Boyd, Korf et al. 2009). NF1 the most common form of Neurofibromatosis and, although it is relatively unknown, one of the most prevalent autosomal dominant disorders in human beings (Huson, Compston et al. 1989; Huson and Hughes 1994). NF1 is treated and understood as a genetic syndrome i.e. a collection of features and symptoms that are identified with a genetic cause. NF1 is a complex disorder which involves potentially all organ systems, principally the skin, iris, skeletal and nervous system. NF1 is related to a loss of neurofibromin, a protein which serves as regulator for cell proliferation and differentiation and is particularly expressed in the nervous system. The loss of neurofibromin leads to an increased risk of developing benign and malignant tumours. NF1 is characterized by major clinical features<sup>4</sup> - which also represent the diagnostic criteria (see Table1). These include multiple café-au-lait spots (CALs), freckling (mainly axillary and inguinal), Lisch nodules, optic pathway glioma, osseous lesions (bony dysplasia) and neurofibromas.

With regard to what can be described as minor clinical features, learning difficulties are estimated to occur in a range between 30-50% of affected individuals. Scoliosis, bony dysplasia, pseudoarthrosis and overgrowth are the most common skeletal complications. Less common, but more serious complications include plexiform neurofibromas, optic and other central nervous system gliomas, malignancy, osseous lesions, vasculopathy and hypertension (Rubenstein and Korf 1990; Huson and Hughes 1994; Ruggieri and Huson 1999; Tonsgard 2006; Ferner 2007).

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<sup>4</sup> See below for a description of the major and minor clinical features.

The average birth incidence of NF1 is 1 in 3000 and this syndrome is equally prevalent among both sexes and all ethnicities (Descheemaeker, Ghesquiere et al. 2005; Levine, Materek et al. 2006). Being an autosomal dominant disease means that if either parent has NF1, their children have a 50% chance of having the disease. In approximately half the individuals who have NF1, the condition also appears in families with no previous history of the condition, as a result of a new genetic mutation - known as *spontaneous* or *de novo* mutations - in the sperm or the egg (Levine, Materek et al. 2006). Thus, approximately half of the affected individuals are the first in their family to present with the condition. The cause of the high spontaneous mutation rate is still unknown (Friedman 1999; Ruggieri and Huson 1999; Boyd, Korf et al. 2009; Ferner 2010). The appearance of many of the diagnostic manifestations of NF1 is age-dependent. Several studies report that approximately 90% of individuals with NF1 will have two or more diagnostic criteria by 6 years of age, 97% by age 8 and all do so by 20 years of age (DeBella, Szudek et al. 2000; Radtke, Sebold et al. 2007). In some cases, diagnosing a young child without a family history of the condition may be challenging as the symptoms may manifest later in the child's life. For this reason, children may need to be followed for several years before the clinical diagnosis of NF1 can be confirmed.

NF1 is a progressive condition. The severity of its manifestations progresses with age and appears to be affected by hormonal changes such as puberty or pregnancy (Huson and Hughes 1994). NF1 is extremely unpredictable and variable, even within families and even in the same affected individual at different times in life. Affected individuals may present innocuous freckles, others severe, but benign, disfigurements, and a few have cancerous growths and may die prematurely. Moreover, many individuals with the condition may have learning difficulties and other neurological symptoms. These

represent some of the reasons that make NF1 a difficult condition for patients and families to deal with and for clinicians to manage. NF1 is primarily diagnosed referring to the diagnostic criteria developed at the NIH Consensus Conference (NIH 1988) (see also Table 1). Since 1995, molecular testing offers another diagnostic approach.

## **Prognosis**

As mentioned above, there is wide variability and unpredictability with regards to how different individuals with the NF1 gene manifest the disorder. Some individuals may just present freckles and café-au-lait spots, while others may have rapidly progressive disorders, such as serious life threatening tumours which can grow in any part of the body. NF1 can also lead to cognitive and learning difficulties, which, once again, can be very mild or serious enough to require educational assistance. Nevertheless, the disfigurement caused by the cutaneous neurofibromas, pigmented lesions (CALS and freckles), and bone abnormalities (osseous lesions) are normally listed among the primary problems of NF1. The major and minor clinical features are grouped and described in the following paragraph.

## **Major Clinical Features**

### **Café-au-lait spots (CALS)**

CALS are innocuous pigmentary uniform lesions with smooth borders. They vary in diameter from 0.5 to 50 cm and they can cause aesthetic problems. CALS might be present at birth or develop within the first 1-2 years of life. They increase in number in early childhood and tend to fade with age or become obscured by numerous cutaneous neurofibromas (Ruggieri and Huson 1999).

The presence of one or two CALS is quite common in the general population; according to the NIH, CALS are considered a clinical criterion if their amount equals to or exceeds 6 (Ruggieri and Huson 1999; Radtke, Sebold et al. 2007).

### **Skinfold Freckling**

This is another innocuous and typical clinical feature of NF1. The freckles are similar in colour to CALS, but are much smaller (1-3 mm in diameter) and often occur in clusters. They are commonly found in the axillary and inguinal regions, but they can occur also in other areas (e.g. around the base of the neck) (Huson and Hughes 1994; Ruggieri and Huson 1999).

### **Neurofibromas**

Neurofibromas are benign tumours that can occur anywhere in the body. They are divided into two main different types.

1) Cutaneous-subcutaneous Neurofibromas. These are discrete nodules that usually protrude above the skin. They are soft, almost gelatinous (viscous) in consistency and violet in colour. They are normally localized on the trunk, but can appear on any part of the body. Rarely painful, they can cause pruritis (chronic itching). Cutaneous neurofibromas are present in almost all patients with NF1 by age 20 years (Huson, Harper et al. 1988; Upadhyaya, Huson et al. 2007).

2) Plexiform Neurofibromas. This term refers to tumours that tend to grow along the peripheral nerve sheath. Most of them are located on the trunk and are not associated with major problems. However, they may grow into healthy tissue and interfere with normal development of tissue or bone. Those located on the face or other exposed parts of the body may be associated with major disfigurement. Large lesions located elsewhere may be associated with significant bony and/or skin overgrowth (Upadhyaya, Huson et al. 2007). These tumours are usually painful and disfiguring and can lead to impairments. Most of them are benign, but there is a risk of their transformation into

malignant peripheral sheath tumours (MPNST). Usually, plexiform neurofibromas and MPNST cannot be easily surgically removed because of the infiltration of surrounding tissue and nerves and the risk of catastrophic haemorrhage since they tend to be supported by a network of blood vessels (Ferner 2007; Boyd, Korf et al. 2009).

### **Optic Pathway Gliomas**

These are tumours of the optic nerve. They represent the most common central nervous tumour system seen in NF1. Approximately 15% of NF1 patients will present them. They can cause loss of visual acuity, abnormal colour vision, optic atrophy and afferent pupillary defect. Normally optic pathway gliomas do not progress or metastasise; however if this occurs, they can cause precocious puberty or increased growth (Huson and Hughes 1994).

### **Lisch Nodules**

These are pigmented harmless asymptomatic hamartomas (benign tumour-like nodules) of the iris that do not affect vision. They are rarely seen outside the spectrum of NF1 and more than 95% of individuals affected by NF1 present Lisch nodules by the age of 20 (Huson, Jones et al. 1987). They develop during childhood after the appearance of CALS but before peripheral neurofibromas (Huson, Jones et al. 1987; Ruggieri and Huson 1999).

### **Skeletal Dysplasia**

Sphenoid or tibial lesions are very typical of NF1. Scoliosis is also another common and relevant skeletal finding. Occasionally, it is the result of spinal neurofibromas. Severe scoliosis can deform the spine, compromising respiration and movement (Huson and Hughes 1994).

## **Secondary Clinical Features**

### **Neurological- Cognitive Problems**

Neurological complications develop mainly from tumours and malformations.

Intracranial lesions or spinal cord compression may cause sensory disturbance, motor deficit, incoordination and sphincter disturbance (Ruggieri and Huson 1999).

Cognitive problems are the commonest neurological complications of NF1 sufferers, who can present a low average range IQ. In some cases patients present a severe intellectual deficit i.e.  $IQ < 70$ , but this is relatively rare (Ferner, Hughes et al. 1996).

The underlying pathogenesis of cognitive problems in NF1 has not been determined (Ferner, Huson et al. 2007). Recent research suggests that altered expression of neurofibromin in the brain may account for the neurocognitive and neurological complications in NF1 (Ferner, Hughes et al. 1996; Ho, Hannan et al. 2007; Boyd, Korf et al. 2009).

The delineation of the spectrum of cognitive features and their severity varies across the literature. In an extensive review published in 2006, it is reported that patients have academic difficulties particularly in mathematics and reading, and have a high preponderance of ADHD (Levine, Materek et al. 2006). On average about 30%-50% of NF1 children are estimated to suffer from specific learning difficulties (Descheemaeker, Ghesquiere et al. 2005; Johnson, Wiggs et al. 2005; Belzeaux and Lancon 2006; Levine, Materek et al. 2006). There is also an increased frequency of speech impairment, visual and spatial difficulties and autistic spectrum disorders (Maunter, Kluwe et al. 2002; Johnson, Wiggs et al. 2005). Another common finding of neuropsychological studies is that NF1 individuals seem to perform better in verbal than non-verbal tests (Huson and Hughes 1994).



## **Psychological and Behavioural Problems**

Sleep disturbance, impaired socialization, poor interpretation of social cues, social anxiety, mild or severe depression and low self-esteem are often quoted as common features of NF1. Instances of attempted suicide, psychosis and sociopathic behaviour have also been reported. These problems are considered to be the consequence of the disfigurement caused by NF1 to the body and the face and by the reaction of others. The complex, variable and unpredictable nature of the disease represents another important factor contributing to anxiety (Huson and Hughes 1994; Johnson, Saal et al. 1999; Radtke, Sebold et al. 2007).

## **Seizures & Epilepsy**

Epilepsy is estimated to occur in approximately in 6-7% of NF1 individuals and it is usually mild. Likewise, seizures are not a prominent feature of NF1; they are estimated to occur in up to 10% of the patients. They can encompass infantile spasms, primary generalized seizures and partial complex seizures (Vivarelli, Grosso et al. 2003; Radtke, Sebold et al. 2007).

## **Headaches**

Headaches span from mild to serious migraines. Atypical headaches accompanied with neurological deficits might be the sign of a brain tumour or cerebrovascular problems (Clementi, Battistella et al. 1996).

## **Growth**

Individuals with NF1 tend to be below average height. It has also been observed that adult patients frequently display short stature. Patients can also present relative or absolute macrocephaly (abnormally large head), which usually develops during childhood (North 1998; Radtke, Sebold et al. 2007).

### **Onset of Puberty**

Although most patients undergo normal pubertal development, both premature and delayed puberty can occur (Sebold, Lovell et al. 2004; Graf, Landolt et al. 2006; Tonsgard 2006).

### **Pregnancy**

The hormonal changes that accompany pregnancy normally exacerbate the symptoms of women who suffer from NF1 permanently, increasing the visibility and sometimes severity of these women's symptoms. The extent of this increase cannot be predicted in advance. Women with NF1 can present for example new tumours or rapid increase of pre-existing ones, and hypertension can significantly increase. A re-assessment of the symptoms can follow from the post-partum period (Ruggieri and Huson 1999).

Early studies also claim that pregnant women with NF1 may have an increased risk of severe complications like intrauterine growth retardation, pre-term labour and stillbirth. At the moment these studies seem to be controversial, probably because of the reliance upon limited particular cases. More recent articles tend to suggest that healthy NF1 pregnant women seem to be more likely to undergo caesarean section more often than non-NF1 women (Radtke, Sebold et al. 2007).

### **Cardiovascular Findings**

Hypertension is commonly found in NF1 patients. This increases the risk of heart attack, stroke and kidney failure (Zoller, Rembeck et al. 1995; Ruggieri and Huson 1999).

### **Pruritis**

Itching is relatively common. It can be localized or general. Normally it is related to the presence or the insurgence of cutaneous neurofibromas (Radtke, Sebold et al. 2007).

### **Unidentified Bright Objects (UBOs)**

These are also known as 'NF1 spots'. These objects are shown by cranial MRI scans in patients with NF1. They tend to disappear with time and have rarely been found in individuals with NF1 over the threshold of 20 years. Some researchers have suggested connection between UBOs and lower IQ, language skill, visual-spatial functioning and academic achievement, but this hypothesis is still controversial (North, Joy et al. 1994; Radtke, Sebold et al. 2007). It has also been proposed to include UBOs as a diagnostic criterion for the diagnosis of NF1 in children. This is due to the fact that some of the diagnostic criteria may not manifest until the individual reaches adolescence or adulthood (Filho, Munis et al. 2008).

### **Tumour and Malignancy**

Individuals with NF1 appear to display an increased risk of malignancy. The spectrum can encompass the already mentioned malignant peripheral nerve sheath tumours and optic gliomas. Leukaemia, lymphoma, embryonal tumour, gastrointestinal tumour and breast cancer are other possible examples. Women with NF1 who are younger than 50 have a fivefold risk of breast cancer; they are in the moderate risk category and should be considered for mammography from 40 years of age (Sharif, Moran et al. 2007).

### Diagnostic Criteria for NF1

NIH consensus development conference (1988)

6 or more café-au-lait macules (> 0.5 cm in children, > 1.5 cm in adults)

2 or more cutaneous/subcutaneous neurofibromas or 1 plexiform neurofibromas

Axillary or groin freckling

Optic pathway glioma

2 or more Lisch nodules

Bony dysplasia

1<sup>st</sup> degree relative with NF1

#### **FIGURE 6**

The NIH diagnostic criteria for NF1 are met in an individual who displays two or more of the listed criteria.

### **Life Expectancy**

According to the literature, except for rare cases of complications, individuals with NF1 can live a normal life with 'normal' education and job expectations. The life span of individuals with NF1 is predicted to be 15 years less than observed in the general population (Zoller, Rembeck et al. 1995; Wolkenstein, Zeller et al. 2001; Radtke, Sebold et al. 2007). The vast majority of studies converge on the estimate that 2/3 of individuals with NF1 are mildly affected and do not require major surgery and do not have life-threatening problems (North 1998; Tonsgard 2006; Radtke, Sebold et al. 2007).

Among the major causes of death, malignancy occupies a significant place. Tumour-related neurological complications can also be added, as well as cardiovascular problems. The literature also reports cases of death from accidents, suicide and complications from surgery (Huson and Hughes 1994; Radtke, Sebold et al. 2007).

## **Research**

Current studies on NF1 are listed on the children's tumour foundation webpage

<http://www.ctf.org> (Dilworth, Kraniak et al. 2006; Radtke, Sebold et al. 2007) .

The principal active areas of investigations include: genotype-phenotype correlations, the aetiology of the extensive phenotypic variability, neurodevelopmental issues, treatments for neurofibromas and other tumours (Dilworth, Kraniak et al. 2006; DeLucia, Yohay et al. 2011).

Animal models are also employed for research. Mice and rats are the most commonly employed for the ease of breeding, large litter and short generation span, but also because their NF1 gene shows strong similarity to the human locus (Huson and Hughes 1994). Another studied model is *Drosophila Melanogaster* (the common fruit fly). However, the occurrence of neurofibromas has also been reported in birds, bicour damsel fish and mammals like cows, horses and dogs (Goedegeburre 1975; Canfield 1978; Bossart 1983; Schmale, Udey et al. 1986; Williams, Lucas et al. 2009).

## **Medical Treatment**

At the moment there is no specific medical treatment or gene therapy for NF1. Medical treatment aims principally at palliative and preventive care for symptoms and improving the quality of life of affected individuals. The main modalities of treatment include: radiation therapy, chemotherapy, surgical resection or decompression of enlarging lesions. These interventions are mainly directed towards the treatment of tumours. Surgical removal of neurofibromas is possible, but unsatisfactory, because the tumours tend to grow again. Scoliosis or other skeletal problems may require, alongside surgical treatment, the employment of a special brace (e.g. Boston Brace).

## **Management of Clinical Problems**

Clinical recommendations state that once the diagnosis of NF1 is made, all available first-degree relatives should be screened by genetic service providers to determine whether the case is familial or sporadic and to guide genetic counselling (Ferner 2010). Because of the variability and unpredictability of NF1, monitoring is considered to be essential; NF1 individuals should be encouraged to seek review of any unusual symptom and ask if they are related to NF1.

In the UK adult individuals with NF1 are advised to see their GPs at least once a year to have their blood pressure checked (since they often suffer from hypertension) and for a general health check- up (Ferner, Huson et al. 2007; Schaefer, Bull et al. 2008).

Young adults aged 16-25 are at a vulnerable stage of life and require education about NF1 and its possible complications. Genetic counselling about disease inheritance and psychological supports are advised, particularly because neurofibromas often start to develop in late adolescence (Ferner, Huson et al. 2007).

The recommended protocol for children with NF1 includes annual review to monitor the disease progression by a Community or a Hospital Paediatrician or a specialised GP (Evans 2011). A detailed developmental assessment of the child should be performed before school. In the UK a special educational coordinator might need to be involved at an early stage and an educational statement should be prepared if appropriate (Radtke, Sebold et al. 2007). School performance should be carefully monitored. Many researchers (Huson and Hughes 1994; Ruggieri and Huson 1999; Tonsgard 2006; Radtke, Sebold et al. 2007) emphasize the importance of early reading readiness in preschool children; speech problems should also be resolutely addressed as early as possible with individual speech therapy. Close liaison between teachers, educational

psychologists, occupational therapists and community paediatricians can ensure that the child receives the optimum assessment and remedial support.

It is very important to note that, given the variety of NF1 symptoms, the management of NF1 patients requires a multidisciplinary team. To give an idea, potential disciplines concerned with treating NF1 might include: Neurology, Neurosurgery, Surgery, Oncology, Endocrinology, Dermatology, Orthopaedics, Ophthalmology, Otolaryngology, Gastroenterology, Urology/Nephrology, Cardiology, Obstetrics/Gynaecology, Physical Therapy, Occupational Therapy, Speech Therapy, Early Intervention Programme, Social Work, Psychiatry, Psychology, Support Groups.

### **Genetic Counselling**

Patients and their family members can be referred to a genetic service provider normally by GPs and Paediatricians. As happens with the vast majority of genetic problems, the service focuses on prognosis, genetic risks to future offspring and psychosocial adjustment (North 1998). Looking at the literature on NF1, it is possible to find suggestions and recommendations about genetic counselling which are specific for this syndrome (Evers-Kiebooms, Fryns et al. 1992; Benjamin, Colley et al. 1993; Counterman, Saylor et al. 1995; Cnossen, Goede-Bolder et al. 1998; Barton and North 2002; Barton and North 2004; Belzeaux and Lancon 2006; Bonnemaïson, Roze-Abert et al. 2006; Ferner 2007; Ferner, Huson et al. 2007; Radtke, Sebold et al. 2007). I have summarised their recommendations below:

1. Counsellors should assess and negotiate the pre-understanding and perception that patients have of NF1. Individuals may have received information about NF1 from sources like the internet, media and other physicians. This information might be outdated, inaccurate and partial, i.e., based only on most severe cases (Ablon 2000).
2. Because of the variability and unpredictability of NF1, the list of potential complications is extensive.

Thus NF1 poses serious challenges to counsellors who have to balance between providing patients with enough information on the syndrome and avoiding frightening them (especially mildly affected patients) about the possible severe phenotypic manifestations of the disorder (Ponder, Murton et al. 1998; Evans 2011) .

Information might be overwhelming, so it is suggested that counsellors use their clinical judgement in selecting the appropriate amount of information to discuss during an appointment and possibly spread the disclosure of the information over more than one appointment. Counsellors are also expected to focus on the other side of NF1 variability: i.e., a considerable number of individuals with NF1 lead productive lives, are able to attend school, and are employed and independent (Ferner, Huson et al. 2007; Radtke, Sebold et al. 2007).

3. The word ‘tumour’ is frightening (Sontag 1979). The counsellor should be careful in employing this word and stress that most of the tumours are benign (i.e., non-cancerous).

4. The cosmetic problems caused by NF1 often have an impact on daily life. Research found for example that NF1 symptoms which have cosmetic implications can lower the quality of life of affected individuals (Wolkenstein, Zeller et al. 2001). This finding is however disputable. In other studies, the relation between severity and self-concept was reported to be complex and multifactorial (Ablon 1996). Other variables, for example the socio-economic status of the patient and/or their family network, play a significant role on the impact of NF1. Overall, this 4<sup>th</sup> point highlights the importance for the counsellor to assess the self-perception of the patient.

5. The counsellor is also expected to elicit the narration of experiences of patients in the social environment since NF1 patients tend to have difficulties with social skills.

6. A genetic diagnosis of a complex and potentially severe disease like NF1 can be received negatively by patients and families. It can lower the self-expectations of the



affected individual and the expectations' of others (including their family network).

Counsellors should discuss patients' and their families' concerns about being labelled with a genetic disorder and negative self-fulfilling prophecies (Radtke, Sebold et al. 2007).

7. The counselling sessions should be adjusted to reflect family comprehension level.

This is particularly related to the possibility of cognitive difficulties throughout the whole family (in familial cases of NF1) (Ferner, Huson et al. 2007).

8. Counsellors are expected to assist the family in navigating the complexities of special education and/or other intervention services. Obtaining additional service (e.g. in school) can be very difficult and might prove to be challenging and frustrating (Ferner 2010).

9. Genetic counselling for a couple considering prenatal testing for NF1 can be offered (if they want it). In addition to the counselling sessions, the paediatrician may be called to counsel a family (prenatal visit) when one prospective parent is affected and the foetus has been diagnosed to have NF1 by prenatal testing and to assist the family in the decision making process. The paediatrician may also discuss the options available to the family for management and raising of the child. In case of early prenatal diagnosis, this may include discussion of pregnancy termination, as well as continuation of the pregnancy and raising the child at home or adoption (Radtke, Sebold et al. 2007).

## **2.4 Molecular Genetics of NF1**

NF1 is caused by mutations on 17q11.2 located gene on chromosome 17 and is autosomal dominant. (Cawthon, Weiss et al. 1990; Viskochil, Buchberg et al. 1990; Wallace, Marchuk et al. 1990). The main NF1 gene product is neurofibromin, a protein that is present at low levels ubiquitously in essentially all tissues and works as a tumour suppressor (DeClue, Cohen et al. 1990; Gutman, Wood et al. 1991; Ruggieri and Huson

1999). Neurofibromin belongs to a family of proteins which have a central role in cell biology and are determinant in cancer development (Upadhaya 2010). Neurofibromin serves as regulator of signals for cell proliferation and differentiation and is expressed in particular at considerable levels in the nervous system. The loss of neurofibromin acquired through mutations leads to an increased risk of developing benign and malignant tumours in affected individuals (Korf and Rubenstein 2005).

The gene for NF1 and its product were discovered in 1990. The research was aided by some important contributions. As already mentioned, a decisive role has been played by the establishment of national and international organizations involved in NF1 research like the National Neurofibromatosis Foundation (NNFF, now called Children Tumour Foundation) in the USA, Let's Increase Neurofibromatosis Knowledge (LINK, now called Neuro Foundation) in the UK, and the International Consortium for Neurofibromatosis Linkage Analysis. These organizations provided funds for research and conferences and encouraged pooling of crucial resources such as families and probes for linkage studies.

Another important step was represented by the NIH Consensus Conference (convened at the request of the NNFF) where the diagnostic criteria for the disorder were established (Stumpf, Alksne et al. 1987; NIH 1988). This helped in identifying affected and unaffected individuals in a family with certainty.

Before the application of molecular genetic technologies to the localisation and characterisation of the NF1 gene, clinical care and research were scarcely coordinated (Huson and Hughes 1994). This was often a consequence of the fact that NF1 patients present such a wide variety of different symptoms that no single medical speciality could determine the overall disorder. Moreover, the molecular understanding of the mechanism of NF1 has allowed researchers to complement the clinical approach to the

condition. Researchers started to be able to identify the problems at the DNA level, rather than proceeding from the clinical manifestation of the disorder (Riccardi 1999). Arguably, the NF1 gene has represented a unifying and defining factor and, after its identification in 1990, the clinical care and research landscape changed significantly, coming together to unify disparate treatments to cohere around NF1.

The NF1 gene is highly penetrant (penetrance is virtually complete after childhood)(Boyd, Korf et al. 2009). This means that the traits expressed by the NF1 gene will always or almost always manifest in an individual carrying the gene. However, the term penetrance is only related to whether individuals express the trait(s) or not; it does not refer to the variation in the degree of manifestation/expression of a specific trait. As discussed previously, the expressivity of the NF1 gene is extremely variable.

Approximately 50% of cases represent new mutations (spontaneous mutations) (Preiser and Davenport 1918; Borberg 1951; Crowe, Schull et al. 1956; Riccardi and Lewis 1988). This mutation rate is one of the highest recorded for genetic disorders (Friedman 1999; Ruggieri and Huson 1999; Spits, Rycke et al. 2005). This is felt to be due in part to the large size of the NF1 gene which makes it more susceptible to mutations (Radtke, Sebold et al. 2007). The large size of the NF1 gene has also made mutation analysis difficult (Ruggieri and Huson 1999). So far, over 500 different NF1 mutations have been described without any predominant one (Upadhyaya and Cooper 1998; Upadhyaya, Ruggieri et al. 1998; Riva, Corrado et al. 2000; Kluwe, Tatagiba et al. 2003).

As mentioned above, NF1 is characterized by extreme variability. After the cloning of the NF1 gene in 1990 (Wallace, Marchuk et al. 1990) a consistent part of the scientific

research on NF1 has aimed to understand the complexity and diversity of NF1 mutations (Tonsgard, Yelavarthi et al. 1997; Upadhyaya, Spurlock et al. 2006; Sharif, Upadhyaya et al. 2011). To move towards this aim, numerous studies have looked at correlations between genotype and phenotype (Tonsgard, Yelavarthi et al. 1997; Castle, Baser et al. 2003; Upadhyaya, Spurlock et al. 2006; Upadhyaya, Huson et al. 2007). Hitherto, only two clear genotype–phenotype correlations have been described.

1) A whole NF1 gene deletion is associated with large numbers and early appearance of cutaneous neurofibromas, more frequent and severe cognitive problems, large hands and feet and sometimes somatic overgrowth and dysmorphic facial features (Upadhyaya, Ruggieri et al. 1998; Ruggieri and Huson 1999; Radtke, Sebold et al. 2007).

2) A 3-bp in-frame deletion of exon 17(c.2970-2972 del AAT) is associated with the typical symptoms of café -au-lait spots and freckling, but it is not accompanied by cutaneous or surface plexiform neurofibromas (Upadhyaya, Huson et al. 2007).

The variability of the NF1 phenotype, among affected family members and even in individuals with the same NF1 gene mutation, suggests that numerous factors other than the specific mutation are involved in determining the clinical manifestations. However, the nature of these factors has not yet been determined (Huson and Hughes 1994; Rasmussen and Friedman 2000). The most accredited hypotheses include the effect of modifying genes, environmental factors (including epigenesis) and chance. It is supposed, for example, that since the NF1 gene has a tumour suppressor mode of action that can be expressed in any tissue type, the specific tissue type receiving the second (somatic) mutation and the time during development within which the mutation occurs, may have a role in determining the actual phenotype displayed. These factors are quite variable among different individuals and even within the same individual depending on their different tissues (Huson and Hughes 1994). Such complexity and the diversity of the NF1 mutations make genotype-phenotype correlation difficult.

It is also important to highlight that the NF1 gene is very large and heterogeneous (involved in many activities). So far, researchers have attempted to explain NF1 features focusing only on about 10% of the entire gene which controls the aforementioned tumour suppressor molecule. Recent molecular research on the NF1 gene has changed distinctly from the previous 'genome' approach (characterized by the genetic material itself, the genetic code and its translation) to an 'Interactome' approach, that focuses on the interaction of the gene product (neurofibromin) with other proteins. Researchers are trying to find (and better understand) the multiple and complex pathways in which neurofibromin participates (Riccardi 2008).

Molecular genetic testing of the NF1 gene is available in the UK; on average, molecular results can be generated within a week (Upadhaya 2010). Nevertheless genetic testing is infrequently employed; the diagnosis of NF1 is normally based on clinical findings (Boyd, Korf et al. 2009). Genetic testing can however be used: 1) as a confirmation for individuals who are suspected to have NF1 but do not fulfil the NIH diagnostic criteria; 2) prenatal diagnosis (PND) or 3) preimplantation genetic diagnosis (PGD) (Upadhyaya, Fryer et al. 1992; Spits, Rycke et al. 2005).

PND and PGD require prior identification of the NF1-mutation in the family.

A significant limitation of genetic testing is the lack of genotype-phenotype correlation knowledge. Genetic testing can be useful as a diagnostic confirmation tool, but it cannot predict the severity of the condition (Boyd, Korf et al. 2009). Another problem related to genetic testing is the price. In the UK the cost of the test can range from £700 to £1000 or more (Upadhaya 2010).

Because of the extreme variability and unpredictability of NF1, requests for prenatal testing are not common. Studies on patients' reproductive decisions and intentions found that the highly variable severity of NF1 was a significant barrier to the use of

prenatal genetic testing and, generally, a factor that complicated patients' decisions about having children (Benjamin, Colley et al. 1993; Ponder, Murton et al. 1998). Clinicians and families may hold different views towards prenatal testing for NF1, especially if prospective parents intend to use the test as a means to pursue pregnancy termination, rather than early diagnosis. PGD is seen as a more ethically acceptable alternative to PND, since it circumvents the problematic issue of 'therapeutic abortion' (Verlinsky, Rechitsky et al. 2002; Spits, Rycke et al. 2005).

In the 13th European Neurofibromatosis Meeting (Oct 30<sup>th</sup> to Nov 2<sup>nd</sup>) held in Killarney in 2008, an important discovery was communicated. Some mild symptoms of NF1 (café-au-lait spots, axillary freckling, macrocephaly), but not others (neurofibromas, Lisch nodules) can be caused by mutations on another gene (SPRED1) on chromosome 15. Thus, there is a 'neurofibromatosis like' disorder which has presumably been confused with NF1 in the past (Bennett, Spurlock et al. 2008; Pasmant, Sabbagh et al. 2008). This new disorder has been newly named Legius syndrome. As more genes that produce NF1-like clinical phenotypes are identified, clinicians will need to update diagnostic and clinical management (Upadhaya 2010) in order to establish more accurate diagnosis and genetic counselling. In Cardiff, for example, all the NF1 patients with mild phenotype (n=75) have been re-examined. None of them display an identifiable NF1 mutation and have been screened for SPRED1 mutations (Bennett, Spurlock et al. 2008; Spurlock, Bennett et al. 2009).

## 2.5 Cognitive, Psychological and Qualitative Studies on NF1

### Cognitive Characteristics and Symptoms

The cognitive profile of individuals with NF1 depicted by the literature varies both for the typology of symptoms, their severity and frequency. This may depend not only on the nature of the syndrome, but also in part on the fact that the cognitive profile of NF1 has been studied with different research aims, employing a variety of methodologies, scales, and comparison groups (Mouridsen and Sorensen 1995; Levine, Materek et al. 2006; Boyd, Korf et al. 2009; Jett and Friedman 2010).

The main conclusion reached by almost all of this research is that individuals with NF1 are more likely to display cognitive impairments (Ferner, Hughes et al. 1996; Johnson, Saal et al. 1999; Ozonoff 1999; Barton and North 2002; Maunter, Kluwe et al. 2002; Graf, Landolt et al. 2006).<sup>5</sup> However, a significant level of research has shown that severe generalized intellectual impairment is unusual (Huson and Hughes 1994; North, Joy et al. 1995; Descheemaeker, Ghesquiere et al. 2005; Johnson, Wiggs et al. 2005). Apart from this aspect, a clear consensus on the cognitive profile of NF1 has not yet been reached.

Children with NF1 often display cognitive deficits or learning problems that qualify them for educational assistance (North, Riccardi et al. 1997). The incidences calculated for example are 30% to 65% (Riccardi 1992; North, Joy et al. 1994; North, Joy et al. 1995; Johnson, Saal et al. 1999; Barton and North 2002; Levine, Materek et al. 2006). Some researchers claim that the cognitive problems that individuals with NF1 can display may jeopardise the achievement of their full academic potential regardless of

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<sup>5</sup> According to (Barton and North 2004), 39% of NF1 children present ADHD. Other studies, for example, showed that 50% of the sample presented with learning disabilities, impaired literacy skills, spelling problems, non-verbal learning disability (NLD) and dyslexia, among other effects (Graf, Landolt et al. 2006);(Johnson, Saal et al. 1999); (Maunter, Kluwe et al. 2002); (Ferner, Hughes et al. 1996);(Sebold, Lovell et al. 2004).

their cultural or socioeconomic conditions (Hofman, Harris et al. 1994; Ferner, Hughes et al. 1996; Ferner, Huson et al. 2007).

It is interesting to juxtapose these findings with other studies where individuals with NF1 with academic under-achievement were found to be “neuropsychologically intact” (Brewer, Moore et al. 1997). This seems to suggest that other factors (poor social skills, behavioural problems, clumsiness, bullying, sleep disorders), rather than specific learning and cognitive difficulties might also play a role in jeopardizing the academic achievement of individuals with NF1 (Johnson, Wiggs et al. 2005; Williams, Lucas et al. 2009).

Neuromotor dysfunctions, impairments in psychomotor co-ordination and gross motor function have been reported in approximately one third of the children (Hofman, Harris et al. 1994; Moore, Alter et al. 1994; Moore, Slopis et al. 2000; Descheemaeker, Ghesquiere et al. 2005) alongside with dyscalculia, dyslexia and spelling problems (Eliason 1986; Aron and Halperin 1992; Dilts, Carey et al. 1996; DeWinter, Moore et al. 1999; Thompson, Viskochil et al. 2010).

In particular, from other studies, lower arithmetic and spelling test scores with no or few reading deficits have been found (Hofman, Harris et al. 1994). Other research highlights verbal linguistic problems along with visual and spatial dysfunctions (Hofman, Harris et al. 1994; North, Joy et al. 1994; Marzocco, Turner et al. 1995; Denckla 1996; Marzocco 2001). Conduct disorder, emotional symptoms, hyperactivity, sleep disturbance (in particular sleepwalking and sleep terrors) seem to be higher in individuals with NF1 (Johnson, Wiggs et al. 2005). Seizures, epilepsy and autism are also listed among the possible complications (Huson and Hughes 1994; Vivarelli, Grosso et al. 2003; Levine, Materek et al. 2006).

Some studies on the cognitive profile of NF1 point out the importance of investigating the potential side effects on cognition and behaviour of the drugs that have been



developed for the medication (mainly of the tumoral complication) of NF1 patients (Descheemaeker, Ghesquiere et al. 2005).

In conclusion, the majority of the studies propose intervention in the form of information for parents, systematic screening with standardised tests along with early screening and treatment for speech, motor, and cognitive problems and early screening and treatment for speech, motor and cognitive problems (Johnson, Saal et al. 1999).

### **Psychological Characteristics and Symptoms**

Individuals with NF1 appear to display psychological or psychiatric characteristics of various types, without conforming to a uniform clinical profile (Boyd, Korf et al. 2009).

Depressive syndrome, anxiety state with vegetative dysfunction and organic brain syndrome represent the most common findings (Samuelsson and Samuelsson 1989; Rubenstein and Korf 1990; Counterman, Saylor et al. 1995; Dilts, Carey et al. 1996).

Fear and uncertainty are also common and there are reported instances of psychosis and sociopathic behaviour (Wolkenstein, Zeller et al. 2001; Ferner, Huson et al. 2007). Even an individual with minimal manifestations of the disorder may live with the burden of constant uncertainty (Mouridsen and Sorensen 1995). Personality characteristics are found to be similar for children with inherited NF1 or children with a new (spontaneous) mutation (Prinzie, Descheemaeker et al. 2003).

The vast majority of research almost unequivocally attests that the main psychological problems stem principally from the disfigurement caused by NF1 and from the complex unpredictable nature of the disease. The extreme variability in the phenotypic expression of the NF1 gene makes it particularly difficult for the affected individuals to comprehend the variety of problems associated with NF1 and studies have also shown that patients, in general, tend to know little about the clinical and genetic aspects of the syndrome (Huson and Hughes 1994; Williams, Lucas et al. 2009).

These problems are thought to depend also on the fact that, as generally happens for individuals who are mildly disfigured, NF1 patients may harbour an exceedingly distorted perception of the self, leading to social isolation (Roback, Kirshner et al. 1981; Hett, Mather et al. 1989). Much research in sociology and social psychology attests that greater physical attractiveness seems to be favourable to a person, whereas less physical attractiveness seems to be less favourable. Society tends to be prejudiced against those with atypical facial (or physical) appearance (Dion, Berscheid et al. 1972; Bull 1990). Individuals of greater physical attractiveness are often perceived to be more socially skilled, live better lives, and have more successful marriages and occupations. It is understandable then, that if a person presents disfigurement or cosmetic problems (even mild ones within the range of NF1), they may experience severe social discomfort which can have diverse and strong consequences on the individual self-perception as a whole (Goffman 1968).

Although only some individuals with NF1 display severe disfigurement, plexiform neurofibromas, CALS, short stature macrocephaly and scoliosis are well known symptoms and complications of the syndrome which can also significantly affect physical appearance (Samuelsson and Riccardi 1989; Samuelsson and Samuelsson 1989). Some studies claim that although patients may have physical symptoms that cause them concern, most seem to cope with such symptoms (Ruggieri and Huson 1999; Reiter-Purtill, Schorry et al. 2007; May, Collier et al. 2008; Schaefer, Bull et al. 2008). However, it also emerges that many patients, for example, complained of short stature (Samuelsson and Samuelsson 1989) and find it very upsetting when they see other people staring at their neurofibromas (Bonnemaison, Roze-Abert et al. 2006). Moreover it is important to remember that the number of neurofibromas can increase during puberty, when young people are most concerned about their appearance

(Mouridsen and Sorensen 1995). Patients also revealed that they conceal their lumps (when possible) in order to avoid embarrassment and having to explain the condition to other people. CALS do not usually cause complications; however some patients are distressed over the appearance and may seek skin camouflage (Ferner, Huson et al. 2007). Individuals with NF1 experience restrictions in social contact (Benjamin, Colley et al. 1993; Ablon 1999) and limited job opportunities (Hofman, Harris et al. 1994; Huson and Hughes 1994; Johnson, Saal et al. 1999; Johnson, Wiggs et al. 2005). Other reported factors that can affect social skills are represented by misinterpretation of non-verbal communication such as gestures and facial expression (i.e. social imperception) and clumsiness which may lead people with NF1 to behave in an inappropriate fashion (Eliason 1986; Eliason 1988; Rubenstein and Korf 1990; Counterman, Saylor et al. 1995).

Research with adult individuals indicates that NF1 is associated with significant psychological burden, poor peer relationships and low self-concept (Cutting, Koth et al. 2000). However, individuals with NF1 appeared to be at greater risk of developing negative perceptions about their self during adolescence and adulthood, than during childhood. This is consistent with retrospective interviews conducted with adults who described adolescence as a very difficult period during which they felt frightened about their appearance (Benjamin, Colley et al. 1993; Ablon 1999).

Other studies (Barton and North 2002; Reiter-Purtill, Schorry et al. 2007) report that young children (8-12 years old) with NF1 tended to have a healthy self-concept. These children demonstrated little understanding of the disorder and its progressive nature. A possible explanation for this could be that most of the children who participated in these studies had mild symptoms that were not noticeable to others and perhaps to themselves (Barton and North 2002; Barton and North 2004). Some children did not even know

they had NF1 or at most recorded that they had ‘something’.<sup>6</sup> This seems to suggest there might be a ‘window of opportunity’ at this early stage of development of people with NF1 to intervene to maintain and improve their self-concept. Some scholars are interested in studying different age groups to determine the indicative age at which self-concept tends to decline (Barton and North 2002).

The research of Dilts and colleagues suggests that family dynamics can contribute to the NF1 self-image (Dilts, Carey et al. 1996). The study compared children with NF1 and sex-matched non affected siblings on a broad range of psychological and behavioural dimensions. From the analysis, the conclusion was that the psychological profile of children with NF1 can also depend on the fact that parents may evaluate them in the light of their non- affected siblings and other healthy children in the extended family, rather than to children in general. Physical and psychological differences among affected and non-affected siblings can become part of the everyday context of family life. Drawing on this context, parents may regard and evaluate their children with NF1 as being low functioning. These parental attitudes can in turn be internalised by children with NF1 and can work as inhibitors or facilitators of their development (Wilson and Yahr 1990).

To conclude, the majority of the psychological studies suggest an increase in the level of intervention in childhood to prevent and treat psychological, social and emotional problems (Johnson, Saal et al. 1999; Wolkenstein, Zeller et al. 2001; Johnson, Wiggs et al. 2005). Moreover it is advisable that children who have been identified as having learning difficulties are referred for an evaluation by a paediatric psychiatrist or clinical psychologist. Information for parents plays a crucial role as it facilitates early intervention and prevention. Interviews with families have documented the importance of providing clear and accurate information about NF1 that is revisited over time (Ablon

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<sup>6</sup> It is also supposed that lower IQ might make patients less able to make comparative judgements.

1992; Evers-Kiebooms, Fryns et al. 1992; Riccardi 1992; Benjamin, Colley et al. 1993; Huson and Hughes 1994). Problems such as attention deficit and mood disorders can be treated with psychoactive medication in addition to behaviour management and counselling (Johnson, Saal et al. 1999). Concerns about effects of NF1 on physical appearance may be addressed by counselling or peer support groups.

Overall, it is important to recall the fact that the true incidence of cognitive impairment, learning problems and behavioural psychological disorders associated with NF1 has not yet been clearly determined. Although it is generally accepted that severe mental retardation is not a typical manifestation of NF1, the incidence and typology of cognitive problems varies significantly (Ferner 2007; Ferner 2010). This is also thought to depend on the selection of participants (Huson and Hughes 1994) and what IQ criteria or test have been used (Mouridsen and Sorensen 1995; Levine, Materek et al. 2006). Some researchers criticize the previous studies pointing out that estimations and conclusions are often based on crude and schematic measures such as number of years of schooling, class placement, and personal history or parental and/or teachers reports, rather than standardized intelligence test scores or behavioural scales (Mouridsen and Sorensen 1995; Johnson, Saal et al. 1999; Radtke, Sebold et al. 2007).

### **Qualitative Studies**

Qualitative research allows for an in-depth investigation of the experiences with NF1 from the perspective of the affected individuals and their basic communication networks: families, support organizations and the healthcare system. Therefore, alongside medical cognitive and psychological research, qualitative studies represent a significant contribution to the understanding of the 'NF1 world'.

To my knowledge, the only substantial qualitative work on NF1 has been conducted by the medical anthropologist Joan Ablon during the last two decades. Ablon's research has revolved around the issues of stigma, disability and social justice.

This scholar has published a monograph about the socio-cultural dimensions of the syndrome (Ablon 1999) and a series of papers which deal with specific aspects explored in the monograph (Ablon 1992; Ablon 1995; Ablon 1996; Ablon 2000; Ablon 2002).

The research focused on illness narratives- primarily experience of stigma- and social support around NF1. It examined the impact of disability and stigma on the life of affected individuals (Ablon 1992; Ablon 1996) focusing in particular on the negative effect of the misdiagnosis of 'the Elephant Man disease' as NF1 (Ablon 1995; Ablon 1999; Ablon 2002). It also investigated the role and functions of NF support groups on the lives of affected individuals (Ablon 1999).

The methodology was based on in- depth interviews (semi-directed open ended qualitative interviews) with adults with NF1 and unaffected parents with affected children. She also conducted follow- up interviews with some of these participants for a total of 54 interviews. The sample was recruited from the NF support group in North California, where the study was set, and the database of genetic departments of two major metropolitan hospitals in the area. Participants were mainly interviewed in their homes or, in some cases, in restaurants and came from diverse economic, social and geographic backgrounds. The research took place around the 1990s.

## Uncertainty

In accordance with the psychological research, Ablon found that the complex symptomatology and unpredictability of NF1 represented the most distressing psychological features of the condition for patients and families. Participants in her study appeared to be worried about future possible complications of the disorder.

According to Ablon, this anxiety was also fomented by the absence of a role model for NF1 ('Elephant Man' aside). She suggested that, differently from other conditions that she studied - e.g. achondroplasia - which have reasonably constant parameters, NF1 because of its extreme unpredictability and variability, is a *condition without parameters* (Ablon 1999; Ablon 2000). Hence, individuals confronted with NF1 do not have a model or candidacy (apart from the extreme cases portrayed by the media) to draw upon in order to understand and manage their disorder.

Ablon focused also on parents' experiences in receiving their child's diagnosis of NF1 (Ablon 2000). The parents interviewed recounted shock, fear, guilt and depression after the diagnosis of their children (see also (Dylis 2006)). Once again, the characteristics of NF1 which were particularly frightening for parents of children diagnosed with the syndrome were the heterogeneity of its presentation, the general unpredictability of its progression as well as the inability of clinicians to predict the appearance or progression of the symptoms of this syndrome. The void resulting from the uncertainty around NF1 (for the lack of parameters) generated a climate of high anxiety in parents (Ablon 1992; Ablon 2000).

The uncertainty of NF1 represents one important interest of this Ph.D. thesis. However, I am extending Ablon's work by exploring how patients and families relate to the uncertainty of NF1 in the light of genetic knowledge, which is something that has not yet been explored in great detail.

## **Family Dynamics**

Ablon also looked at family dynamics in relation to NF1. She highlighted that the majority of the interviewees who had parents with NF1 did not seem to have found their parents to be helpful role models for living with the condition. Participants reported a significant or even total lack of communication about the syndrome with their parents (Ablon 1999).

Ablon suggested that parents with NF1, albeit potentially more aware of the difficulties their affected children may experience or develop, did not necessarily display more efficacy in managing the condition of their children. In the first place, as already pointed out, the syndrome can be very variable within the same family; therefore family members can experience a very different symptomatology. Dealing with the unknown in a child with NF1 is also a very challenging task for a parent. Furthermore, parents could also experience a sense of guilt for having transmitted their condition to their children. Finally, Ablon observes that parents with NF1 frequently present cognitive or physical difficulties. This could prevent them from being ready to promptly and effectively tackle the difficulties their diagnosed children experienced. Therefore, Ablon argues that because of their own physical or cognitive complications, parents with NF1 may find it particularly arduous to effectively relate to healthcare professionals, teachers and other providers to negotiate services for their affected children.

Nevertheless, according to Ablon, at the time of her research (1990s in the US) this situation was already improving as a result of more general awareness among participants of the second generation about genetics and NF1 (Ablon 1999). Affected parents of second generation appeared to be more knowledgeable about their condition than their parents' generation as well as more willing to talk about the condition with their affected children. For Ablon, having more information about the syndrome- especially on its genetic basis- reduced feelings of guilt, fear, stigma and religious



attribution that may have prevented former parents from openly discussing with their children their shared disorder.

Ablon strongly emphasises that a supportive familial environment in which NF1 is openly discussed is extremely important for individuals with NF1 especially in early life.

Ablon could not identify any clear correlation between clinical severity, and the way affected individuals coped with their syndrome (Ablon 1999: 15). However, she found that the most important factor in mediating individual adaptation and coping strategies was the family environment. She claimed that a strong and united family is likely to give more confidence and security to individuals, encouraging a better adjustment to the syndrome and increasing in general their quality of life. In particular, she also pointed out that an open and good communication about NF1 in the family is important and can help to bolster self-esteem and confidence in affected children (Ablon 1996; Ablon 1999). It is however also possible to argue that other families may prefer to adjust to the syndrome by constructing different identities not founded on NF1. This Ph.D. aims to bring the study of the familial experience with NF1 to the fore. Whilst Ablon's participants were either NF1 individuals or parents of affected individuals, I am exploring family networks as much as possible interviewing many family members within the same network. Furthermore, I combine this family sampling strategy with a key interest in the links between individual and familial experience and sense making practices around genetic knowledge.

### **Impact of the syndrome**

Ablon described NF1 as having a considerable impact on individuals' self-confidence, educational careers, earning ability, economic status and reproductive decisions (Ablon

1999). Participants with NF1 tended to have negative perceptions of their selves. Many participants in her study recalled that as children they were teased at school about their physical attractiveness, appearance, their academic performance and felt lonely. The presence of visible tumours (neurofibromas) was strongly embarrassing and made them feel different and isolated (Ablon 1999). Moreover, some individuals who were mildly affected reported to be very anxious about the stigma associated with NF1 and /or the possibility of developing tumours or other complications. However, there were also seriously affected persons with skeletal abnormalities and disfiguring tumours who deployed a positive, normalising attitude towards their condition and showed high self-esteem.

This diversity of NF1 experiences reported by Ablon is consistent to a key principle of the disability rights perspective (Hughes and Paterson 1997; Bickenbach, Chatterji et al. 1999; Kerr and Shakespeare 2002) that is, individuals cannot be defined and do not necessarily define themselves by their disease or impairment only. The experience and impact of a disease is mediated by many contextual factors like access to medical, financial and educational resources, individual personality, family acceptance and broader sociocultural instances (see also Chapter 6).

Ablon found for example that the impact of NF1 also depended on the socio-economic status of the affected person and/or their family network (Ablon 1996). For instance, many individuals raised in middle class lifestyles were able to keep only low level or part-time jobs with few or no benefits and tended to live on the economic borders of society. Often, this was due to the effects of learning, cognitive and behavioural difficulties as well as the necessity for frequent absences from work for surgery or for medical appointments in general.

In a recent study based on semi-structured interviews of young individuals with NF1 recruited through a clinical genetics database, participants perceived themselves to cope well with their syndrome, albeit they also claimed to feel isolated and expressed concerns about their future (May, Collier et al. 2008). The researchers also suggest that female participants appeared to be more influenced by cosmetic complications related to NF1 (as concerns employment and their life in general).

In contrast, Ablon argued that the syndrome had a more significant impact on the men rather than the women interviewed (Ablon 1996; Ablon 1999). She found that women in her sample had higher rates of marriage and economic achievements, than men in comparable conditions. According to Ablon, men tended *prima facie* to deploy a more pragmatic, stoic and positive public attitude towards their condition than women did. But, in reality, they were more affected in their manhood and personhood by NF1. As result of this, they tended to isolate themselves from society, missing many life-stage opportunities to realize their potential. Therefore, for Ablon the reason why women with NF1 may appear to be more affected and concerned relied on the fact that men are socially expected to be more stoic and reluctant in asking for help than women (Ablon 1996). This Ph.D. thesis method samples and looks at both genders within a family context.

### **Reproductive decisions**

In the Ablon study, many individuals with the condition decided not to have children. This was dependent, according to Ablon, on a series of explicit and implicit circumstances (Ablon 1999). Firstly, this decision could depend on the fact that pregnancy can permanently increase the visibility and sometimes severity of women's symptoms.

More commonly, both men and women decided not to have children on the basis of the heritability and potential severity of the condition; i.e. the child could be seriously affected and have a shorter life. They also draw on their personal negative experience with the condition to justify their reproductive decisions. Following from this, Ablon claimed that individuals' reproductive decisions were also considerably influenced by an internalised low self-concept and feelings of devalued status. Thus, for Ablon alongside the possibility of giving birth to a seriously affected child the stigma and humiliations experienced by NF1 individuals in their lifecourse could underpin their reproductive decisions (Ablon 1999; Ablon 2000).

Similar results have been reported in two qualitative studies which explored the reproductive decisions of adult NF1 patients in the UK (Benjamin, Colley et al. 1993; Ponder, Murton et al. 1998). A particularly interesting point is made by Ponder, Murton et al (1998): family dynamics can influence decisions about reproduction. For example, NF1 parents who often felt guilty for having passed the condition to their children, presented contrasting feelings towards the possibility of having grandchildren. This may put pressure on individuals with NF1 who are considering having children and, overall, causing or exacerbating family conflict. Having this in mind, the authors suggested that more support should be made available to families as a whole to help communication and management of the condition (Ponder, Murton et al. 1998).

### **Stigma**

The central focus of interest of Ablon's research on NF1 is individuals' adaptation to stigma. This issue is investigated in the monograph (Ablon 1999), and is also explored in the academic journal publications (see in particular Ablon 1995; 1996; 2002) where

Ablon draws also on data from her research on other conditions (like achondroplasia and ontogenesis imperfecta).

Probably this interest in stigma stems also from the historical period in which Ablon was conducting her investigation (mainly the last decade of the 20<sup>th</sup> century). This period was relatively close to the re-diagnosis of the Elephant Man as having Proteus Syndrome and not NF1, which occurred at the end of the 1980s (Cohen 1988). Ablon has devoted considerable attention to exploring the effect of the association between NF1 and what has been called the ‘Elephant Man disease’ on individuals with NF1. She found that this association had a considerable negative impact, increasing the stigmatization of this genetic condition.

It is difficult to tell whether this correlation between NF1 and the ‘Elephant Man’ is still pervasive nowadays, as media coverage appears less focused on this in the UK.

However, a recent ethnographic study conducted in Canada reports that individuals with NF1 and their parents can still be told by doctors they are affected by the ‘Elephant Man disease’ (Legendre, Charpentier-Cote et al. 2011).

In the conclusion of her monograph, Ablon (1999) also made a few points that are particularly relevant for this research. She seemed to suggest that participants’ feelings of stigma coloured their relation to the genetic nature of their condition. According to Ablon, genetics was regarded as something deeply entrenched in the body and the essence of individuals. Therefore, participants with NF1 tended to see their disease as something inherent in their essence, something permanent that would stay with them for their whole life. There were feelings of shame related to the idea of having a genetic disorder, that is, of bearing some permanent defect within the body. Furthermore, participants experienced feelings of guilt and blame for passing the condition on and/ or saw their condition as constraining their reproductive choices.

Nonetheless, these points are not developed further as the role of genetic aspects in individual and familial experiences with NF1 was not the focus of Ablon's work. This thesis follows in the wake of Ablon's incipient theories about genetics by providing an in-depth empirical investigation of the genetic and familial understanding of NF1.

The phenomenon of stigmatization is also tackled in the work of Rozario, based on the life-narrative of a UK-Bangladeshi woman affected by the syndrome (Gaff and Clarke 2007; Rozario 2007). This paper shows how stigma can emerge within the family network, and not only from society. Paradoxically, in this woman's narrative, the social environment appeared to be more open than the familial one. For the family of origin of this woman, who had a Bangladeshi cultural background, appearance, gender, sexual identity and marriage appeared to be central concerns. The woman interviewed recounted that because of her dark skin and visible tumours her parents and relatives regarded her as 'unmarriageable'. Furthermore, despite her intelligence and competence, this woman was not accepted by her family of origin and wider UK Bangladeshi acquaintances. In particular, in her narrative, emphasis was put also on how she was considered to be morally corrupted. Thus, a different physical appearance seems potentially to be a valid reason to undermine the whole status and value of a person to whom automatically fewer rights are recognized.

### **Support Groups**

Ablon looked also at the function of NF1 support groups on individual experiences with the condition (Ablon 1999). The NNF had a branch in North California, where Ablon's study took place. The foundation sponsored local support groups organising many scientific events for researchers as well as activities more tailored to patients and families, like discussion meetings and summer camps for teenagers with NF1.

Many participants interviewed either did not join any group or joined to receive the newsletters, but preferred not to attend the meetings. One rationale they gave for not attending meetings was that they were afraid of encountering people who were more badly affected, for instance, with multiple tumours or severe skeletal deformities. In accordance with what has already been stated concerning gender response to NF1, Ablon observed that the majority of men justified their choice of not attending by stating they did not need any support, thus conveying a public image of independent and pragmatic masculinity (Ablon 1996).

Those who attended at least one meeting evaluated the experience in diverse ways. Some participants confirmed being relieved to see other individuals who were more seriously affected than themselves, whereas the same experience appeared to terrify others.

Sharing experiences and information about the everyday management of the condition did not necessarily provide more confidence. On the contrary, for many participants it amplified their anxiety as it was seen as a vivid showcase for the possible NF1 symptoms and complications they may develop. Paradoxically, although designed to provide assistance to patients and families and to relieve them from the distress of living with NF1, support groups could also have the opposite effect (Ablon 1999).

The distance of venues for the meetings also surfaced as a hindrance to their attendance. However, according to Ablon, the main obstacle to individuals' engagement with genetic support groups for NF1 was not structural, but depended on the extreme variability of the condition. In her interpretation, the diversity and multiplicity of the manifestations of NF1 can challenge the creation of a sense of community based on the same interests and worries that normally underlies the formation of genetic support groups. Individuals with NF1 because of the variability of manifestation of the condition may feel to have nothing in common with each other (Ablon 1999).

My research intends to develop Ablon's points further. First it is situated in a different temporal, geographical and cultural context, that is, the 21<sup>st</sup> century and the UK.

Furthermore, it looks at support groups in relation to genetic knowledge. I am interested in how genetic knowledge may influence individuals' illness identity and their use of genetic support groups. In other words, I empirically investigate whether there is 'biosociality' for NF1, and - if so - how it is structured.

### **Healthcare System**

Concerning medical practice, Ablon highlighted that participants in the study appeared to be generally dissatisfied with the healthcare service received (Ablon 1992; Ablon 1999; Ablon 2002). Furthermore, there appeared to be more medical services and attention available for children with NF1 than adults. Paediatric patients tended to be monitored and managed better, whilst it was easier for adult patients to be forgotten by the medical system.

Overall, Ablon reported a frequent lack of continuity of care both for adult and paediatric patients. Despite the importance of regular monitoring for NF1 patients, the participants interviewed perceived many difficulties in managing to see specialists. This was caused by problems in juggling the timetables of different specialists, long waiting periods, patients' feelings of hopelessness towards doctors due to previous negative experiences, as well as patients' cognitive and learning difficulties. For these reasons, Ablon suggested that more attention towards psychological and cognitive complications related to NF1 would also be beneficial for patients. She also highlighted the need for an health care service which is more holistic, focused on the patient lifecycle and which can offer a more effective system of continuity of care (Ablon 1999).

From the accounts of participants, doctors appeared to be paternalistic in style in terms of the communication of diagnosis and information about the syndrome. For instance,



they tended to try not to alarm patients in the process of disclosure (e.g. they reveal only some aspects of NF1) which is in accordance with the 2<sup>nd</sup> point of the Genetic Counselling paragraph in this chapter (§ 2.3). In Ablon's work it also emerged that physicians were often the bearer of the NF1 diagnosis and they appeared to be unready and lacking in communicative skills, which increased the anxiety in parents (Ablon 2000). In addition, during the process of diagnosis sometimes parents referred to the case of the 'Elephant Man'; occasionally some physicians did the same. The greatest trauma for parents occurred when the diagnosis of NF1 was accompanied with the labelling of the 'Elephant Man disease' (Ablon 1995; Ablon 2000).

According to Ablon, clinicians should give to patients a realistic context for expectation. She suggested that guiding patients and families in seeking information on the syndrome would be crucial, as the available information on NF1 often revolves around extreme cases which are likely to cause anxiety. She also pointed out that more attention should be devoted towards the setting and style of disclosure. Patients and families may be shocked at the initial time of disclosure and may not be able to formulate any questions. Therefore, Ablon suggested that healthcare professionals should devote more time to listening to patients and should offer more follow-up appointments. Ablon observed that patients referred to a geneticist or a genetic counsellor found these specialists to be the most helpful. Moreover, she also stressed that support groups can provide a crucial service that should be complementary and coordinated with the medical one (Ablon 1992; Ablon 1999; Ablon 2000).

Patients diagnosed with NF1 or other genetic disorders in the UK have been reported to find difficulties in knowing about and gaining access to genetic services (Ponder, Murton et al. 1998; Evans 2011). Because of the late onset of NF1, diagnoses are often delayed even when there is a family history of the condition. Mildly affected NF1

patients do not always receive counselling. Seriously affected patients are normally seen by specialists who have expertise in treating the specific symptom, but do not deal with the syndrome as a whole, its heritable nature and other aspects of the syndrome that can be relevant to patients (Ponder, Murton et al. 1998; Evans 2011).

In another study based on interviews conducted in the UK (Benjamin et al 1993), patients' levels of knowledge of main features of NF1 were generally good, but as concerns complications, they were more influenced by individual and familial experience, rather than biomedical knowledge. Those who received counselling were overall more informed.

An interesting example of negotiation between familial and medical knowledge was the fact that some parents with a child who was found to be a *de novo* mutation believed to be at high risk of having another affected child, even after having been examined and counselled. Furthermore, affected parents of children with NF1 were overall more informed and demonstrated greater knowledge of the condition than patients without a child (see the discussion of concept of genetic responsibility in the next chapters).

Keeping these important considerations about patients, families and the healthcare system in mind, this Ph.D. thesis will further explore the situation in the UK, with its National Health Service infrastructure, at a different point in time. Furthermore, it will look at the role of genetic knowledge and practices in the healthcare management of individuals and families with NF1.

## 2.6 Concluding Remarks

In this chapter I have looked at the medical, psychosocial and qualitative research on NF1. A key point that can be drawn from the historical, medical and molecular genetic sections is that NF1 has become a genetic syndrome, even while highly variable. The investigation of this point and its relation with the role of monstrosity images, the stigma of having a genetic syndrome and the healthcare management of this disorder can provide interesting insights to our understanding of social construction of genetic disorders.

From the review of the qualitative and psychosocial research, it emerges that some important issues strictly linked with the syndrome have not been sufficiently investigated. Although NF1 has become a genetic syndrome, there is no research that extensively explores the way genetic knowledge influences individual and familial experiences with the condition. Questions concerning how genetic knowledge can be employed to define self, in interpersonal management and in structural support such as support groups have not yet been explored in great details.

Ablon has touched on this issue, but despite the title of her monograph - 'Living with genetic disorder. The impact of Neurofibromatosis 1' (Ablon 1999) - her research remained focused on stigma and psychological adjustment and did not directly explore how the genetic aspects of the condition were experienced.

A great deal of emphasis has so far been placed on the issues of stigma, psychology of appearance, self-perception etc. The problem of uncertainty, dependent on the variability and unpredictability of NF1, has been explored, but not in the light of genetic knowledge and practices, probably because of the historical period in which this research was undertaken. Also the issue of stigma itself has not been significantly correlated with genetic information and the consequences of being 'labelled' with a

genetic syndrome (Beck 1992; Featherstone, Atkinson et al. 2006) have not sufficiently studied in relation to NF1.<sup>7</sup> Furthermore, Ablon has investigated the role played by NF1 support organizations for doctors, patients, and parents. But, once again, the links between the genetic aspects of the syndrome and support groups have not been explored.

It has also to be noted that overall the cognitive, psychological as well as the qualitative studies are limited in their generalizability because they are based on a limited sample, which is often collected from hospital clinics and family support groups where it is considered more likely to find individuals who have a more serious affliction. Given that the symptoms of NF1 are highly variable, the number of less seriously affected people has not been taken into account. Thus, it is important to observe that in the reporting of frequency figures, the actual occurrence of more serious manifestations may be overstated because of a failure to ascertain mildly affected cases in which a medical opinion had not been sought. Ablon sampled from metropolitan hospitals and support groups, while participants to this research project have also been recruited from outside NF1 associations and clinical settings.

This research draws on Ablon's work, introducing important points of novelty, like the focus on genetics, a deep exploration of family networks, a different sampling strategy and geographical and cultural context.

Ablon provides very rich and interesting qualitative data (probably the most comprehensive available so far) on individual experiences of NF1. Her research is very informative and shows the utility of qualitative methods as a means to gain an insight into the experience of living with this syndrome.

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<sup>7</sup> This is the same point made in the paragraph Genetic Counselling (§ 2.3).

Nevertheless, although Ablon organises the exposition of participants' narratives around clear categories, such as psychosocial issues, diagnosis, gender differences and socioeconomic status, she did not seem to employ a formal approach to derive common themes from her data. Her monograph is prevalently descriptive; the findings are not strongly framed around and tied to an in-depth theoretical discussion of disability, genetics and other relevant sociological and anthropological concepts.

This choice could be seen as determined by a noble intent. Given the limited (almost inexistent) information on the psychosocial aspects of the syndrome, especially before her research, Ablon's principal aim was to provide healthcare professionals, social service workers as well as affected individuals and their family with information on the experiences and concerns of people affected with NF1. She clearly refers to this "mission" in the preface to her monograph (Ablon 1999: ix).

In-depth cultural analyses of genetics, disability and theoretical discussions of other relevant sociological and anthropological issues may have been 'sacrificed' to the aim of giving more space to the voices of patients and make their needs and experiences easily available and readable to families, healthcare providers and other researchers interested in NF1 (Landsman 2000; Shuttleworth and Kasnitz 2004). Ablon's choice to let patients and families voices speak for themselves could be seen as a 'weakness' from a theoretical point of view, but is also a strength in terms of the richness of data on the experiences and coping patterns of individuals and families with NF1.

I deeply share Ablon's intent to "reveal the human situation of her informants" (Shuttleworth et Kasnitz 2004:146) and to help individuals and families with NF1 as well as clinicians who work with them. However, whilst giving space to empirical qualitative data on NF1, I will also try to critically reflect on relevant literatures that explore patients and families experiences with and health care professionals'

management of genetic disorders. I will draw on this body of literature to set the research aims and to discuss the findings.

Many studies of the social impact of genetic medicine have focussed on the experiences of late onset disorders with known and predictable phenotypic expression, like Huntington's disease and breast cancer. Because of its high variability and uncertainty - that is its being *without parameters* - NF1 provides a novel and challenging case to explore the psychosocial impact of genetic disorders.

In the next chapter, I outline the theoretical framework I employ for filling these gaps in the literature and I formulate the research questions.

### **3. Theoretical Framework**

#### **3.1 Introduction**

In the last four decades, a growing number of diseases and conditions have been found to have a genetic component. Consequently, more and more individuals can be identified as being genetically ‘at risk’, as having carrier status or a genetic disorder. This gradual expansion of genetics in medicine has become the subject of interest for an increasing number of social scientists who have explored the potential and actual impact of genetics on individuals’ subjecthood and on society.

The study of the construction of genetic disorders, perceptions of risk, heritability, and attitudes towards genetic technologies has important social implications. It explores whether and how genetic diagnoses affect individuals’ subjecthood, their interpersonal relationships and the ways they plan and conduct their lives. Furthermore, gaining insight into how individuals experience genetic conditions also has significant clinical applications. It can highlight aspects of patients’ experience which may not always be visible in clinical settings, ultimately contributing to a more patient-oriented service.

Genetic information also has important familial implications. If an individual is diagnosed with a genetic disorder, for example, it is possible that their siblings or offspring may also have inherited the same condition. Exploring the experience of genetics and family patterns of disease within families can produce a more nuanced understanding of the psychosocial impact of medical genetics. With regards to the healthcare service, it can offer a greater insight into how family dynamics and processes can influence the way patients face genetic conditions, genetic futures and healthcare.

As illustrated in Chapter 2, the psychosocial, qualitative and anthropological works on NF1 which have been conducted so far have predominantly explored stigma and social-psychological adjustment, but they did not extensively focus on the genetic aspects of the syndrome. Questions concerning how genetic knowledge influences individual experiences with NF1, how it can be used to define the self, in interpersonal management and in structural support such as support groups or websites have received little or no attention. This may well reflect the fact that genetic explanations have historically become more prominent only in the last decade. One of the main aims of my thesis is to fill this gap by examining how the genetic aspects of NF1 influence the lived experience of the syndrome.

There is also a lack of research that directly explores individual and familial experience with NF1, focusing on how individuals and families make sense of genetic knowledge and manage this variable and unpredictable syndrome. Ablon's research provides valuable insights on family dynamics around the syndrome, but mainly on disability, stigma and uncertainty, rather than genetic knowledge. Furthermore, Ablon did not extensively explore family networks. She interviewed only adults with NF1 and unaffected parents of affected children. As I will further discuss in the methodology chapter, in this Ph.D. thesis I interviewed as many family members as possible within the same network. I believe that this family network approach can further tap the psychosocial dimension of NF1, giving access to the views of different family members on the disorder, shedding also more light on the interrelation of individual and familial experience with the syndrome. Collecting interviews from family networks is also particularly significant, since families are important sites for the investigation of a given experience with genetic conditions; as already stated, genetic diagnoses occur at a familial level and have implications for other family members. Furthermore, Ablon



recruited from clinical database and support groups; she herself acknowledged the impossibility of establishing whether this sampling is representative, and, considering the high variability of the disorder, she also suspected it could be composed of individuals who are more severely affected (Ablon 1999; Ablon 2000). Being aware of these limitations, in order to secure a more representative sample, I did not directly recruit from NF1 support groups, but I sampled both within and outside (through snow ball sampling) the NHS clinical genetic database of the a hospital in the South of England.

As stated in the previous chapter, my research develops Ablon's work, further with a focus on genetics, a deep exploration of family networks, a different sampling strategy and geographical cultural context. With this study, I also intend to contribute to the qualitative research on NF1 and to sociological studies on the construction of genetic disorders. I am interested in relating the case of NF1 to the relevant literature in the sociology and anthropology of genetic medicine, to explore the possible modalities in which genetic knowledge and technologies are changing individual and familial construction of diseases such as NF1, their health related practices and their social relations and values.

This chapter presents and critically evaluates the theoretical constructs related to this thesis's research aims. It will explore studies on the psychosocial implications of genetic knowledge and technologies for individuals and their families and the role played by the healthcare system in individual and familial experiences and understandings of genetic conditions. It will also provide a rationale for my research and set out the research question of this thesis.

## **3.2 The Genetic Self**

### **Genetic Medicine and Subjecthood**

Since the last decade of the 20<sup>th</sup> century, the Human Genome Project and the burgeoning of genetic knowledge and technologies have been accompanied by a growing widespread attention to and interest in the genetic and inheritable aspects of human traits. Individuals' genetic profiles have come to be regarded as a determining factor in disease risk. Consequently, many diseases are being redefined in genetic terms, and genetic practices are entering the medical domain.

Sociological and anthropological studies of genetic medicine have explored whether this implementation of genetic practices in healthcare has profoundly changed how individuals identify themselves and act in relation to their diseases. This strand of research stems from a long tradition of sociological and anthropological studies on the social impact of public health and the process of medicalization, and then geneticization by which non-medical problems came to be increasingly redefined and treated as medical and subsequently as genetic (e.g. alcoholism, hyperactivity etc.) (Zola 1972; Zola 1975; Illich 1990; Lippman 1991; Hoedemaekers 2001; ten Have 2006).

The work of Petersen and Bunton (2002) provides an excellent point of access to this research, allowing an exploration of important broad issues related to genetic medicine, healthcare systems and patients' experiences of disease. The authors point out that genetic technologies are pervasively portrayed by nation states, professional literature, media and other social organs as capable of delivering significant treatment and prevention innovations. Compared to other typologies of medical information, genetic information is considered to be novel as it is predictive, it can be relevant to other people who are genetically related (i.e. it is relational) and it is perceived as something

that is tied into our biological essence. Following from this characterisation, genetic information tends to be widely depicted as having an empowering effect. The possibility of detecting genetic risks, susceptibilities and disorders is generally presented as beneficial, as it provides individuals with more and useful information about their body, enabling them to actively pursue health management strategies and engage in lifestyle modifications to improve their wellbeing and /or to minimize risk and disease. Genetic information is thus conceived as capable of promoting patients' autonomy and choice.

This 'genetic view' has fostered and justified the development in recent years of a significant number of public health initiatives to increase populations' level of understanding of genetics, often termed genetic literacy (see for example the initiatives of the WHO, NowGen and their programmes for increasing citizens - especially young individuals' - education on genetics, or more generally the so called 'participatory technology assessment' or 'upstream public engagement' (Harris and Rose 2010)). Thus, according to Petersen and Bunton, 'gene talk' has become pervasive in our society; individuals seem to be directly and indirectly encouraged to become more aware of the role played by genetics on their health. Discourses about genetic risk, predispositions and genetic explanations for traits and even behaviours have become increasingly common.

Moreover, genetic literacy does not solely bear the potential for altering - at the individual level - one's construction of the body and self. As already pointed out, unlike other forms of risk, genetic risk is inevitably distributed among family networks, having potentially great repercussions on familial relationships. Affected individuals, for example, can face significant challenges concerning their reproductive choices as well

as disclosure and management of genetic risk with kin and extended family (Petersen and Bunton 2002).

Because of its intergenerational implications, genetic information entails knowledge about one's family history oriented towards the disclosure of the inheritance of disorders, risk and predispositions. Family medical history and genetic maps are used to effectively chart and treat the patient. Thus, drawing on works of scholars who, like Finkler (2000), have a research interest in genetics and kinship, Petersen and Bunton point out that with the advent of genetic medicine, the family represents a primary site for risk detection and preventive intervention. It can be argued that it is the family, rather than just the individual, that becomes the real patient (Finkler 2000; Petersen and Bunton 2002; Godard, Kaariainen et al. 2003; Bunton and Petersen 2005). As family dynamics (not just individual experience) are a key focus of this thesis, I will explore this in further detail in section 3.3.

Petersen and Bunton also point out that not only patients, but also healthcare workers have been encouraged to improve their genetic literacy. As more and more diseases have been found to have a genetic basis, genetic practices have come to be regarded as essential diagnostic, management and prevention tools. Consequently, healthcare systems are expected to undergo processes of reorganization in order to integrate clinical genetics into specialised and primary healthcare practice. European countries, for example, are developing common frameworks of core competences for clinical geneticists as well as healthcare professionals in the primary and secondary sector to implement the overall education in genetic medicine of healthcare providers (Skirton, Lewis et al. 2010). There is also a growing demand in medical systems for collaboration between geneticists and healthcare specialists in primary and secondary care and to

mainstream genetics in medicine. Primary healthcare providers (for example GP and general paediatricians) are expected to play a crucial role as gatekeepers for patients' and families access to genetic services. Since they are generally supposed to have established a long rapport with patients and they often deal with families, they are thought to be in the position to detect the possibility for genetic risk and refer patients and families to the proper specialists.

I would argue that this conception of continuity of care is based on an idealistic and old fashioned view of GPs and of family. In current practice, for example, it is very common for patients to see different GPs in the same health centre. Moreover, the idea that family members see the same GP as the 'family' doctor is a rather outdated.

Nonetheless, this view of GPs appears to be pervasive. It is also reflected for example in the importance given to GPs very recently in the last healthcare reform of the UK Department of Health (DoH, Health and Social Care Bill 2010).

Mirroring a prevailing trend within sociology of genetics and other different scholarly discussions of genetic medicine (see for example (Lippman 1992; Hoedemaekers and Have 1997; Hoedemaekers 2001; ten Have 2001; Rose 2006; ten Have 2006; Arnason and Hjorleifsson 2007), Petersen and Bunton assume that governmental, healthcare professionals and individuals' understanding of health and illness are increasingly influenced by genetic practices (Petersen and Bunton 2002). However, in contrast to this trend, Petersen and Bunton openly recognise that the collaborations between clinical genetics and other specialties encounter some barriers. These include the inadequacy of the training in genetics of GPs in the primary and secondary sector and the difficulties in translating research outcomes in genetic medicine into clinical practice.

According to Petersen and Bunton, there is another important element associated with genetic information that governmental and other forms of public health discourses convey in a more subtle, although still compelling, way. The efforts to promote genetic literacy are driven by an “unstated assumption” (Petersen and Bunton 2002) that patients and individuals do not only have the right to genetic information about their health, but they also have a duty to become informed about their genetic constitution and to undertake risk minimization strategies to monitor and modulate their own risks (Petersen and Lupton 1997; Weiner 2010). In my opinion, behind this claim there is probably also the idea that *because* these genetic technologies are made available and offered (at least in theory), individuals may feel pressurised to engage with them and make informed choices as part of their duties as responsible citizens. Thus, for Petersen and Bunton, the notion of genetic risk is not morally neutral, as it may seem at first glance, but it is charged with governmental discourses of active citizenship. Genetic risk is also seen as a tool or justification used by de-centralised, post-welfare states for reducing structural interventions to increase people’s quality of life and rely instead on the agency and responsibility of individuals and families to intervene on and regulate their own risk (Lippman 1998; Rapp 1999; Petersen and Bunton 2002; Lock and Nguyen 2010).

This position on genetic technologies can be seen as a continuation of a more general argument made by Petersen and Lupton about wider individualising trends in public health towards the encouragement of individual responsibility to manage risk and illness and optimise health (Petersen and Lupton 1996; Petersen and Lupton 1997). In recent neo-liberal forms of government, states have reduced their welfare intervention and their responsibility for the provision of social services. At the same time, states have become interested in genetic explanations of many health and social problems that were

previously addressed with the adoption of structural changes and intervention (e.g. the reduction of the level of pollution in cities, better provision of housing and better working conditions). These moves, merged with the strong promotion of an individualistic entrepreneurial culture that characterises many 'advanced' neoliberal societies, has fostered the model of the active and autonomous citizen who is expected to become genetically literate to make informed choices and take a greater responsibility for managing and minimizing their own risk.

To summarize, Petersen and Bunton's work is critical of the idea that genetic practices increase personal choice and are empowering. They contextualise and problematize this idea in the light of the trends in public health thinking and wider political agendas.

Whilst genetic information could be empowering in certain cases, it is also understood as potentially putting pressure on individuals to undertake actions to minimise risk, transforming problems that were previously seen as public concerns into private individual and familial matters. Following from this, Petersen and Bunton also suggest that the increasing interest in public health on genetic aspects of disease may deflect attention from environmental and structural interventions which could be more urgently needed to improve health, wellbeing and reduce inequalities.

Petersen and Bunton's analysis provides an insightful critique of social implications of genetic technologies. However, their view, which rests on a juxtaposition between the structural (governmental) and individual (private) levels, may overlook some of the ways individuals relate to these technologies. It is possible, for example, that an individualistically informed choice model which bridges the structural and individual levels could also conform to how people understand and engage with genetic practices.

Other scholars like Lippmann, Kerr and Shakespeare hold a more critical position towards genetic technologies. They radically question the notion of informed choice itself (Lippman 1991; Lippman 1992; Lippman 1994; Lippman 1998; Kerr and Shakespeare 2002; Kerr 2004). They argue that individuals who enter the biomedical system are not empowered, but rather subjected to political, social and cultural pressures. These commentators emphasise that genetic technologies deliver complex information which can be difficult to interpret, and can be both intellectually and emotionally overwhelming for patients. Secondly, choices always happen in a social, political and economic context. The healthcare system, families, friends and wider society (e.g. media, internet etc.) exert a strong influence on the views that patients hold, and the decisions they make in our society. It is tacitly assumed that responsible action consists of the avoidance of unnecessary private and social problems, as well as economic burdens. Thus, for these scholars, genetic technologies compel individuals to engage in risk minimization behaviours, rather than empowering them.

It is plausible to think that in their attempt to inform social change in order to reduce potential oppression and inequalities stemming from biomedicine, scholars like Kerr, Shakespeare and Lippmann conceptualise patients and individuals in an overly restrictive way i.e. as being particularly passive and defenceless vis-à-vis biomedicine, society and other forms of authority. In fact, in emphasising how genetic practices could mask operations of power and act as constraints to individuals' autonomy and free choice, these scholars conceive individuals as being unable to make sense of genetic information, exert their agency or resist biomedical models of disease. Furthermore - as has already been observed with respect to Petersen and Bunton - this approach dismisses the possibility of a conflation of individual and structural agendas (Clarke, Shim et al. 2009).



The writings on biological citizenship by Rose and Novas provide a different interpretation of the influence of genetics on subjecthood (Novas and Rose 2000; Rose 2001; Rose and Novas 2004; Rose and Novas 2005; Rose 2006). Rose and Novas are critical of the trend in sociology and anthropology (embodied by scholars like Kerr, Shakespeare and Lippmann) to react negatively towards innovation in biomedicine. The position held by these scholars strongly recalls the 'genetic view' prevalently held by nation states, professional literature, media and other social organs described in the work of Petersen and Bunton. Rose and Novas have in fact extensively argued that technological developments in biomedicine ultimately do have empowering effects, as they place individuals in the position to exert more control on their bodies and lives. Rose and Novas argue that innovations in biomedicine are contributing to a broad process of transformation of personhood along biological lines which characterises our contemporary society; in the authors' parlance, genetic risk is linked to the birth of the 'somatic individual' (Novas and Rose 2000; Rose 2006). For Rose and Novas, genetic practices are novel 'technologies of the self' which allow individuals to intervene in and modify an increasing number of aspects of their existence and reshape their identity.

Genetics is seen as providing useful and salient knowledge about the body, susceptibilities and risk, and with this knowledge, individuals benefit from the possibility of modifying and improving their lives, engaging for example in risk management and minimization behaviours - which the authors term 'genetic prudence' or 'genetic responsibility'. Rose and Novas apply motives of neo-liberalism to describe the impact of biomedical innovation on individuals. For the authors, the idea that life is a project and individuals are expected to work on themselves permeates the culture of our society. Hence, taking action to understand and minimize genetic risk is seen as an implementation of the general work already undertaken by individuals towards their

autonomy, self-actualization and optimization of life. Notions like genetic prudence and responsibility are incorporated into this supposed pre-existing idea of work on the self, and inform the way individuals conduct their life, their reproductive decisions, and their career aspirations, to cite a few examples.

With the thesis that biological life has entered the domain of choice, Rose and Novas eschew negative reactions to genetic technologies, like the ones of Lipmann, Kerr and Shakespeare. In contrast to models that envisage passive patients ineluctably subjected to biomedicine and other forms of social pressures in disguise, Rose and Novas propose an individualistic ‘informed choice’ model in which individual agency is more acknowledged and where there is also room for a conflation of individual and structural agendas.

Nonetheless, recalling the position of Petersen and Bunton and many other critics of the impact of biomedicine on society, Novas and Rose claim that citizens are expected to benefit from engaging with these technologies, but, at the same time, may also feel obliged to do so because of external social and political pressure. Rose and Novas point out that, ignorance about genetic information and fatalistic and hopelessness behaviours in the light of this information tend to be discredited in our society (Rose and Novas 2004). In other words, the promotion of empowerment, freedom of choice and life planning has a flip-side: citizens’ obligation and responsibility to engage with these technologies and to act to minimize risk. Therefore, Rose and Novas claim that in the socio-political context of many ‘advanced’ countries, active biocitizenship is not only desirable, but is also becoming obligatory.

However, Rose and Novas seem to loosen this tension between the structural and the individual by linking their conception of biocitizenship with the assumptions that guide

the neo-liberal forms of government discussed above. In their writings, the duty of citizens to seek information about their health and engage in risk minimization strategies tend to *coincide with* the individual's 'voluntary' desire to realise their potential, seek their wellbeing and optimize their life.

The core message conveyed by Rose and Novas is that biology, in other words our corporeality, is not a given and unchangeable fact. With scientific developments, our bodies are made increasingly knowable to us. Furthermore, with this knowledge comes more freedom i.e. the possibility of choosing strategies to modify one's life. Rose and Novas' (bio)citizens are active agents, who are increasingly in control of their health and disease and autonomously seek and *demand* advice on how to plan their life strategies from healthcare providers as well as alternative sources. Citizens are not subjected to expert knowledge and are not the victims of de-centralised post-welfare forms of governments. On the contrary, Rose and Novas view citizens more as informed consumers.

They also acknowledge that genetic information, for example a positive test result for Huntington Disease (HD), can cause feelings of anxiety, despair and fatalism in affected individuals and their families (Rose and Novas 2004). However, they claim that individuals invest in science and its progress: life sciences and biomedicine are prevalently linked to feelings of hope, for example, for future treatments. The authors also claim that for those directly and indirectly suffering from a genetic ailment, gaining more knowledge about the condition could serve as a technique or a guide in order to conduct and manage life in the face of illness.

Overall, it can be argued that with respect to the social implications of genetic medicine, the position of Petersen and Bunton represents a theoretical middle ground between the strong critical claims - which presuppose passive patients and a strong state - of scholars

like Kerr, Shakespeare and Lippmann and the more 'neo-liberal' view of Rose and Novas. In fact, Petersen and Bunton - whilst presenting the idea that the advancement in genetics may increase individual empowerment and control over risk and disease - also question the actual uptake of genetics in clinical settings and manifest concern towards the possibility that genetic practice may mask operations of power and control, deflecting attention from more urgent public health and structural reforms.

It is also important to highlight that Petersen and Bunton's position, despite being less critical than the perspective of Kerr, Lippman and Shakespeare, is nonetheless more prone to address crucial sociological issues of exclusion and marginalization in relation to genetic practice than Rose and Novas' account. In reading Rose and Novas' writings, it is tempting to ask *who* these biocitizens are. Rose and Novas' version of biocitizenship presupposes subjects who are ready and able to understand and engage with genetic medicine. Hence, their account may overlook those who may not experience innovations in biomedicine as primarily empowering and promoting the self-management of risk, like, for example, some NF1 patients who have learning difficulties and have to deal with uncertainty.

In addition to Petersen and Bunton, other commentators like Braun, Raman and Tutton have also raised the question of potential exclusion and marginalization with regard to biological citizenship (Braun 2007; Raman and Tutton 2010). These scholars point out that Rose and Novas do not sufficiently take into account marginalised populations (e.g. the poor, migrants, people in the underground economy etc.) which instead should be included as the central foci of social theories on biosciences (Braun 2007; Raman and Tutton 2010). In other words, the self-actualised, empowered citizens who can make choices are prevalently middle class wealthy people. I would also stress that, even in the

UK, where there is one of the world's most comprehensive publicly-funded health services, not all individuals may be able to access genetic services or understand genetic information.

Other studies, for example the work of Weiner, question the actual pervasiveness and generalizability of genetic responsibility and biocitizenship. Weiner focuses on patients' constructions of Familial Hypercholesterolemia and shows that also when individuals are equipped with the instrument to actively engage with genetic information they may not conceptualise and understand themselves in genetic or molecular terms despite the fact that genetic discourses for this condition are widespread (Weiner 2006; Raman and Tutton 2010; Weiner 2010). These studies highlight that genetic conditions do not have to be construed as genetic to be manageable. Thus, structural, economic and intellectual barriers as well as pre-existing ideas of self, health and responsibility can prevent citizens from actively engaging with genetic medicine.

I find these points about marginalization and exclusion particularly relevant with regards to NF1 and possibly with other conditions which can cause cognitive and physical difficulties. In fact, it is possible that under these circumstances, individuals and families may not be able to perform the behaviours of active citizenship envisaged by Rose and Novas and by public health discourses. Ablon hinted at this problem in her work showing that many NF1 parents found it difficult to coordinate and access healthcare service for their children and themselves (Ablon 1999).

Therefore, while agreeing with Rose and Novas about the importance of acknowledging the possibilities opened up by biomedicine, I am more convinced by Petersen and Bunton's concern that biomedicine may not be very accessible and democratic and may

even mask, or serve as a justification for, the lessening of welfare and public health structural reforms.

### **Genetic Subjecthood and Support Groups**

Rose and Novas argue that individuals' involvement with their genetic conditions can imply linking up with patient support groups or forming new ones. This collective aspect of biological citizenship further amplifies individuals' empowerment, as support groups can ultimately influence research to address their disease-related concerns. Hence, they claim that it is increasingly common for individuals diagnosed with a genetic disease and for their families to use this knowledge to seek (or to create) structural support like genetic support groups and websites. To corroborate this idea, they mainly draw upon data from web forums and online support groups of people affected by HD. They observe, for example, that HD bloggers tend to be involved in many activities like posting new research findings, making donations for research, engaging in various fundraising activities to promote the search for a cure, and are willing to take part in experimental clinical trials for potential therapies to cure HD (Novas and Rose 2000). It is, however, important to highlight that HD is a very distinct (and thus not necessarily generalizable) genetic disorder characterized by a very severe outcome, early loss of life and high penetrance. Nevertheless, although Rose and Novas' focus is mainly limited to this distinct condition, to specific patient organizations and to individuals who *do* participate, rather than those who do not, they extend their claims about new forms of activism based on shared biological identities to genetic disorders in general as well as susceptibilities for more common conditions.

In doing this, Rose and Novas link the idea of widespread collectivising implications of biocitizenship to Rabinow's notion of biosociality. Biosociality is a heuristic tool coined by Rabinow (Rabinow 1996) to name groups and identities that are forming or changing

around biomedical sites of knowledge (genetics, molecular biology, genomics) and institutions (industrial, academic, medical) (Taussing, Rapp et al. 2003). It mainly refers to the formation of new social and political groups related to a specific genetic asset where people align themselves in terms of genetic narratives and practices. In the wake of the Human Genome Project and the drive to find genetic underpinnings for disease and broader human characteristics, Rabinow hypothesised that genetics would play an increasing role in people's lives and subjecthood. Foregrounding the activities of certain patient groups (once again mainly on HD) Rabinow suggested that knowledge of genetic 'identities' would lead to the creation of new communities of shared recognition, joint advocacy or even activism (Hacking 2006).

With reference to my research, it is interesting to recall that Rabinow specifically mentions NF1 support groups as an example of biosociality:

There already are, for example, neurofibromatosis groups whose members meet to share their experiences, lobby for their disease, educate their children, redo their home environment and so on. That is what I mean by biosociality. I am not discussing some hypothetical gene for aggression or altruism. Rather, it is not hard to imagine groups formed around the Chromosome 17, locus 16,256, site 654,376 allele variant with a guanine substitution. Such groups will have medical specialists, laboratories, narrative, traditions and a heavy panoply of pastoral keepers to help them experience, share, intervene, and "understand" their fate.(Rabinow 1996:102).

The notions of biosociality and biocitizenship are both grounded on the core assumption that individuals can increasingly intervene and exert control over their lives (Rabinow and Rose 2006). Biosociality and biocitizenship require that scientific and technological developments in the field of genetics create novel and important opportunities for patients to understand and act upon their diseases, reshaping how they organise themselves into groups and the activities they undertake. These two notions are

ultimately the claim that genetic support groups and other forms of group activism based on biological conditions play a decisive role in individuals' forms of subjectivity and disease management.

In more practical terms, Rabinow, Rose and Novas see genetic support groups as being important sources of information, representing a valuable platform for patients, family members and interested clinicians to liaise and/or share important aspects of their lived experiences with a genetic condition. Another determinant role served by these patient organizations is campaigning for the rights of patients, normally to de-stigmatise a disorder, increase its visibility and to obtain more healthcare service for patients.

Rabinow, Rose and Novas also claim that biocitizenship and biosociality represent an innovative style of agency and activism around disease. Whilst they recognise that the existence of patient organizations and support groups predated the advent of genetic and genomic medicine, they point out that 'pre-genetic' patient organizations had different characteristics. The authors claim that 'pre-genetic' patients' groups were mainly involved in psychological support and fund raising activities. Biosocial patients' groups, instead, often form direct alliances with scientists as they are involved with researchers and medical experts with the aim of finding a cure and/or treatment for a condition (Rose and Novas 2004; Rabinow 2008). The crucial innovation of biosocial groups is that they are increasingly playing an active role in shaping the direction of biomedical scientific research. For Rose, Novas and Rabinow, biomedical progress and the consequent genetic knowledge of diseases has opened new possibilities of investigating the nature of diseases, giving the opportunity to patients groups to ally and collaborate with scientists, shaping research.



The ideas of biocitizenship and biosociality have been the object of investigation of many scholars (Gibbon and Novas 2008). Callon and Rabeharisoa conducted a detailed analysis on the genetic support groups for Muscular Dystrophy MD in France, the Association Française contre le Myopathies (AFM) (Rabeharisoa 2003; Rabeharisoa 2006; Callon and Rabeharisoa 2008). At first glance, it could be argued that the case of AFM appears to be an emblematic example of biosociality or biocitizenship. The authors show how individuals with MD, thanks to the AFM association's intense engagement in scientific research (e.g. supporting research to find gene mutations for MD) and social activities (e.g. the annual television programme Telethon), were able to construct an MD individual and collective genetic subjecthood.

For Callon and Rabeharisoa the MD gene played a key role in this process. The discovery of the MD gene mutations in fact not only had a significant impact on subsequent genomic research, but it also provided a biological explanation for the impairments and problems of people with MD. This phenomenon of re-biologization of disability also had very important de-stigmatising consequences, turning the status of individuals with MD from "marginalised freaks of nature" into "human beings in their own right" (Callon and Rabeharisoa 2008).

Alongside the gene, the second crucial aspect that made this association successful was the fact that members were directly involved in its activities. Since its inception, the AFM governing board was composed of patients and their families who directly engaged with scientific and clinical research and in the development of policy and management strategies. Patients and families became real stakeholders in genetic, medical and social networks; they actively participated, for example, in the creation of research facilities, in the diffusion of knowledge and collaboration with scientists in

genetic and medical research. This engagement represented for Callon and Rabeharisoa another important aspect that progressively built patients and families' MD subjecthood.

Although the AFM seems to possess enough of the requisites to corroborate the idea of biocitizenship, Callon and Rabeharisoa hold a more sophisticated view on the formation and functions of genetic support groups. For the authors, AFM represents an exceptional situation rather than a generalizable one. They seem to agree with the idea that the developments of economic markets, combined with technological and scientific innovations in medicine, can facilitate the rise of patient organizations. However, they also acknowledge that these organizations are not necessarily successful. As Callon and Rabeharisoa point out, concerned groups, particularly at their inception, do not necessarily present a shared identity and interests; these tend to be achieved as a result of the activities of the group, but only when these activities are successful as it appears for the case of the AFM. Genetic support groups may not be economically sustainable and may easily face marginalization when trying to generate research, as the diseases are generally rare. Pharmaceutical companies, for example, may refuse to invest in areas of research that will only benefit a limited number of customers. Moreover, as there may not be so many individuals affected with a given condition and they may be located in different parts of the world, even when there are clinicians interested in further researching these diseases, it may be complex to collect a sufficient amount of cases for the clinicians to study (Panofsky 2011).

Thus, notwithstanding the claims of biosociality and biocitizenship, understanding the actual agency of genetic support groups and of individuals with genetic conditions and their families is a very challenging task. Genetic support groups tend to be composed of patients, families as well as researchers and clinicians who have an interest in the

specific condition. It is not easy to schematise and generalise the internal dynamics of these social assemblages. Clinicians and scientists may strongly influence the establishment and agenda of support groups, limiting patients' control and involvement. As some scholars point out, clinicians and medical professionals still dominate the membership of important committees - in particular the ones related to research funding and public education, which represent core interests of support groups (Petersen and Bunton 2002; Kerr 2003; Raman and Tutton 2009; Panofsky 2011).

The influence of support groups is more complex than envisaged by the biocitizenship and biosociality formula, under which patients and lay persons have the potential to influence experts and shape research. Again, the idea of empowerment that pervades the notions of biosociality and biological citizenship seems to be modelled upon cultural motifs of neo-liberal societies, especially the emphasis on the values of individual autonomy and entrepreneurship and the de-centralization of the state. Biosocial support groups appear to be the social version of the entrepreneurial autonomous individual. In de-centralised forms of government where the welfare state is slowly disappearing (due to structural changes and the effect of recession) it is very unlikely that the state will invest in research and the provisions of structural support for people who are diagnosed with rare genetic diseases. Thus, if the medical support provided is not sufficient and the disorder is difficult to manage, affected individuals and their family members may coalesce to form networks of mutual support. Given that genetic technologies are regarded as capable of increasing the understanding of the underlying mechanisms of diseases and developing better forms of treatments, individuals with a specific genetic condition may be expected to attempt to form alliances with clinicians and scientists interested in their conditions. As argued by Rose, Novas and Rabinow, these interested

individuals may foster the research on their disease attempting to minimise risk and optimise their lives.

I would argue that the description of patient groups as examples of biocitizenship or biosociality could be regarded as a possible embodiment/example of the current British Prime Minister Cameron's Big Society. It is very tempting to draw structural similarities between Rose, Novas and Rabinow's claims that affected lay people are/ought to be empowered by the 'new genetics' (and become increasingly capable of influencing their lives, shaping genetic research) and the communitarian rhetoric of the Big Society. The Big Society aims to create a robust citizenship in which individuals actively seek 'to do good' including for example getting involved in setting up their own local free schools, running local libraries or purchasing road signs. The government programme is to delegate power and responsibility to local groups and citizens, withdrawing from its responsibility to provide particular services. This withdrawal is justified by the belief that services are better administered by interested local communities composed of active citizens, but is probably also caused by the need to save money (Britain as of 2011 - has one of the worst deficits in the European Union). Concerning genetic disorders and their managements, given the lack of resources available in a de-centralised state, individuals and affected groups are expected to exert their autonomy and their role as active citizens in managing their disease, thanks to the possibility offered by new technology and the free market elicited by the de-centralised liberal state. The power (and responsibility) of intervening on health and disease is devolved from experts and political authorities to citizens and lay people.

From a more micro-level perspective, Petersen's analysis of illness narratives of members of genetic support groups for cystic fibrosis (CF), haemochromatosis,

haemophilia and thalassemia reported that genetic support groups played an important role in their illness experiences (Petersen 2006). Participating in support group activities was presented as part of the process of learning how to manage their genetic conditions. These groups were described as important sources of information, especially around the time of diagnosis and whenever healthcare professionals' opinions were conflicting or unclear. The possibility of sharing and comparing illness experiences with others, could also allow support group members to better understand the nature and scope of the constraints that their condition could pose on their life, helping in reconstructing their identity.

In line with the ideas of biosociality and biocitizenship, support groups were often seen as important sites of normalisation, identification and management of uncertainty and other hindrances posed by genetic conditions. However, as Petersen himself points out, participants in his study were recruited from those who joined support groups, but he did not interview those who did not join. As stated above, being aware of this problem in my research, in order to secure a more representative sample, I did not recruit from NF1 support groups, but from the database of a local hospital, as well as outside the NHS, among the so called healthy volunteers.

Ideas about the importance of support groups, and the suggestion that individuals with genetic conditions or susceptibilities identify themselves and guide their conduct according to genetic information, have been questioned more openly by several studies. In Lock's work (2008) on Alzheimer's disease (AD) for example, individuals with an AD family history who were involved in research on genetic susceptibilities for the condition did not appear to endorse genetically informed identities. Participants appeared to have filtered and interpreted the estimates they had been given through their

experience with family members with AD, the recollection of their family history and wider media and social discourses on AD. The vast majority of respondents did not join support groups. They relied instead on familial or other networks, as they were too busy to participate in support group activities, or they appeared to be in denial, due also to feelings of shame and stigma associated with having family members with AD. Support groups were also avoided because they were seen as sites for unnecessary and potentially overwhelming pseudo-counselling activities where members share their problems (Lock 2008).

Similar findings are also reported with other disorders; of particular interest for this thesis is Ablon's qualitative research on NF1 (Ablon 1999). As already mentioned in Chapter 2, Ablon noted that many respondents with the condition did not join NF1 support groups (Ablon 1999). Feelings of anxiety related to fear of meeting others with the condition and denial emerged as important aspects that prevented the actual attendance of support groups. Some respondents who attended at least one group meeting affirmed that seeing other individuals with NF1 (especially if more severely affected) helped them to balance the perceptions of their own condition. For others, meeting individuals with similar or even worse symptoms was described as a distressing experience. It also emerged that, notwithstanding their symptomatology, many individuals (especially male ones) did not join because they felt no need of support (Ablon 1996). Ablon hypothesised that the specific variability of NF1 could represent a hindrance for the construction of a common (disease) identity and sense of community- in Ablon's terms 'a common world and concern'- that are the foundations of genetic support groups (Ablon 1999).

Ablon's hypothesis of the lack of 'a common world and concern' for NF1 draws important links between the variability of the condition and how it can influence the

way it is experienced by affected individuals. Nonetheless, as I discussed, Ablon's main interest was adaptation to stigma (Ablon 1995; Ablon 1999; Ablon 2000; Ablon 2002); she did not extensively explore the role played by support groups on individuals and family experiences with the condition in the light of genetic knowledge and technologies. These aspects instead will be important foci of my research.

### **Uncertainty**

The assumption that individuals engage with genetic knowledge about their condition and that the more individuals are informed about their health and disease the better they manage it, remains largely undisputed in works such as that of Rose and Novas. Yet - as discussed in the next section- genetic information is linked to uncertainty and therefore, as commentators like Lippman, Kerr and Shakespeare have highlighted, it can be difficult to manage both by healthcare professionals and patients (Lippman 1998; Kerr and Shakespeare 2002; Kerr 2003; Kerr 2004). Experts may disagree on how to interpret a test result and a genetic diagnosis may be more confusing than empowering. Often diagnoses are not accompanied by (clear) therapeutic pathways, and there may be a therapeutic gap between diagnosis and treatment (Marteau and Croyle 1998; Marteau 1999). Thus, not only might affected individuals be overwhelmed by the uncertainty related to genetic information, but they may also not have the means to engage in risk minimization strategies.

Empirically grounded research on patients' experiences with a range of different genetic conditions and risks has questioned the actual uptake of genetic aspects in individuals' illness accounts, showing the complexity and heterogeneity of their discourses on health and disease. Notably, patients' accounts appear to incorporate un-geneticized, 'lay' perspectives, including familial and broader intersubjective dynamics (Sanders, Campbell et al. 2007; Clarke, Shim et al. 2009). Moreover, the development of genetic

practices is not necessarily accompanied by a radical change in professional and patients' conceptions of disease.

Cox and Starzomski's qualitative study of Polycystic Kidney Disease PKD (a common but relatively unknown genetic disorder) raises a number of important points in relation to these issues (Cox and Starzomski 2004). Their study, based on a series of interviews with healthcare providers, patients and their family members, focused on the social construction and clinical management of the condition. Although PKD is a genetic condition with clear genetic markers and testing, the authors found that there were two distinct constructions of the condition: 1) a chronic disorder that can cause gradual loss of kidney function and 2) a life-threatening inheritable (in autosomal dominant fashion) genetic condition. Interestingly, the second construction, the genetic, was the less predominant one.

As regards the healthcare system, interviews with medical professionals showed a certain distance and lack of collaboration between nephrologists and medical geneticists. While treating patients, nephrologists displayed a tendency to deflect on the genetic aspects of the condition or even to avoid presenting PKD as a genetic hereditary disorder. They feared in fact that dwelling on the genetic aspects of the condition would foster feelings of fatalism in patients, creating an obstacle to patients' compliance to treatment. Moreover, since PKD also has a variable and uncertain prognosis within families, nephrologists were afraid that stressing the genetic side of the condition may lead patients to trace a parallel between their disease trajectories and other affected family members. The utilization of the family as a benchmark could lead patients to deceptive and unfounded optimism or pessimism towards the development of their own condition.



Moreover, genetic pre-symptomatic testing, despite its availability, was not employed in clinical practice, but diagnoses were based on clinical evidence. This mainly depended on the cost of genetic testing and on the fact that both patients and healthcare professionals saw pre-symptomatic genetic diagnosis as having potentially discriminatory and stigmatising repercussions (for example in terms of life insurance and employability).

Also patients and families constructed PKD predominantly as a chronic kidney disease. Not only did they overlook the genetic aspects of the condition, but they also appeared to have an erroneous understanding of the mechanism of inheritance.

In summary, PKD was mainly constructed and managed as a chronic kidney disease and not as a genetic condition. With this study, Cox and Starzomski provide a remarkable example of how the discovery of the genetic mechanisms of disease and the development of genetic testing may not necessarily find immediate application in clinical practice and may not necessarily alter patients and families experience of disease.

These findings can be significant for this Ph.D. research particularly because, as with PKD, whilst genetic testing is theoretically available for NF1, professional literature reports that it is not commonly employed and diagnosis is mainly performed on clinical evidence. Moreover, NF1, like PKD, is a variable condition. Thus, there may be some common patterns in the way PKD and NF1 are constructed by healthcare professionals, patients and families. For example certain clinical characteristics of NF1 may be as - or more - salient than its genetic aspects.

Similarly nuanced scenarios have been found in investigations that explored other genetic conditions. For example, Bharadway and colleagues conducted in- depth

interviews with individuals affected by, or at risk from, iron overload from genetic hemochromatosis; their results show an interesting relation between the medical classification and patients' experiences of the disease (Bharadway, Atkinson et al. 2007). Although the genetic base and inheritance pattern of this condition has been found, the links between genotype and phenotype are still unclear. The disorder has highly variable penetrance, symptomatology and severity; thus a positive test result simply reveals that the individual is susceptible to the development of the condition. Consequently, as with the case of PKD, the disorder is still mainly diagnosed and managed clinically. The patients interviewed attributed minor importance to the genetic aspects of their condition, but they were more engaged with its clinical side, because for them early and accurate clinical diagnosis was very important. In fact, if untreated, hemochromatosis can cause serious organ damage (due to iron overload), but if it is detected at an early stage the clinical management is straightforward (the iron overload is reduced by regular bleeding i.e. phlebotomy). As the onset of this condition is insidious and its symptomatology has not been systematically determined yet, a timely and accurate diagnosis can be difficult to achieve and the disorder can progress unrecognised. To diagnose the condition, clinicians rely upon a set of principal features of the disorder based on previous medical studies. However, the patients interviewed presented with symptoms that were divergent and different from the clinical description of the disease, and they challenged clinical authority, claiming that these symptoms were part of their disease.

In summary, as with PKD, patients' illness discourses leaned more towards the clinical side of their disorder and on aspects of the symptomatology that affected them personally, rather than the genetic nature and origin of their condition. However, though

participants were not passive towards medical practice, they contested the clinical definition of their disease in order to redefine it and include their own nosography.

In contrast to accounts such as Kerr, Shakespeare and Lippmann's which envisage individuals as being eminently passive to medical practice, this study illustrates interesting modalities in which patients can express their agency and resist medical authority. These patients, in negotiating the clinical definition of their disorders with healthcare professionals conform much more to Petersen and Bunton's critique of individualising trends in public health towards patients' responsibility toward management of diseases, or Rose and Novas' model of active biocitizenship.

Furthermore, the issues of negotiation of the clinical definition of the disorder and the need for timely and accurate diagnoses could be potentially important with respect to NF1. Early diagnosis may increase monitoring and early intervention which can be crucial with a syndrome that can cause, among other things, malignancy and cognitive problems. Yet, NF1 is mainly diagnosed upon clinical evidence, and, since the manifestation of its features is age dependent, individuals with the condition may be diagnosed only later in life.

Concerning patients' experience of illness, Klitzman's interviews of individuals who had or were at risk from Huntington's disease, breast cancer or alpha1 antitrypsin deficiency revealed that their ways of relating to and engaging with genetic information were highly variable and subjective. Klitzman suggested that personal aspects linked to the disease, family history and other familial dynamics were important factors that could influence the modality and extent to which genetic information affected their identity (Klitzman 2009).

Petersen's analysis of illness narratives of members of genetic support groups for cystic fibrosis (CF), hemochromatosis, haemophilia and thalassemia provided even greater evidence of patients' minimizing and downplaying attitudes towards their conditions (Petersen 2006). Participants in his study, whilst acknowledging their health problems, tended to minimize the overall impact of genetic disorders on their lives. They compared, for example, their situation with what they perceived to be more serious scenarios of other individuals, emphasising as well the positive aspects of their disorder and the "lesson learnt" from dealing with it. Interviewees tended also to present themselves as active agents who were in control over their genetic condition and indeed able to be of assistance to others. These discourses appeared to be common, irrespective of the severity of the disorders.

The fact that individuals with different genetic disorders may not necessarily endorse a univocal medical disease identity and engage in monitoring and risk minimization behaviours, challenges many of the assumptions of biocitizenship, but also its opposite model of the passive patient subjected to biomedicine. Empirical studies show a nuanced picture in which individuals do not necessarily engage with their disease or risk in a uniform and predictable way. Notably, affected individuals and their family members can also deploy "downplaying" attitudes towards genetic disorders (Sanders, Campbell et al. 2007).

Therefore, I would argue that social theorists like Rose and Novas, in producing general frameworks for understanding the implications of human sciences on a global socio-political scale, may lose contact with people's experiences of genetic disease, and often bypass the clinical and lifeworld (Mishler 1986) complexity of engagements (and disengagement) with genetic information. The same point can be raised with respect to a

lot of research in this field -including works that take a more negative attitude towards genetic technologies (Lippman 1991; Lippman 1998; Kerr and Shakespeare 2002). The work of Petersen and Bunton maintains instead a more critical and open viewpoint on the social impact of genetic medicine also questioning its actual uptake in clinical settings. Thus, it offers a theoretical framework which can also better accommodate the fine-grained results emerging from empirical research into patients experience with and healthcare management of genetic disorders.

### **3.3 Genetic Medicine and Family Dynamics**

#### **Medicalization of the Family**

The interrelation of individual and familial dynamics around genetic conditions is a central interest of my thesis. Because genetic diagnoses are inherited and distributed amongst family networks, families are important sites for exploring their experiences with genetic disorders. In this section, I will review anthropological and sociological studies on genetic medicine which have focused on the cultural and social implications of biomedical technologies on individuals' familial relationships and bonds.

Some of this research, like that conducted by Finkler (Finkler 2000), suggests that advances in genetic medicine are leading to a process of medicalization of the family and kinship, which implies that family members are brought closer by their shared genetic profiles. Finkler embraces the already discussed common view that biomedicine is profoundly altering individuals' experience of health and disease as well as medical practice. Following from this premise, and by emphasising the familial implications of genetic information, she concludes that biomedicine is also significantly changing how individuals construct their familial relations and bonds. As already mentioned with regard to Petersen and Bunton's work, Finkler argues that since diseases are

increasingly rooted to genetic inheritance and families have come under close surveillance, blood ties have become more salient.

The idea of medicalization of family and kinship is presented by Finkler as stemming from her reflections on the effect of media and public hyped discourses on the 'new genetics', alleged changes in biomedical practice and from fieldwork she conducted in the USA. The fieldwork is an ethnographic research based on open ended interviews with women with breast cancer, men and women with a family history of cancer and adoptees in search of their biological mothers (Finkler 2000; Finkler 2005).

The process of medicalization introduces a new way of conceiving familial relations that emphasises genetic problems over other aspects like social, economic status as well as affective and geographical proximity. Individuals are described as becoming more aware of sharing genotypes with their kin and more prone to engage with their kindred in practices of disclosure, health management, control, surveillance, sharing as well fears and anxieties related to their health. Blood relatives, for example, are brought closer even when they are affectively or geographically distant, not just by the common burden of having a genetic problem, but also because they experience the general sense of sharing the same genotypes. Blood relatives share 'the same genes and the same destiny' (Finkler, Skrzynia et al. 2003).

The medicalization of the family can result in the construction of new family networks which are not built upon choice, but upon angst of having or being predisposed to a disorder, or more generally of a shared biological makeup (Finkler 2005). Recalling the idea of biosociality, these alleged new networks can be artificial from a social and affective point of view, but natural from a biological one. In strengthening conventional

categories of biological relatedness, this process of medicalization, can revive or reinforce kindred bonds, but, at the same time, it can also counter individuals' freedom to choose whom to regard as family. In fact, the genetic bonds that are made explicit by a clinical family history do not necessarily coincide with what counts for individuals as family.

Therefore, the medicalization of the family, by narrowing family to genetic bonds, is counter to the changes that family have undergone since the second half of the 20<sup>th</sup> century. Our contemporary experience of the family (in Finkler's words 'the family in the postmodern society') which emphasises choice over biological ties, is altered and brought back to a traditional definition - pre 1960s- based more on biological ties (like consanguinity, marriage and procreation) than choice (Finkler 2000).

In more general terms, the medicalization of the family is also regarded as an obstacle to individuals' autonomy and freedom of choice. Being autonomous, independent, standing outside socially defined groups and choosing freely one's lifecourse are mentioned by Finkler as being important values of many contemporary societies, especially in North America, where her study is set. As the medicalization of the family emphasises biological connectedness, promoting a fixed and circumscribed model of family, it is at odds with these values. Kinship relationships based on genetic inheritance can potentially limit individuals' choice of whom to regard as family and it can also constrain life choices for example by generating dilemmas and responsibility concerning disclosure and risk management practices.

These aspects touched by Finkler resonate with the question of the relation between medical genetics and individual agency already discussed in the previous section. In particular, there are important correlations between the notion of genetic responsibility

and medicalization of the family, as Finkler, Rose and Novas draw more or less directly on a neo-liberal model of individualism, choice and autonomy in their interpretation of the social impact of biomedicine. It can be argued that both these notions imply a tension between individual and family, that is, the familial bonds stressed by genetic relatedness are mainly interpreted as possible constraints to individual autonomy, independence and freedom of life choices.

In her writings, Finkler seems to agree in principle that individuals can resist this process of medicalization and that family and kinship relations can still maintain some flexibility. Finkler's presentation and discussion of her data show cases where individuals' experience of kinship did not necessarily coincide with its biomedical model. For example, some of her participants with a family history of breast cancer appeared to feel closer to their stepsiblings or other non-biologically related members of the family, while distant towards other family members to whom they were related by blood (Finkler 2000; Finkler, Skrzynia et al. 2003). Nevertheless, Finkler emphasises that these individuals were living with the constant worry of developing cancer. By dwelling on these feelings, they were also reconfiguring their family structure, bringing blood relatives -albeit affectively and /or geographically distant -closer.

For Finkler, genetic discourses and practices are part of a dominant belief system that strongly affects the construction of subjectivity and families; in her words we are living the age of the 'hegemony of the gene' (Finkler 2000; Finkler, Skrzynia et al. 2003). Thus - as other scholars like Petersen, Bunton, Rose and Novas have argued- according to Finkler, individuals are bombarded with media, governmental and other forms of discourses that convey the idea that health problems and other aspects of life are explainable in genetic terms.



Moreover, for Finkler, individuals' hypothetic residual forms of resistance to this genetic world view are further undermined once they enter the medical system where the ideology of genetic inheritance is incorporated into clinical practice. This point is not dissimilar to the model discussed earlier of the passive patients subjected to biomedicine, as conveyed by scholars like Kerr Shakespeare and Lippmann.

In summary, according to Finkler, as a result of this strong and pervasive 'genetic' bombardment, individuals' family and kinship relations are likely to undergo a process of reconfiguration upon which biological ties rather than affective and other social attributes are emphasised.

The work of Finkler, by increasing researchers' awareness of the importance of families and the implications of genetic medicine on kinship and family relations, has certainly provided a significant contribution to the research on the social implication of genetic medicine. Nonetheless, the idea of medicalization of the family rests on an assumption - namely, that genetic discourses and practices are pervasive in clinical practice and exert a strong impact on individuals' identity- which also underpins the already discussed work of other scholars (like Petersen, Rose and Kerr) and can be problematic. For example, as I showed in the previous section, research on patients' illness experiences with a range of diverse genetic conditions and risk has highlighted many aspects that can mitigate the idea of a strong uptake of genetic positioning. Genetic practices may not enter into healthcare settings and alter the everyday clinical management of disorders; and above all they may not profoundly change patients' illness experience (Petersen and Bunton 2002; Cox and Starzomski 2004). Moreover, Finkler's view- like the theoretical constructs which assume passive patients subject to biomedicine (Lippman 1991; Kerr and Shakespeare 2002) - may overlook individuals' agency and forms of resistance to medical, public health and broader social discourses on genetics.

If this is the case, then the medicalization of the family could be a less widespread phenomenon than suggested by Finkler. In fact, it is reasonable to think that individuals who do not significantly experience their disorders in genetic terms may also not necessarily be inclined to reconstruct their familial bonds according to genetic ties. Finally, it is important to stress that she focused on breast cancer which is an exceptional condition particularly tied with public health campaigns and pre-existing discourses of womanhood and motherhood.

### **Genetic Responsibility**

As already observed, given its intergenerational implications, genetic information can pose challenges and dilemmas for affected individuals concerning, for example, their reproductive choices as well as disclosure and management of genetic risk with other people, particularly with kin and extended family members. These particular aspects of the genetic subjecthood have been encapsulated in the notion of ‘genetic responsibility’. The term ‘genetic responsibility’ epitomises important intersubjective and biographical factors that can influence individuals’ constructions and management of information about their genome. Broadly conceived, it denotes the possible relations and duties that individuals with genetic susceptibilities, risks, or disorders appear to have towards themselves in terms of their own identity, reproduction and health and towards other people, mainly family members (Graham 1979; Hallowell 1999; Novas and Rose 2000; Petersen and Bunton 2002; Hallowell, Arden-Jones et al. 2005; Arribas-Ayllon, Sarangi et al. 2008).

Petersen’s empirical study based on interviews of members of genetic support groups for cystic fibrosis (CF), haemochromatosis, haemophilia and thalassemia shows that individuals can display concerns in transmitting their genetic conditions to future generations to the extent of deciding sometimes not to have children. In justifying these

decisions, they tended to present themselves as morally responsible citizens who sacrificed their personal aspirations of parenthood in the interest of the quality of life of the hypothetical child (Petersen 2006). Making the decision about whether to take the risk of having children can also be interpreted as an attempt to regain some sense of control over genetic conditions, especially towards their uncertainty (Petersen 2006; Kelly 2009).

From the standpoint of the medicalization of the family, the cluster of familial links and duties encapsulated within the notion of genetic responsibility could be interpreted as possible forms of constraint that limit individual action and life. However, other studies have shown that genetic responsibility behaviours can be construed and enacted by individuals in more complex and fine-grained ways.

A compelling example can be found in Hallowell and colleagues' work on patients' perceptions of their genetic risk for hereditary breast/ovarian cancer (HBOC) (Hallowell 1999; Hallowell, Arden-Jones et al. 2005; Hallowell, Arden-Jones et al. 2006).

Hallowell and colleagues conducted several studies based on in-depth interviews with patients identified to be at risk for HBOC. Their research suggests that participants, both men and women, were worried about the potential familial implication of their risk and described genetic testing as a familial duty. In line with the ideas of genetic responsibility and of medicalization of the family, their principal motivation for undergoing BRCA 1/2 testing was to ascertain the risk to their children and other family members, rather than their own. However, this research also suggests that both women and men actively constructed and re-constructed genetic risk and responsibility in ways that were functional to the affirmation of their identity as individuals, parents and family members. Genetic information was received and interpreted within and adapted to a pre-existing array of identities.

The women interviewed undertook risk monitoring practices despite the fact that some of these medical procedures (e.g. annual mammography and ovarian ultrasound) were potentially iatrogenic. They also disseminated information about risk prevention and management to their family and even friends. For Hallowell, part of the justification of these women's behavior resides in the already presented general assumptions underlying public health and biomedical discourses that individuals should increasingly take control over their health (Petersen and Lupton 1996).<sup>8</sup> With regard to genetic medicine, the care of the self is extended to kin and family and intertwines with general discourses of parenthood.

Hallowell suggests that genetic testing limited women's choices about how to manage their health and conduct their lives. Because of their propensity to feel responsible for the care of other family members, women may feel obliged to know their risk and engage in risk minimization actions. However, the women interviewed actively re-interpreted genetic practices by integrating them with their constructions of the self as women and mothers. Engaging with genetic information and risk minimization behaviors was presented by them as a part of the fulfillment and affirmation of their identity as careers and relational selves.

Thus, in line with Finkler's idea of medicalization of the family, genetic ties were relevant for the women interviewed. Not only did women mainly justify engaging in risk assessment and monitoring behaviors as a parental duty towards their children, but they were also involved in disclosing information about the importance of risk prevention within their family network. Nevertheless, in contrast to Finkler's interpretation, genetic practices did not necessarily alter or constrain the ways these

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<sup>8</sup> See also (Lippman 1991; Lippman 1992; Lippman 1994; Rose 2001; Kerr and Shakespeare 2002; Petersen and Bunton 2002).

women construed their identity and family relations. Rather, Hallowell's analysis illustrates how women's narratives conflated elements of genetic responsibility with more general preexisting discourses of motherhood and womanhood (Graham 1979).

The investigations conducted on men show even more manifold forms of engagement with genetic risk and responsibility. The men interviewed presented different typologies of genetic responsibility discourse. In the first place, like the women, all men interviewed justified undergoing testing as a duty towards their children. Furthermore, the authors suggest that participants deploy other understandings of genetic responsibility and risk which appeared to be dependent on their carrier status.

Those who were found not to be carriers for the BRCA 1/2 mutations presented contradictory feelings of relief and guilt. With respect to their children, men appeared relieved for not carrying the mutation. Nevertheless, they also tended to express feelings of unhappiness and survivor's guilt for their negative results if other family members were found to be positive. On the other hand, men who actually found to be positive carriers for the BRACA 1/2 mutations, instead of blaming themselves and feeling responsible, adopted a fatalistic attitude towards genetics to decrease their guilt and responsibility. They tended to adopt a deterministic view of genetics distancing themselves from the idea that they transmitted the mutations. This attitude towards genetic risk appeared to be functional to rid carriers from the sense of guilt related to passing on to children the mutation. Individuals adopted fatalism to absolve themselves from the responsibility for their own health and that of their children (Keeley, Wright et al. 2009).

As mentioned above, genetic responsibility could be interpreted in line with Finkler's idea of medicalization of the family, as a possible form of constraint to individual choice and actions. However, by showing these different complex responses to genetic

practices, Hallowell and colleagues suggest that both men and women can draw on discourses of responsibility, care and guilt to affirm their identity as individuals and family members. These studies also reinforce the idea presented previously that individuals' modality of interpretation of genetic knowledge are unpredictable, complex and situated.

Nonetheless, it is worth mentioning that Hallowell's and many other studies of genetic responsibility have recruited participants from clinical genetic services and have focused on distinct and known conditions like HBOC or HD which have a relatively straightforward genotype/phenotype correlation. Patients and family responses to genetic information may differ in the case of multisystem genetic disorders like NF1 which are highly variable and unpredictable and may be managed, alongside clinical genetics, by many other different medical specialties.

### **Lifecourse Foregrounding of Genetic Risk**

Empirical research conducted on familial experience with genetic conditions has also highlighted that, contrary to the idea of medicalization of the family, individual and familial perceptions of relatedness were not significantly determined or reconfigured by a biomedical definition of inheritance. Patients can express complex and sometimes inconsistent beliefs about inheritance and genetics, which are often influenced by their interactions with other family members (Cox and McKellin 1999; Featherstone, Atkinson et al. 2006; Lock and Nguyen 2010). Furthermore, empirical research has also suggested that the way individuals perceive inheritance is shaped by their perception of emotional relationship, duty, and responsibility that compose family ties (Richards and Ponder 1996). Therefore, the experiences and courses of actions of families with genetic conditions are influenced by complex biographical, temporal and familial aspects

(Parsons and Atkinson 1992; Cox and McKellin 1999; Featherstone, Atkinson et al. 2006).

Parsons and Atkinson conducted extended unstructured interviews with mothers and daughters in families with Duchenne Muscular Dystrophy (DMD) to analyse the ways they construct and define their carrier risk status (Parsons and Atkinson 1992).

DMD is a muscle wasting disease that manifests only in males, but is genetically transmitted by females. Women who have a male family member with a DMD are normally referred to medical geneticists who generate an estimation of their carrier risk based on family history and clinical tests.

All the women interviewed were given estimations by specialists of their carrier risk at a certain point in their life. In discussing these estimations with the researchers, they demonstrated varied personal reactions and attitudes towards their risk. Risk appeared to be prevalently latent and back-grounded, but it also tended to become salient at certain critical junctures in the lifecourse of these women; the authors call this phenomenon 'life course fluctuating relevance of risk'. Commonly, these junctures were related to familial, social interactions and in particular to reproductive decisions. In fact, the majority of the women in this study who displayed an interest in their genetic risk were younger women who were planning to have a family or were still family building. Women who were potentially or actually involved in reproductive decision making, placed their risk in zones of high relevance because they saw it as something of immediate personal concern. They tended for example to convert numerical risk figures into more simple terms (i.e. "low/ high/ fifty- fifty" risk of passing the condition).

In summary, risk was not only understood in terms of its biomedical calculation and its perceived relevance was not constant and consistent throughout individuals' lives. The

relevance of risk was the result of a fluid process of negotiation between medical calculations, and individuals' everyday life and needs, particularly familial reproductive aspects.

Similar results emerged in Cox and McKellin's study based on in-depth interviews of individuals at risk for HD and their family members (Cox and McKellin 1999). Despite the differences between MD and HD, the authors found that also the relevance of genetic risk for HD appeared to be 'fluid and contingent', changing throughout participants' lifecourses. Once again, the relevance that participants gave to their hereditary risk for HD was shaped and differentiated by nuanced biographical and familial factors, rather than simply by its biomedical calculation. Moreover, the authors point out that geographic and emotional closeness to an affected family member appeared to be as important as biological ties in explaining how participants and their families constructed their hereditary risk for HD. Family members that are close in a family tree may be very distant geographically and emotionally in real life. Thus, in contrast to the medicalization of the family model which presupposes a correspondence between biological and social closeness, the pre-existing closeness or distance of family bonds and relations could shape how the 'objective' genetic closeness was perceived by participants.

Notwithstanding the objective diagnosis of a parent, if HD did not enter everyday family discussions and/or if the affected parent died of unrelated causes, children tended to conduct their lives without taking their risk too seriously. Another important element that altered the relevance of genetic risk was related to family dynamics and the natural course of aging. Probably due also to the late onset of HD, whilst still young, individuals minimised their risk, perceiving it as something distant. However, getting



closer to the age of onset of HD of a family member, could bring the risk closer. Moreover, becoming adult can coincide with important life planning decisions, for example about having children or about career aspirations. For some interviewees, reaching adult age could bring their risk closer, leading them for example to decide to request predictive genetic testing for HD.

Cox and McKellin's study shows the limitations of biomedical models of inheritance and kinship, highlighting the importance of understanding the construction of genetic risk within the lived experiences of individuals and their family members. HD risk did not necessarily appear to be inherently problematic for individuals. They rather interpret and re-interpret genetic information: they can downplay their risk but also give it high relevance at certain critical junctures in their lifecourses. It is particularly remarkable that such varied and nuanced interpretation of the relevance of genetic risk were also found in a study on HD which is a condition that has a relatively clear penetrance and straightforward phenotypic outcomes.

This study also shows that the subjectivity and responses of individuals at risk for HD are much more complex than depicted by Novas and Rose. Unlike Novas and Rose's ideas, individuals at risk for HD do not necessarily react by endorsing a HD subjectivity, seeking more information and knowledge about their condition and acting in genetically prudent and responsible ways. They may instead downplay or decide to ignore their HD risk, or –as reported in other studies (Soltysiak, Gardiner et al. 2007) - their adjustment to the diagnosis may not be immediate. It is also indicative as Cox and McKellin report in their study that during their research in British Columbia, Canada, where the study was conducted, only 10-15% of the population found to be at risk for HD requested the genetic test.

In summary, individuals and families tend to display more fluid, original and creative ways of adapting to genetic practices. It is also very important, in the light of my thesis' interest on family dynamics, to reiterate that a significant part of these critical junctures appear to be related to family (Petersen 2006; Keenan, Teijlingen et al. 2009). Family dynamics are strongly intertwined with individuals' lifecourses and represent a crucial heuristic space that shape individual experiences of self, disease and genetic knowledge. An analysis of the evidence above shows that the notions of medicalization of the family and genetic responsibility correctly individuate the familial implication of genetic knowledge. However, they tend to depict overly abstract and fixed models of description of individual and familial engagement with genetic knowledge. Alongside recognising the importance of familial implication of genetic knowledge it is also important to look deeper into families' lifeworld, acknowledging that the same families and individuals can significantly change their modality of engagement with genetic knowledge throughout their life. Constructions of genetic knowledge can fluctuate depending on individual and familial lifecourses. Rather than assuming that genetic knowledge alters individuals' subjectivity and construction of family, it is probably more useful to think that the opposite occurs, that is, individual and familial relations alter the way genetic knowledge and diseases are experienced.

### **Families and Uncertainty**

Another important theme for my thesis is the uncertainty related to genetic information and its implications for patients and families as well as healthcare providers. As the vast majority of the studies analysed have shown, there is always a certain degree of uncertainty associated with genetic diagnoses and risk. Medical genetics often results in probabilistic outcomes which can leave patients uncertain. It is very difficult to predict gene-gene, gene-environment interactions, hence there is uncertainty with regard to

phenotypic expression both in cases of complex multigenic disorders or predispositions and in cases of monogenic disorders with clearer prognostic pathways (Kitcher 2003). NF1 is a valuable case study through which to examine the effect of uncertainty in experiences and understandings of genetic information, as it is variable in different ways (e.g. from café-au-lait spots to life threatening tumours, and it can cause both cognitive and physical difficulties); recalling Ablon, NF1 is a ‘condition without parameters’ (Ablon 1999).

As I discussed above, empirical research has highlighted that patients attempt to make sense of clinical risk estimation by translating numeric values into something that is more meaningful to them (Parsons and Atkinson 1992). Moreover, family members with a genetic condition can be used for example as tools of inspection and benchmarks to visualize possible future trajectories of the disorder (Cox and Starzomski 2004).

Through the interpretative lens of Finkler’s medicalization of the family (Finkler 2001), the increasing prominence and significance of biomedicine, with its inherent uncertainty, is introducing a new sense of risk and anxiety, changing individual and familial experience of disease and resulting in new constructions of personhood.

According to Finkler, families struggle with the unknown in diagnoses, experiencing feelings of constant worry about their future. It can be argued that these feelings of uncertainty and anxiety can become the strings that unify geographically or affectively distant family members who are sharing the same genes and destiny.

For Finkler, individuals are becoming *perpetual patients*, i.e. because of their worry towards the future possibility of developing a certain disorder, they interiorise the role of patient into their present life (Finkler, Skrzynia et al. 2003). People who believe themselves to be, or are actually found, to be at risk for a genetic condition react by altering their status of healthy individuals, becoming patients before it is necessary.

From being an uncertain possibility in the future, risk is brought into the very essence of their personhood, consequently becoming a constant worry in individuals' lifeworld. Hence, in Finkler's view, genetic medicine forces risk and bonds of consanguinity into people's lives. Individuals tentatively address risk and its inherent uncertainty by entering, when possible, into the medicalized domain to seek clearer prognoses, unequivocal labels and in general to find more stability and certainty.

Nevertheless, it is important to observe that the dimension of uncertainty remains often uncontrollable and unmanageable within the biomedical domain too. Individuals' requests to healthcare professionals for clearer diagnostic or prognostic scenarios often remain unanswered (Featherstone, Atkinson et al. 2006; Whitmarsh, Davis et al. 2007). Moreover, other research shows that patients can also emphasise and value the uncertainty inherent to genetics, by transforming it into a site of resistance to biomedical definition of disease and risk. An interesting example can be found in Whitmarsh and colleagues' study which examines how families with Klinefelter, Turner and Fragile X Syndrome make sense of the uncertainty related to these disorders (Whitmarsh, Davis et al. 2007). The research is based on semi-structured interviews with parents and relatives of children with these conditions and focuses on their understanding of the syndrome and the significance given to the diagnosis. Klinefelter, Turner and Fragile X Syndrome are variable conditions associated with a wide spectrum of cognitive and physical complications which are difficult to predict. All these disorders can be diagnosed also in the absence of clinical manifestations, simply relying on genetic testing.

Participants' accounts revealed a tension between the certainty of the diagnosis and the uncertainty of its prognosis. Especially around the time of the diagnosis, they tended to

assign importance to the genetic and clinical aspects of the syndromes. Yet, the medical authority of the diagnosis clashed with the lack of definition and uncertainty related to the phenotypical expression of the syndrome. Over time, the experience of uncertainty actually distanced many families from the biomedical definition of these syndromes. Participants drew on their experience of raising children with these syndromes to question and resist medical diagnoses. Further they used the experience of uncertainty to express their frustrations towards the medical understanding of disease and to free their children's individuality and potentialities from the constraints of medical diagnoses and the clinical descriptions of the disorders. They highlighted, for example, the accomplishments of their children and other positive aspects related to the syndromes that were not included in its medical description. They saw their children as having an open ended future that could comprise possibilities that were beyond the limits imposed by the diagnosis. Some families challenged the diagnosis itself, especially when the children did not present any symptoms.

To sum up, many families actively embraced uncertainty to resist biomedical labels and to challenge or even question the value of genetic labels. As parents of children diagnosed with these syndromes and dealing with the uncertainty in everyday family life, participants gained confidence to question the authority of healthcare professionals, negotiating with them the nature and implications of the diagnoses (Whitmarsh, Davis et al. 2007).

Therefore, individuals can relate to the unknown in genetic diagnosis not only, as Finkler suggested, by principally feeling a constant worry about the future and/or seeking medical help. These studies equally challenge theoretical constructs like those of Kerr, Shakespeare and Lippman which envisage passive patients subjected to

biomedical authority. I would argue that the agency shown by these participants is more in line with Novas and Rose idea of the active (bio)citizen who as patient and consumer demands control over their condition from health providers and is also ready to challenge medical authority if their demands are not met by the healthcare system. Nevertheless, the uncertainty of genetic conditions and the effect it can have on their clinical management and on individual and family responses to genetic information, can be seen as factors that mitigate theoretical constructs like Rose and Novas' which presuppose a profound change of subjectivity in the light of genetic information. Uncertainty can in fact become a factor that undermines the authority and salience of genetic information for individuals' illness experience.

These aspects are particularly interesting in the light of my thesis which studies a highly unpredictable condition like NF1. As discussed in the previous chapter, Ablon suggested that the uncertainty of NF1 (that according to her is *a condition without parameters*) was the most distressing psychological feature of the disorder for patients and families. However as I already observed, Ablon did not extensively focus on the genetic aspect of the experience with the condition. With my research I will explore the interplay between genetic knowledge and uncertainty in individual and familial experiences with this condition.

### **Practical Kinship**

The complexity and situatedness of individual and familial dynamics around genetic risk emerged from the empirical studies discussed above is nicely captured and synthesised in the work of Featherstone and colleagues (Featherstone, Atkinson et al. 2006). For these authors, research on the effects of genetic medicine on people's lives cannot disregard the importance of systematic explorations of how individuals make sense of inheritance and risk within their familial relations. Thus, alongside being an

essay on the impact of genetic medicine on individuals and families, this study is an extensive investigation of the social meaning of relatedness and how it is enacted in the everyday lives of family relations.

Throughout their work, Featherstone and colleagues compellingly argue that the relations of inheritance and kinship are constructed within cultural and social contexts which are independent from biomedical knowledge. Hence, for the authors, although medical genetics may strengthen biological relatedness, individuals' constructions of inheritance and kinship are not straightforwardly medicalized - as suggested for instance by Finkler - but are mediated by social and other contextual factors.

Drawing on anthropological and sociological literature, Featherstone and colleagues highlight how the 'natural' bonds of consanguinity are themselves as cultural and social categories. In the first place, historically, knowledge of diseases occurring in the family as well as mutual surveillance is not novel, but predates the advances of molecular genetics and medicine. Moreover, Featherstone and colleagues mention, for example, reported cases in anthropological studies of matrilineal kinship systems in which individuals are closer to the male siblings of their mothers, rather than to their biological fathers. Hence, the same biological ties can be construed in different ways depending on cultural, social and local contexts.

Following from this, Featherstone and colleagues argue that it is misleading to assume that individuals' everyday experience of kinship is strongly shaped by, or even coincides with, its biomedical representation. Therefore, contrary to the position held by commentators like Finkler, they claim that biomedicine and genetic technologies are not strongly reshaping people's understanding of kinship and inheritance. Family bonds defy biomedical classifications; rather than being merely naturally determined entities,

they also vary upon the particular interactions and the processes of mutual recognition among individuals.

To capture this irreducible complexity and agency of families and individuals, Featherstone and colleagues coin the notion of ‘practical kinship’ (Featherstone, Atkinson et al. 2006). Families are the result of everyday interactions and practical acts of mutual recognition between members, like the exchange of favours, mutual obligations, everyday contacts, the celebration of festivities and other forms of gathering. Individuals’ understandings of genetic risk and diagnoses are intertwined with this gamut of past and present interactions that constitute families. The way patients make and remake their family bonds and the value they give to inheritance are contingent and can vary considerably with their lifecourse. Moreover, the boundaries of the family and the salience of familial relations often vary across different family members. In other words, each individual can potentially assign different values to family interactions and bonds. Once again, this is indicative of the fluid nature of family relations which, in turn, can influence the diverse idiosyncratic responses to genetic medicine deployed by individuals.

Featherstone and colleagues’ research is grounded on in-depth interviews with families with a range of genetic conditions who have attended genetic clinics in South Wales, and on ethnographic work based on observation of the work of different clinical teams in these genetic clinics. Many of these genetic disorders were variable and unpredictable both within and between families and over the lifetime of affected individuals.

Therefore, often the diagnosis of these conditions was not necessarily accompanied by clear prognoses, assessment of severity, age of onset and effective treatment pathways (Featherstone, Atkinson et al. 2006). The analysis of their fieldwork provides important



insights into how biomedical understanding of disease is strongly mediated by familial dynamics.

Many family members interviewed appeared to be involved in practices of mutual surveillance. Younger generations were often inspected by older ones to detect new possible occurrences of the genetic condition within the kindred. However, these surveillance patterns conflated biomedical definitions of inheritance with elements of practical kinship. All the offspring, for instance, could be inspected, including those who were found by clinicians not to be at risk of inheriting the disorder. As with studies discussed above (Cox and McKellin 1999), observing the impact that the diagnosis and the management of genetic conditions had on the lives of affected family members, could also significantly influence individuals' personal decisions about seeking diagnosis and treatment. On the basis of the illness experience of other family members, some participants refused to seek a diagnosis for themselves or to follow clinicians' recommendations to undergo regular monitoring, even when complying with medical monitoring procedures could be beneficial to their health.

Disclosure appeared to be a complex and problematic process. Families were not 'aseptic' environments to be smoothly filled with genetic information. On the contrary, they encapsulated pre-existing relations, bonds and biographical elements which influenced the processes of disclosure and management of genetic disorders (Arribas-Ayllon, Featherstone et al. 2011). Individuals often did not communicate the news of having a genetic disorder to all the members of their kindred, but only to those they regarded as being part of the family and/or those whom they judged to be capable of understanding and properly coping with this information. Therefore, the flow of

communication did not necessarily follow the lines of biomedical inheritance and clinical recommendations, but was strongly mediated by instances of practical kinship.

Furthermore, communication often was a diffuse and partial process. Disclosure was diluted throughout the everyday life of practical kinship (Metcalf, Coad et al. 2008). When discussing genetic disorders, individuals, for instance, tended to omit for as long as possible certain pieces of information that could be distressing for other family members. Even close family members tended to avoid having conversations about their genetic disorders because this was considered to be a delicate topic that could generate anxiety, a sense of guilt or other distressing feelings and revive problematic family dynamics. This phenomenon has also been called ‘family protectiveness’ (Skirton 2001). Moreover, participants tended to interpret- and then explain to other family members – the information about their genetic disorders in the light of their personal experience and beliefs. Even when individuals had had contact with clinicians and genetic counsellors, their beliefs about their genetic condition and its patterns of inheritance could be said to be inconsistent with medical knowledge. Similar results have been reported in other studies. A longitudinal research on patients’ experiences of living with presymptomatic genetic test results of HBOC highlighted, for example, that health behaviours and disclosure to children and other family members represented common challenges and concerns that genetic testing could pose to family life and relationship (Hamilton, Williams et al. 2009).

Featherstone and colleagues’ research represents some of the most detailed and comprehensive piece of work available on the complexity of biomedicine and of the life world of families who live with genetic disorders. The notion of practical kinship nicely encapsulates this complexity, providing an open and useful framework that allows an

investigation into the social impact of genetic medicine, without losing contact with the experiences and needs of patients and families. Contrary to ideas of medicalization according to Finkler, which adopt abstract, stereotypical models of the family characterised by simplified familial ties, the notion of ‘practical kinship’, implies that familial ties are also the result of everyday recognition and interaction between family members (Featherstone, Atkinson et al. 2006; Metcalfe, Coad et al. 2008). Individual and familial reactions to genetic conditions cannot be simply reduced to, and understood in terms of, medical definitions of disease and inheritance.

In relation to my thesis, it is also worth emphasising that there is an important link between the notions of practical kinship and lifecourse fluctuating relevance of genetic knowledge. It can be claimed that the experience of disease and risk conflates individual and familial biographical factors, mundane and other contingent elements alongside biomedical, governmental and other broad social discourses. Family dynamics are important factors that shape and influence individuals’ fluctuating relationship with genetic knowledge. Complex familial dynamics can underlie the lifecourse fluctuating relevance of risk in individuals’ lives. Thus the complexity of family dynamics is reflected in the complex and variable individual response to genetic knowledge.

In the same way as Featherstone and colleagues work, with my thesis I intend to examine the interrelation of individual and familial experience of disease and genetic knowledge, focusing in particular on individual and familial meaning making practices around uncertainty.

### 3.4 Concluding Remarks

In summary, widespread sociological accounts of the impact of biomedicine on individuals like biological citizenship and the medicalization of the family assume:

- A clear genetic underpinning of diseases.
- A strong uptake of genetic practices in medicine (as diagnostic and management tools)
- A profound change in individual subjectivity resulting from the individual and familial uptake of genetic practices in everyday lives, promoted by government and public health discourses (genetic practices have been presented both as empowering and as forms of subjectification).
- A strengthening (or medicalization) of family bonds due to the familial implications of genetic practices.
- An increasing role of support groups as part of individuals' and families' engagement with their genetic disorders (novel forms of social political biomedical assemblages).

Contrary to these theories, more empirically grounded research has highlighted that:

- Understanding the mechanisms of disease is an enormous task. Genetic risk and conditions are diversified, multifarious and linked with uncertainty.
- Genetic practices are not necessarily adopted in clinical settings.
- Individual and familial responses to genetic medicine are heterogeneous and situated: the medical understanding of genetic risk and disorder does not necessarily coincide with the 'lay' understanding. To date, the responses of individuals with genetic disorders appear mixed, including the ones of people who come from families with rare, deadly single gene disorders. Individuals can ignore, downplay or challenge test results or their conditions; the relevance of risk is not static, but can vary throughout the lifecourse of individuals, reaching zones of high relevance that are often related to familial dynamics (for example reproductive choices).

- Family dynamics do shape how genetic conditions are lived. However not only the experience of risk, but also the experience of kinship does not necessarily coincide with its medical representation. Families are irreducibly complex. Familial ties, rather than being just naturally determined entities, are also the result of everyday recognition and interaction- between family members.
- The role of support groups in individuals and familial experiences of genetic diseases is complex.

It seems unclear whether genetic medicine is profoundly changing individuals' subjectivity and their relation to health and disease. What seems clear from this review is that individuals' construction of genetic disease and the psychosocial consequences of genetic practices defy sweeping generalizations about changes in subjectivity. Such generalizations - like for example the notion of biological citizenship - seem also to mirror the general hype associated with genetic and genomics that pervades a lot of governmental and media discourse (Lock and Nguyen 2010).

Moreover, alongside overlooking important aspects of the lifeworld of patients and family, these theories can also directly or indirectly contribute to deflecting attention from potential problems and inequalities related to biomedicine. It seems unclear whether broad theoretical constructs like biocitizenship and biosociality are conceived to be descriptive or normative. Nevertheless, independently from the intent of those coining such expressions, I would argue that these constructs have normative repercussions. The idea of patient empowerment and autonomy and the consequent climate of duties and expectations described/fostered by biocitizenship - have the potential to mislead policy making. The claims of biocitizenship - which already strongly resonate with governmental and general public discourse over genetic medicine - may in part lead healthcare professionals and policy makers into thinking that patients

and families with different genetic conditions significantly engage with genetic knowledge, support groups and are empowered by this. Even if they are not adequately safeguarded by the healthcare system and find medical and psychological help from genetic support groups, web forum and other source. But, in the light of the studies discussed above this may not always be the case. Therefore, in line with the more critical stance of Petersen and Bunton towards the social impact of genetic practices, it is plausible to argue that models like biosociality and biocitizenship may divert attention from, or even cover up, situations in which patients do not or cannot engage with their genetic disorders and bring forward their health-related needs. Furthermore, it can be argued that notions like biosociality and biocitizenship can potentially contribute to the social pressure patients may experience in case they do not or cannot conform to the disease activism and agency model these notions ultimately convey.

As I stated in the introduction of this chapter, studying individuals' experiences with genetic disease can provide healthcare professionals with valuable information. Hence, it is important that more empirical detailed studies are conducted with a greater variety of conditions in order to delineate a more nuanced and encompassing picture about the social impact of genetic medicine.

The same line of argument that has been used to highlight the limitations faced by theorizations of individuals' response to genetic risk can be applied with respect to family and kinship. It is reasonable to claim that genetic medicine can strengthen ties of biological relatedness. Family dynamics and bonds do shape how genetic conditions are lived. Nevertheless, not only the experience of risk, but also that of kinship does not necessarily coincide with its medical representation.

This outline of research on the experiences of individuals and family with genetic conditions suggests that genetic knowledge, rather than having an increasingly

hegemonic role in determining health-illness identity and behaviours, is just part of a more complex set of identity discourses and practices. Individuals and families are not empty boxes that are simply filled with biomedical information. Rather, they receive and interpret genetic knowledge in the light of their biographies which can change and adapt over time.

As argued by Featherstone and colleagues, simplistic models, like Finkler's, that overlook the complexity of families and emphasise traditional familial bonds, can actually exacerbate the distress experienced by individuals, who may feel their family does not fit this ideal image (Featherstone, Atkinson et al. 2006). In other words, the narrowing of families to genetic bonds has additional normative implications.

Families conflate biological, cultural and social elements. Their status and structure are flexible and unpredictable: divorces, adoptions, reproductive technologies, or simply absent or problematic family relations are common in our society. Research that conveys an idyllic and traditional image of families where disclosure and family bonds are unproblematic, may contribute in generating a climate of misleading expectations on how families should deal with genetic disorders. Explorations of the impact of genetic medicine on families should move beyond the stereotype of the traditional nuclear family and respect the practical and empirical aspects of family life. This type of research may produce results that could benefit families which are living with genetic disorders (Skirton 2001; Skirton, Williams et al. 2010).

Therefore, there is a pressing need for more empirical investigation into how families interpret and manage genetic risk and diagnoses. Investigating individual and familial experiences with genetic disorders is difficult (for example it is not always easy to gain access to families given the complexity of their structure and relations) and produces

thick (complex) results (Cox and McKellin 1999; Featherstone, Atkinson et al. 2006). Nevertheless, having greater insight into the dynamics and processes by which families face genetic conditions, genetic futures, and healthcare can enrich research on clinical settings, shedding more light on the needs of individuals with genetic conditions.

### **3.5 Rationale for my Research**

Theoretical constructs like genetic responsibility, biocitizenship, biosociality, medicalization of the family - whilst acknowledging the complexity of genetic information- rest nonetheless on the assumption of a link between genetic information and a sense of certainty and identity. Genetic testing or diagnosis is viewed as determining whether a patient has or will develop a disorder and thereby leading to a significant improvement of prevention, treatment and intervention capacities.

However, I have shown that the application of genetic technologies in medicine generates also uncertainty. Genetic tests can reveal if somebody is at risk of developing a particular disorder, but the tests are based on probabilistic estimations which can be complex for healthcare professionals and patients to interpret. Genetic diagnoses do not always coincide with clear prognoses; conditions which are characterised as being partly or wholly dependent on genetic factors can still be variable and unpredictable.

Predicting genes-genes, genes-environment interactions can be an enormous task, both in cases of complex polygenic disorders or predispositions and in cases of monogenic disorders with clearer prognostic pathways. Moreover notions like genetic responsibility, biocitizenship, biosociality, medicalization of the family are often associated with HD, or HBOC, which are condition of a less complex and variable than multisystem genetic syndromes. Sociological research based on a variety of different conditions (Cox and Starzomski 2004; Petersen 2006; Bharadway, Atkinson et al. 2007) - including also HD (Cox and McKellin 1999) and HBOC (Hallowell 1999; Hallowell,



Arden-Jones et al. 2005; Hallowell, Arden-Jones et al. 2006) - showed a more heterogeneous and nuanced picture. Patients and healthcare systems responses to the increment of genetic practices in medicine are significantly heterogeneous and situated.

With this thesis I focus on NF1 to examine whether having a genetic basis makes this complex syndrome more coherent as concerns individual subjecthood, family experience and the way it is managed by the healthcare system. It is theoretically and clinically important to investigate whether and how NF1 is associated with a strong and specific identity and community based on genetic knowledge, that is, whether the notions of biosociality and biocitizenship are relevant for NF1. The constructions and management of diseases may vary in relation to different genetic conditions and the features of these conditions such as variability and symptomatology. A transformation of patients' subjecthood in genetic terms may be less evident in the case of multisystem, highly variable and unpredictable genetic disorders like NF1 which may be managed, alongside clinical genetics, by other different medical specialties.

As NF1 is very variable, patients and healthcare providers may emphasise clinical aspects of the syndrome other than the genetic ones. Moreover, the multisystem nature of NF1 and the fact that its management may involve diverse medical specialties may pose considerable challenges to the healthcare system. Thus, the variability and unpredictability of NF1 can make it a very difficult condition for patients and families to deal with and for healthcare professionals to manage.

Given the relational and familial implication of genetic information, I will also focus on genetic responsibility as a familial and relational experience. This approach will offer a greater insight into the complex entanglement of patients and family dynamics (i.e. practical kinship) around genetics, illness and identity. I will examine whether and how affected individuals are placed in a web of relations of responsibility to family

members, other people and themselves and whether they make life plans such as decisions about getting married, having children and a career in relation to NF1. Furthermore, with particular reference to uncertainty, the literature analysed has highlighted that individuals with genetic conditions may be inspected by other family members as a way of predicting the development of their own disorder or, more in general, may influence other family members' modality of engagement and disengagement with a genetic disorder. However, at least in theory, the case of NF1 problematizes this scenario as the syndrome can vary considerably within the same families and over the life time of affected individuals. Exploring the clinical management of and patients and families sense making around uncertainty and genetic knowledge appears to be very challenging for complex highly variable conditions such as NF1.

### **3.6 Research Aims**

The objective of this thesis is to study the impact of genetic knowledge on experiences and understandings of NF1 with respect to individuals with the syndrome and their family. The research aims to investigate:

- (1) The relevance of the notions of genetic responsibility, medicalization of the family, biocitizenship and biosociality to NF1, that is, the ways in which knowledge about the genetic basis of NF1 influences individual and familial experiences with the syndrome, the definition of the self, the management of the condition and the use of structural support such as support groups.
- (2) The interrelation of individual and familial experiences of disease, particularly individual and familial meaning making practices around uncertainty.
- (3) How patients and families are managed by the healthcare system, how they experience it and how genetic classifications play (or do not play) into this.

## **4. Research Method and Process**

### **4.1 Introduction**

I have addressed the research aims formulated at the end of the previous chapter by conducting qualitative, semi-structured interviews with individuals with NF1, their family networks, and healthcare providers involved in the management and treatment of the syndrome. The following chapter aims to explain this research method and outline the research procedure. It is divided into two sections.

Section 4.2 will discuss the methodological approach, the relevance of semi-structured interviews for this research project and it will provide a description of how the interviews have been analysed. In particular, this section will also account for the theoretical and methodological importance of exploring family networks' experiences with genetic disorders.

Section 4.3 will describe the specific research design and procedure adopted in this thesis.

### **4.2 Research Method**

#### **Qualitative Methods**

Qualitative methods are indicated to explore areas of research which are novel or not well understood, where the relevant variables have not yet been fully identified (Glaser and Strauss 1967; Strauss and Corbin 1990; McAllister 2001) and specifically in research which focuses on lived experiences and attitudes (Silverman 2005).

Differently from quantitative methodologies, qualitative methods do not allow the investigation of general laws, causal relationship between different phenomena and to make claims about trends and regularities in a particular population.

The main strength of qualitative research lies in its ability to explore and analyse how a phenomenon is locally constructed. It investigates people's views and behaviours in their natural context (Silverman 2006), rather than through categories defined by the researcher at the outset.

Therefore, this is a relatively flexible methodology in terms of data collection and analysis. The qualitative research process tends to be significantly iterative i.e. aims, theoretical framing, data collection and analysis can mutually inform and enrich each other during the lifecycle of the research. Consequently, qualitative research often generates novel insights and understandings, offering a more nuanced and fluid perspective on issues that are normally taken for granted. By allowing for an in depth analysis of participants' perspectives, qualitative methodologies can provide revealing and interesting examples of contextual sensitivity (Silverman 2006) i.e. an apparently stable phenomenon and institution (e.g. a medical diagnosis or the family) may acquire different meanings in different contexts.

Qualitative research gives considerable space to participants' voices and provides rich descriptions of people's experiences and subjective meanings. This methodology enables the researcher to engage with participants on their own terms and to sensitise to participants' diverse forms of expression. It allows to study how a certain phenomenon is locally constructed and to investigate the broader social context in which participants responses are elicited.

For its flexibility, holistic approach and closeness to people contexts and meanings, this methodology has been increasingly adopted in healthcare service and sociological research to ascertain the needs and perspectives of patients (Skirton 2001; Bowling 2004) and to explore the lived experiences of disease.

All these features make qualitative methods particularly suitable for this doctoral thesis which explores NF1 families' experiences and needs and healthcare professionals view's on the management of this highly variable syndrome.

As this study of living with NF1 is relatively novel and explores delicate and complex issues (e.g. disease, genetics, uncertainty, family), it is important to employ a methodological approach that enables respondents to express themselves as freely as possible and to raise issues that are relevant to them in their own languages and expressions. Moreover, qualitative methodologies allow the contextualization of patients' accounts of disease, genetics and uncertainty in the light of familial dynamics, the healthcare system and broader social environment. Therefore, a qualitative research approach is very useful to explore in-depth the possible interrelation of individual and familial experiences of disease and meaning making practices around genetic knowledge and uncertainty.

This qualitative research combines both Grounded Theory (GT) and Narrative Analysis (NA) inspired methods to analyse and interpret the data.

### **Grounded Theory**

GT is a qualitative research method articulated by Glaser and Strauss which has been commonly utilised in social sciences (Glaser and Strauss 1967). It consists of inductively building theory from the collected and analysed data in a way in which data collection, analysis and eventual theory stand in close relationship to one another (McAllister 2001). The strength of this method resides precisely on the fact that it is designed to build theory about psychosocial processes from a strong evidence base, rather than through prior hypotheses (McAllister 2001; Silverman 2006).

The researcher employing this methodology is expected to produce a clear description not only of the results, but also of the process of data analysis in order to make the whole study more accessible and potentially replicable.

Thus, GT is presented as being very versatile and ‘permeable’ to the richness of qualitative research data, and at the same time as being a rigorous method of building up hypotheses and theory from the data collected (Strauss and Corbin 1990; Strauss and Corbin 1998).

GT has been adopted by diverse disciplines in a variety of ways and is therefore rather heterogeneous (Charmaz 2000a; Silverman 2005; Charmaz 2006). Following in the wake of Glaser and Strauss initial articulation (1967) and other scholars’ accounts (e.g. Charmaz 2006), I would argue that GT could be seen as a flexible set of tools that can be adopted by the researcher as and when appropriate.

I did not follow the procedures of GT rigidly, also because there are many and sometimes divergent version of this research method.

For example, I did not follow an important tenet of early GT that the researcher should engage with the review of the literature only after data collection and not before or during. The rationale behind this strategy is that doing literature review can influence the researcher to draw on pre-existing findings while analysing a phenomenon, potentially missing the richness and originality of the data collected (Strauss and Corbin 1990).

However, this strategy presumes that the researcher is a *tabula rasa* without preconceived ideas and knowledge developed from experience and literatures already read. In more practical terms, this strategy is at odds with how PhD and other research are conducted. Normally, in order to start a project, the researcher is expected to produce a proposal which is informed by a review of the relevant literature on the topic of interest (Friese 2007). In my own experience, I believe the reading of the literature

(particularly the one on NF1) made me more capable of understanding and tune with the accounts provided by the interviewees.

The GT analytical process followed by this thesis has been mainly inspired by Strauss and Corbin (1990; 1998).

Data analysis mainly occurs through two processes: coding and memoing.

Data are closely read and carefully coded line by line (also word by word when necessary) in order to isolate processes and actions. Often, participants' own words are used to name codes and describe concepts in order to maximise the emergence of theory from data (Silverman 2005; Corcoran, Mewse et al. 2007). Codes are successively refined; more general analytical categories are derived from codes via an analytical process which is based on systematically comparing, making connections and establishing causal relationships between codes (axial coding). As the analysis progresses, the researcher starts to focus on some categories which appear to be particularly relevant, to relate them to other categories and to fill categories which require further refinement and development (selective coding) (Strauss and Corbin 1990; Charmaz 2006).

Categories can also be further checked and clarified through theoretical sampling i.e. the sample and sources are not necessarily fixed at the outset of the research, but can include successive sites if new leads emerge from the analysis of the initial interviews (Glaser and Strauss 1967; Strauss and Corbin 1990; Silverman 2006).

The analytical process is recursive i.e. these different types of coding do not occur in a linear fashion. Codes and categories derived from the analysis of the initial interviews are tested against subsequent interview data. Moreover, interviews schedules, sampling, coding, theoretical underpinning of research are amended and refined in relation to data collection and analysis. This important aspect of GT is also referred to as the *constant comparative method* (Strauss and Corbin 1990; Charmaz 2006). Therefore, the



researcher who employs GT methods is significantly guided during the lifecycle of the research by the data collected.

The analytic process is also accompanied by the writing of memos. Memos are written notes which allow the researcher to elaborate thoughts and reflect on the analytic process. Memos are normally used to reflect on data, codes and to relate the analytic process to the literature (Charmaz 2006). Similar to memos, another important principle that underlies GT is researcher's reflexivity, which stands for the idea that the researchers should produce critical accounts of their engagement with the research and fieldwork and their role in the research, including acknowledging potential researcher bias (Strauss and Corbin 1990; Bowling 2004; Silverman 2005).

GT appears to be suitable for this thesis which intends to investigate an exploratory area focusing on the terms and concepts participants use themselves, rather than presupposing them.

GT methods have been increasingly used in genetic counselling research as they provide an effective means of developing evidence-based theoretical frameworks about psychosocial issues around genetic counselling (Skirton 2001). GT research can be relevant to counsellors and other genetic service providers and can produce a basis for clinical intervention (McAllister 2001). This type of research has explored patients' perspectives and needs and has also focused on how genetic information is interpreted and understood both during and after clinical appointments (McAllister, Payne et al 2008).

GT has also been used in sociological studies to analyse illness experiences (see for example Charmaz 2000; 2006). It is important to remember that Glaser and Strauss GT approach was first developed in relation to their study of dying on an hospital ward (Glaser and Strauss 1967), thus in a similar research area. GT methods of analysis

appear to be a particularly appropriate framework for this qualitative research, and for the analysis of data.

GT has been a useful resource that has guided me in thinking about the issues arising from patients and health care professionals experiences of NF1.

However, in order to explore more in depth and unravel the interrelation of individual and familial experiences of NF1, I have also employed another analytical tool: Narrative Analysis (NA).

### **Narrative Analysis**

Most scholars regard narratives as a means through which individuals make sense of experiences and communicate their meanings to others (Viney and Bousfield 1991; Chase 1995; Bowling 2004; Czarniawska 2004). Through narratives, individuals interpret and elaborate experiences by relating them to their own biography and by drawing on broader social cultural models, traditions or motifs, the so-called ‘narrative stocks’ (Murray 1999). These stories or narratives exert and mirror several psychosocial and heuristic functions; from helping to develop and maintain a sense of identity to providing a guide by which to live and to cope with events or problems (Viney and Bousfield 1991). Narratives incorporate cultural conventions, recurring structures and specific genres (Frank 1995; Atkinson and Silverman 1997; Thomas 2010) and are viewed as important creations which enclose in their very content and structure, the dialectic between the individual and the socio-cultural spheres.

Narrative analysis approaches have been increasingly adopted in health research as a means to access patient illness<sup>9</sup> experiences. This method has been particularly

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<sup>9</sup> Consistent with sociological literature on health and illness (Kleinman 1988 ) I use the term disease to refer to the biological disorder diagnosed by healthcare professionals whereas I use the term illness to refer to the person’s experience.

employed to study patients' construction of lifelong conditions i.e. chronic diseases (Charmaz 1983; Kleinman 1988; Charmaz 1991; Bury 2001).

Narrative studies have contributed to the biomedical understanding of disease, giving more visibility and importance to patients' voices and their experiences with disease in their everyday life (Mishler 1986; Kleinman 1988).

A narrative approach is particularly fruitful to this thesis project, as it allows for a nuanced appreciation of how genetic knowledge and risk are understood in relation to individuals and families' lifecourse (see Chapter 3). If part of the narrative process consists in making sense of events by relating them to personal biographical aspects, then exploring illness narratives can provide a valuable access into the richness of the meanings individuals assign to genetic information and the way these meanings are merged with - and vary throughout - their lives. For these reasons, narrative analysis can offer a fine grained insight into the interrelation of biographical elements in individuals' experience with NF1 and genetic knowledge.

NA is also complementary to the GT analysis. Systematic coding is a very effective way to manage a considerable amount of data and allows the researcher to easily make links, trace patterns and group the different interviews.

However, this analytic technique fragments the data and does not allow the researcher to retain the 'whole' i.e. a sense of continuity and contradiction through any of the individual accounts. In this way, important aspects that should be considered in the process of theoretical analysis and interpretation of data, such as the complexity of the research subjects and their unique individualities can be lost. In fact, the same (de-contextualised) code can easily normalise and obscure the heterogeneity of meanings (Braun and Clarke 2006). The first immediate expedient to prevent this is the

employment of inclusive coding (Strauss and Corbin 1990), instead of the more 'canonical' sentence-by-sentence or even word-by-word coding strategies (Browner and Preloran 2000; Smith and Sparkes 2008; Kelly 2009). Inclusive coding implies generating codes that incorporate as much transcript as possible, as long as it is meaningful.

NA allows the researcher to go beyond inclusive coding. The concept of the story integrates and brings together, rather than separates, elements of human experience (Bowling 2004; Silverman 2005), allowing the researcher to get closer to participants and their picture of events. Consequently, by identifying narratives within the whole interview and the whole interview as a narrative, NA helps to capture something more of the complexity of the research subjects, since it allows for a holistic interpretation of data, informed by biography and the individuality of participants. For these reasons the narrative approach will also offer an excellent point of access to unravel the interrelation of individual and familial experiences with genetic disorders.

### **Semi-Structured Interviews**

Qualitative semi-structured interviews represent a flexible and explorative method which gives to the researcher the possibility of pursuing salient issues which arise during the field-work period, rather than having to know in advance all the important aspects of the area being researched (Kelly 2009).

Semi-structured interviews are guided by a set of questions, or schedule, formulated in advance by the researcher. Normally, the interview schedule is relatively fixed to make sure that the same core research topics are covered consistently in all the interviews, making the comparison between interviewees easier.<sup>10</sup> Nonetheless, although prompted

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<sup>10</sup> However, it is also possible to argue that qualitative interviews assume shared meanings i.e. a transparency of language. There is in fact an implicit assumption that the words (in the question) mean the same thing for the interviewer and interviewee (Hollway and Jefferson 1997).

by a schedule, semi-structured interviews allow for a significant level of flexibility. This interviewing method gives to the researcher the freedom to be attentive towards the language of the interviewees and to follow their narratives and directions. The flexibility of semi-structured interviews is highly suited to the exploration of a novel area of research, as in the case of this dissertation.

Interviews are particularly appropriate to explore the meanings that participants give to a set of events, experiences and broad phenomena investigated by the researcher (Bowling 2004). They allow the researcher to go beyond superficial responses and to obtain from the interviewees detailed and ‘thick’ accounts, that is, valuable insights on the meanings that they assign to events, attitudes, behaviours and experiences (McAllister, Payne et al. 2007; Kelly 2009). The data gathered from interviews also gives the researcher the possibility to analyse in depth the language and communication styles used by the interviewees. Furthermore - compared with other common qualitative research methods (e.g. focus groups) - interviews are more prone to encouraging introspection. This is mainly due to their confidential setting. For these reasons, qualitative interviews are generally used to explore private issues, emotions, experiences, thoughts, intentions and the meanings individuals attach to events. Therefore, this method can be fruitfully employed to discuss sensitive issues, for example- as concerns this thesis- self-perception of NF1 individuals, familial relations and constructions of NF1, attitudes towards genetic knowledge.

In summary, qualitative semi-structured interviews are suitable for this thesis project since, as stated in the previous chapters, (i) the role of genetic knowledge as in patients’ and families’ experience have not been investigated in great detail yet in relation to NF1 and (ii) the exploration of experiences and attitudes is essential to this dissertation project. This interview method has been adopted because of its potential to provide rich data on the lived experiences of NF1, especially with reference to complex issues

related to genetic knowledge, their impact on health behaviours and illness identity (Saukko 2003).

In the literature the status of data gathered by interviews is highly contested. The interview is regarded as a form of social interaction in which the researcher, the research and research subject all occupy social positions (Silverman 2006). The researcher never truly disappears and does not simply act as a conduit for interviewees' experiences (Hammersley and Atkinson 1994; Bowling 2004). On the contrary, in interviews there is potential for interviewer bias (Voysey 1975; Baruch 1981; Cornwell 1984); i.e. power relations, or some other forms of influence may explicitly or implicitly occur between the interviewer and the interviewee. Interviewees' discourses can conflate 'public', or 'politically correct' accounts, and 'private' accounts (Silverman 2005; Silverman 2006); this may be seen as diminishing the value of the data.

What participants say in an interview depends also on who they think they are talking to and in what situation. Interviewees can be motivated, whether consciously or unconsciously, to disguise the meaning of at least some of their feelings and behaviours. Interviewees can embrace particular positions to protect vulnerable aspects of the self or of others, they may not know why they experience or feel events in the way that they do or may even not hear the question through the same meaning-frame as that of the interviewer or other interviewees (Hollway and Jefferson 2000).

Therefore, the data collected by qualitative interviews may present idealizations (Hollway and Jefferson 1997), abstract intellectualisations (Dingwall 1997; Murphy, Dingwall et al. 1998), impression management, a dance of expectations (Baruch 1981; Silverman 2006), moral tales or tales of personhood (Hollway and Jefferson 2000). Not only are the meanings of a person not unique, but they also depend on the relational encounter with the interviewer (Cornwell 1984).

Nevertheless, the more the researcher manages to adopt measures to increase disclosure and comfort in the interviewee, the more likely it is that the level of communication reaches some 'depth', allowing the interviewee to reveal inner perceptions, feelings and behaviours, opening the space for 'private' rather than 'public' accounts (Hollway and Jefferson 2000). The logistic of the interview can play a decisive role; it is important that interviews take place in a setting that will be comfortable for the interviewee (e.g. interviewees' homes or any other place chosen by them as opposed to more 'authoritative' and 'suppressant' settings such as hospitals or work environment). Emphatic and attentive listening on the researcher's side can also be determinant in facilitating disclosure.

Furthermore, the conflation of individual (private) and social elements do not necessarily skew and devalue the data gathered by interviews. Research subjects can in fact be thought to be a combination of unique biographical events and socially shared meanings, (Willig 2001) or stock of knowledge (Schutz 1970). Individuals receive and construct their experiences within social, cultural, physical contexts and schemes which impinge on these meanings (Hollway and Jefferson 1997; Hollway and Jefferson 2000; Silverman 2005; Silverman 2006). Consequently, experiences themselves are never pure, but inherently embedded and negotiated in a social and cultural web (Hollway and Jefferson 2000; Kelly 2009). Therefore, interviews achieve not only information about the research subjects' experiences, but also an insight into the social fabric of their interpretative framework and system of knowledge. This blend of individual and social that characterises interviews can lead to an enrichment of data rather than diminish them.

In sum, interviews provide data in context (Hollway and Jefferson 2000). Interview data offers the possibility of focusing on the discourses and language of participants allowing the researcher to locate cultural, social and individual stories and to uncover their structures. It is therefore highly important to analyse the data at different levels and consider the context of the interview. The researcher's self-reflexivity occupies an important place in this context.

In conducting and analysing the interviews for this research project, both their content and context will be taken into account. An important means adopted by this research project to obtain more contextual information is represented by the method strategy of interviewing many family members from the same network. This strategy is explained below.

### **Family Networks Approach**

The narratives of family networks are sought in order to explore the lived experiences of NF1 and its genetic aspects within the links of familial relations. Family networks are composed of individuals who are related by a blend of biological, social, legal and affective bonds (Gaff and Bylund 2010). As already discussed in the previous chapter the significance and very 'structure' of these bonds can vary across different individuals within the same families.

For this reason, I let participants define their own family network by asking questions about the influence of the syndrome on other family members and by adopting snowball sampling (Silverman 2005), that is, by asking them at the end of each interview whether they thought there could be other members in their family interested in participating to the study (see below § Research Procedure).



As stated at the end of the last paragraph, taking into account the ‘whole’ context of an interview is a crucial part of the analytical process of its theoretical interpretation. By looking at the wider socio-cultural contextual frame of an interview, the researcher can more easily avoid taking interviewees’ account at face value, i.e. avoiding treating the interviewees’ accounts and points of view as explanations (Cox and McKellin 1999; Silverman 2006). Therefore, enlarging the scope and collecting interviews also from family networks can be a useful methodological and theoretical strategy which provides the researcher with a richer set of information, increasing the possibility to detect and analyse more details and contradictions within interviews (collecting information that would have not been visible otherwise).

The family network approach provides the possibility to obtain a multiplicity of points of view, accounts and attitudes i.e. the perspectives of different family members on the same issues (NF1, genetic knowledge, coping strategies etc.) for each familial case, allowing for a methodological micro-triangulation. This will convey a richer and broader picture of the lived experiences of NF1 and their social relations and values. It also offers the possibility to detect patterns in the perceptions of NF1 within different family networks, for example, between the experiences of parents of individuals with NF1 and the experiences of siblings of individuals with NF1. Therefore the family networks approach is at the same time a method of sampling and data analysis.

On a more theoretical level, as discussed in the previous chapters (Chapters 2-3) the family occupies an important role in the social construction and management of genetic diseases. Families can be seen as a first (or at least a highly relevant) site where meanings and attitudes towards NF1 are formed and negotiated. Diagnoses often occur at a familial level and have familial consequences, for example they may be linked to

instances of genetic responsibility (Hallowell 1999; Finkler 2000; Featherstone, Atkinson et al. 2006; Williams, Skirton et al. 2009).

The collection of family narratives aims to explore how individuals' attitudes and behaviours towards NF1 are constructed as a result of, or in relation to, manifold family process. Therefore, families are also important sites to study and trace the genealogy of attitudes and understandings towards the syndrome.

Despite the fact that recent discussions about the social implication of the new genetics have highlighted the importance of studying genetic disorders and hereditary risk within the context of familial beliefs and dynamics (Cox and McKellin 1999; Fanos and Puck 2001; Downing 2005; Hallowell, Arden-Jones et al. 2005; Featherstone, Atkinson et al. 2006) this typology of family studies is still relatively novel. Empirical studies of patients' perspectives on genetic syndromes tend to focus primarily, or mainly, on individuals' accounts and the psychological implications of receiving an informative test result. Few studies examine, for example, how affected patients and their families jointly construct the meaning of genetic risk within their everyday lives and how, in turn, such pre-existing constructions shape the experience of genetic risk (Ablon 1999; Featherstone, Atkinson et al 2006; Whitmarsh, Davis et al 2007; Ashida, Hadley et al 2009; Skirton, Williams et al 2010; Weiner 2010). Moreover, this empirical research on experience with genetic disorders which has focussed on familial dynamics around genetic knowledge is normally based on interviews of one or at most two individuals within the same family (parents or partners) and tends to analyse individuals in the family as separate units (see for example Weiner 2010).

This research project aims instead for a deeper exploration of family illness narratives and dynamics which is achieved by interviewing as many family members as possible within the same network.

In short, the family network approach developed in this doctoral research is a relatively novel method of sampling and analysing data.

This method is conducive to studying how a condition with a variable symptomatology and prognosis (which can also vary within the same family) such as NF1, affects the family and whether and how it constitutes genetic subjectivities in family networks. In particular, this method appears to be suitable to unravel possible interrelation of individual and familial sense making practices around genetic knowledge and uncertainty.

Nevertheless, exploring family networks as a method also presents some problems. As discussed in the previous chapter, families can be problematic, fragmented complex entities full of psychological tension. Therefore, it can be quite delicate and complicated to explore them. With regards to this research study, the subjects touched on in the interview (e.g. the experience with NF1), can be a distressing familial topic and thereby it may be linked to uncomfortable issues or even taboos. Recruiting many individuals within the same family network is not always an easy task. Even the most collaborative families can find this form of recruitment to be too invasive. Some participants may find the idea of other family members being interviewed quite uncomfortable. This recruiting process can potentially be uncomfortable both for the one who recruits and the one who is recruited; participants can also feel the condition of confidentiality to be undermined.

On the other hand, this recruiting process, with its problems, can represent a source of useful information about family dynamics. In fact, interviewees' potential refusal to involve other family members in this study (and the rationale they may provide) may

represent in itself a relevant piece of contextual information about family dynamics and communications around genetic disorders.

### **Healthcare Providers Interviews**

Patients and families' accounts are also complemented by interviews with healthcare providers who are involved in the treatment or management of NF1. The aim was to cover as many relevant medical specialties significantly involved in the treatment and management of the syndrome as possible (e.g. medical genetics, paediatrics, oncology, dermatology, surgery, orthopaedics).

The main objective for pursuing these further interviews is to analyse and compare professionals' accounts and understandings of NF1 with the ones of patients and families. This allowed for a comparison between the so called 'lay' understanding of genetic knowledge with the one of healthcare professionals (Kerr 2003; Cox and Starzomski 2004). Furthermore, given the diversity of medical specialties involved in the treatment and management of the syndrome (see also Chapter 2), the interviews of healthcare providers may highlight different perspectives on the syndrome related to different medical approaches.

It is particularly important to collect information from professionals, not only because this provides a broader platform for a deeper insight into the psychosocial world of NF1, but also because it allows the researcher to have a more direct understanding of the scientific, medical and clinical thinking on NF1, one which is not based purely on the analysis of the available literature and patients' and families.

Moreover, on a more practical level, the juxtaposition of patients, families and healthcare providers' interviews will facilitate the identification of significant patterns or differences in patients and professionals' accounts of NF1. This information may be

relevant to the making of recommendations for clinical practice within the NHS relating to NF1 and other genetic conditions.

### **4.3 Research Procedure**

#### **Sampling and Recruitment**

The sample comprised:

- Individuals with NF1 and their family networks (n=30)
- Healthcare providers involved in the treatment and management of the syndrome (n=11)

I initially sought and obtained ethical approval from the HuSS Research Ethics Committee at the University of Exeter in relation to non-NHS patients, and from the NHS LREC in relation to NHS patients and health care providers. Copies of these certificates are provided in appendix 1.

#### **Inclusion criteria**

**Age:** all participants were aged 16 or older.

**Gender:** both male and female participants were included.

The inclusion of male and female and young and old people was relevant with regard to patients' and families interviews (Bowling 2004; Cox and McKellin 1999). It allowed me to tap a broad range of experiences which included a diversity of family members (e.g. grandparents, parents, and children) which is important for the aims of this research.

**Disability:** the research included individuals affected by NF1. Some of them presented with physical disabilities e.g. tumour/facial disfigurement as well as minor cognitive impairment e.g. dyslexia, difficulty with numerical calculation, minor learning

difficulties. However, individuals who were not able to consent were not included in the research.

**Ethnic origin:** no ethnic origin was originally excluded. However, in the end, the sample was mainly composed of white Caucasian individuals.

### **Data Collection**

I have worked with the lead consultant in a Clinical Genetic Department in the UK who acted as a professional collaborator. Data collection took place from 2008 to 2010.

Participants were obtained from two sources: a) NHS; b) non-NHS.

a) NHS patients were selected for interview by the lead consultant from the Clinical Genetics database of patients with neurofibromatosis.

A purposive sampling strategy (Bowling 2004) - i.e. a non-random method of sampling, often adopted in qualitative research, where a group of people with a specific characteristic are sampled - was employed.

The consultant selected patients with different symptomatology and degrees of severity, in attempting to cover the high variability of NF1 and obtaining a representative sample. However, to comply to the NHS Ethics requirements the consultant selected only patients who were able to consent, excluding individuals with severe cognitive impairment.

On my suggestion, and on the basis of professional collaboration, the consultant selected as well a number of healthcare professionals who could be invited to participate in the study. Given the large number of specialties which can be involved in the management of NF1 (see Chapter 2) we aimed at sampling from professionals with

different backgrounds. However, we also wanted to interview specialists who were reasonably familiar with the syndrome.

Genetic counsellors, genetic consultants, medical genetics scientists, oncologists, ophthalmologists, paediatricians, surgeons, educational psychologists, family advisors were invited.

I managed to interview professionals from all these backgrounds, but educational psychology and oncology.

Healthcare professionals and patients were invited on headed paper from the Clinical Genetics unit, ; the invitation letters included a reply slip and the information sheet. The invitation was forwarded with a covering letter signed by the lead consultant . I waited for participants to return the reply slip to the genetic unit stating their willingness to participate before contacting them for the interviews. Copies of these documents are provided in appendix 2.

b) Non-NHS participants ('healthy volunteers'). Some patients and family members (n= 9) were not obtained from the NHS database, but through researcher's personal contacts with individuals/families with NF1 and from asking the participants recruited through the lead consultant if other family members wished to participate. Thus, a snowball sampling strategy (Hollway and Jefferson 2000) was also employed. This method involves the researcher asking an initial group of respondents to recruit other people they know who could be suitable to participate in the study. Healthy volunteers were not invited by letter, but were normally contacted by phone or email by the researcher.

For this research project -particularly in the light of the variability of NF1- it was important to explore both clinical and non-clinical settings to ensure that a more

representative sample of family networks with NF1 is recruited (not just those who have contact with Clinical Genetics). The snowball sampling strategy also allowed the sampling to take place from different geographical areas of the UK.

## **Interview Format and Process**

### **Patients and families interviews**

Individuals who agreed to participate in this study were contacted by the researcher and asked to suggest time and venue for the interview (in order to make the interview setting as comfortable as possible for them). Three participants opted for the clinical room in the department of Clinical Genetics in Exeter Hospital, two were interviewed in the University of Exeter, one in a park, the rest decided to be interviewed in their homes.

Interviews were face-to face and lasted from 40 to 120 minutes.

All interviewees agreed to the interviews being recorded and all were recorded digitally.

Interviews were semi-structured, i.e. guided by a schedule elaborated in advance.

The interview schedule was slightly amended after the first three interviews i.e. some questions were just reworded in order to be simpler.

Patients and families interviews principally focused on illness experience from personal, familial to wider social level, as well as on experiences with the healthcare system. The sequential order of the questions and their open nature were designed to elicit narratives of illness, health behaviours, familial dynamics, inheritance, genetic aspects of the syndrome and impact of illness on identity without directly forcing these themes on the interviewees (Groleau, Young et al. 2006). Interview schedules for patients and family members can be found in Appendix 3.

Participants from the same family network were interviewed separately in order to allow confidentiality and to talk freely about their personal and familial illness experiences.



Normally towards the end of the interview each participant was asked if they knew other family members who could be willing to participate in the study. This implies that participants were put in the situation (if they were willing) to explain to other family members the nature of the study and to reveal their involvement in the study. Often during the interviews, or afterwards, participants gave detailed explanations and justifications on why other family members were or were not or could not be asked to participate, providing fruitful prompts for further discussions and valuable information on the coping strategies and familial relation around NF1.

### **Healthcare Providers' Interviews**

Healthcare providers who agreed to participate in this study were contacted by the researcher via telephone or email.

They were all interviewed in their offices or departments' facilities apart from one who was interviewed in the University of Exeter. Participants were located in different geographical areas of the UK, but I will not provide more specific geographical details to protect their anonymity. Interviews were face-to-face and lasted approximately 30 to 50 minutes.

All interviewees agreed to the interviews being recorded and all were recorded digitally. Interviews were semi-structured and focused on participants' professional experience with NF1, the management of the syndrome in the NHS, their relation with NF1 patients and families with a particular interest on how genetic information is communicated to and understood by patients. The exact questions used in each interview depended to some extent on the speciality of the participant and their role in the management of NF1. Interview schedules for healthcare providers can be found in appendix 3.

## Demographic Details

### Patients and family Sample

TABLE 1

Family networks	Interviews	Individuals with NF1 diagnosis interviewed	Men	Women
I	2	0	1	1
II	1	0		1
III	5	1	4	1
IV	2	2	1	1
V	4	4	1	3
VI	2	1	1	1
VII	1	1		1
VIII	2	1	1	1
IX	3	2	2	1
X	1	1		1
XI	2	2		2
XII	2	1	1	1
XIII	3	1-2 <sup>11</sup>	2	1
<b>Total</b>	<b>30</b>	<b>17-18</b>	<b>11</b>	<b>19</b>

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<sup>11</sup> One of the family members was unsure about their NF1 diagnosis.

TABLE 2

Family network	NF1-related symptoms reported (individuals interviewed and family)	New mutation	Family history
I	Café au lait marks, internal neurofibromas, external neurofibromas, dyslexia		1-2 generations
II	Café au lait marks, internal neurofibromas, external visible neurofibromas, learning difficulties, epilepsy	X	
III	Café au lait marks, internal neurofibromas, external visible neurofibromas, bone problems, physical impairment		3 generations
IV	Café au lait marks, external neurofibromas bone problems, learning difficulties, physical impairment		2 generations
V	Café au lait marks, internal neurofibromas, external visible neurofibromas, eye problems, learning difficulties, cancer		3 generations
VI	External visible neurofibromas	X	
VII	External visible neurofibromas	X	
VIII	External visible neurofibromas, eye problems		2-3 generations
IX	External visible neurofibromas		3 generations
X	Bone problems, learning difficulties, physical impairment		3 generations
XI	Café au lait marks, learning difficulties, external neurofibromas		2 generations
XII	Cancer, learning difficulties, external neurofibromas		2 generations
XIII	Eye problems, bone problems, hydrocephalus physical impairment		1-2 generations

TABLE 3

<b>Reported Experience with Healthcare System</b>	<b>N. of family networks</b>
<b>Removal of neurofibromas</b>	<b>4</b>
<b>Orthopaedic and other surgical intervention</b>	<b>3</b>
<b>Cognitive-learning Support</b>	<b>4</b>
<b>Psychiatric support</b>	<b>1</b>
<b>Educational support</b>	<b>1</b>
<b>Social service support</b>	<b>3</b>

TABLE 4

<b>Age</b>	<b>Interviews</b>
16-30	3
31-40	11
41-50	10
51+	6

TABLE 5

<b>Occupational Class</b>	<b>Interviews</b>
<b>Managerial, Professional and Intermediate</b>	<b>14</b>
<b>Routine and Manual</b>	<b>13</b>
<b>Student</b>	<b>2</b>
<b>Unemployed</b>	<b>1</b>

## **Professionals' Sample**

Genetic counsellor (n=1)

Genetic medicine consultant (n=2)

NFA staff member with additional experience as NF family advisor<sup>12</sup> (n=1)

Ophthalmologist (n=1)

Paediatrician (n=3)

Surgeon (n=2) (surgeon1: orthopaedic surgeon, surgeon2: plastic surgeon)

Scientist (n=1) (medical geneticist)

**Total: n=11**

## **Analysis of the Interviews**

### ***GT***

Interviews were analysed during and after the data collection period. The collected data were fully transcribed (verbatim) and analysed, with the help of the computer software for textual analysis NVivo9. The transcription was already a way to familiarise myself with the data. Systematic open coding was employed, i.e. codes were generated inductively, assuring that the emerging concepts were fully grounded in the data. This involved breaking down the transcriptions into sections which appeared significant for the research questions, and/or for theoretical, methodological or any other reason and labelling or coding these sections. Codes were inductively developed as a dialectical iterative process of moving between the interview transcripts, the emerging code list and the research questions, making sure that the concepts expressed by the codes were firmly grounded in the data (Strauss and Corbin 1990). Data collection was combined with analysis and theory building in an on-going process, in order to encourage continuous review of earlier stages of analysis. This procedure is called the *constant*

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<sup>12</sup> Family advisors are NF specialists advisors who work within hospitals and give support to anyone affected by NF1 (cf Chapter 8).

*comparative method* which is a crucial feature of GT (Strauss and Corbin 1990; Hollway and Jefferson 2000).

The line-by-line analysis of transcripts produced a considerable number of codes (over 150). These initial codes were compared, related and collapsed as new data were collected and at the end of data collection.

Through axial coding, categories were grouped into more abstract themes. NVivo made the coding process easy to handle and possibly less laborious than using Word documents.

The supervisors gave feedback during the whole process of analysis discussing codes, categories and relationships.

Throughout the lifecycle of the research I kept a journal (memo) in which I noted reflections on the data, the interviews settings and my impressions and feelings about the interviews as well as on my interview skills. I also compiled a summary for each interview in which participants quotations were accompanied with background information on the participant, observations about the interviews and theoretical reflections. I also tended to make notes and compare the interviews of different family members. These notes and memos were extremely helpful in refining the codes and in identifying major themes.

#### **NA**

In order to further unravel the links between individual and familial experiences of NF1 and genetic knowledge I decided to employ also NA method of analysis. Towards the end of the GT analysis process I selected a family from the sample on the basis of the richness of the interviewees' accounts and because it was the family I explored the most (I interviewed five individuals from this family network).

In this research project, although almost all the interview questions were designed to be open and elicit narratives, the interviews did not technically fit the in-depth narrative

model (narrative interviews are generally longer and have a biographical approach (Hollway and Jefferson 2000)). Nevertheless, several interviews of individuals in the same family network can create a wider and more complex narrative space.

As for GT, the target of the NA conducted in this research project comprised not only the whole transcripts of each participant, but notes, the self-reflection of the interviewer about the participant, the whole interview. Accounts of other family members on the participants were also part of the NA analysis process. The NA approach has also allowed me to further reduce the problem of fragmentation of data, which is very difficult to avoid using GT and other coding strategies,

To conduct the narrative analysis I re-read the uncoded transcripts of the family members interviews trying to identify and extract core narratives, or basic plot lines (Bury 2001; Frank 1995). I compiled for each member a detailed profile which comprised: biographical information (Hollway and Jefferson 1997) including experience with NF1 and relevant information about links with other family members; core narratives accompanied by extensive quotations and my notes. By comparing the core narratives of each family member, I started to refine and integrate them. I also rewrote in my own words the storylines of each individual, in order to help me to reflect on the phenomena described by the family members and individuate the most salient ones.

**TABLE 6**

Example of raw data and codes

Theme	Sources	Quotation
Clinical Experience	24	D: You were not able to find a doctor...?  No really, I found them a bit... scary...
Genetics/ Family History	22	I don't remember I have ever seen her and that. But mum said, my mum said, she [grandmother] used to have lumps and bumps all over her face....but I never saw her so I don't know and dad didn't talk about it, so...it was just....my mum thinks I have inherited it from... whether I did or not...don't know it has not been proved' cause she was dead when I was...god knows so....
Identity/ NFI Downplayed ("don't want to know")	24	This is a mentality, I didn't want to know a lot about it, the lumps were coming gradually getting more, I've got a lot more now, thousands I would think, if I bother to count, but I haven't

### **Data Management**

In terms of data protection, all of the interview transcripts, tapes and voice files are held anonymously (i.e. with participant numbers as identifiers). The key containing the identifying names, addresses and contact numbers was held on the password-protected University of Exeter system as a separate Word file by the researcher/supervisor.

Interview data will be held at the University of Exeter in a locked cabinet for 5 years before being destroyed. All the names employed in the results chapters are fictional.



## **Potential Problems**

In terms of assessment of possible harm, some people, especially parents of NF1 children, could find discussion of this genetic condition distressing. However, sharing experiences about the disease with the researcher could also alleviate some of this stress. This was stated on the participants' information sheet, which contained as well information about the national support group which operates a helpline, in addition to a recommendation to contact the GP with any concerns. In the invitation letter the entirely voluntary nature of the participation was clearly stated.

In terms of risks for the researcher, the researcher visited the majority of the participants in their homes, while healthcare providers were interviewed in their offices. The supervisors had the contact details of each visit and had been informed both before and after the occurrence of every interview by the researcher.

## **4.4 Concluding Remarks**

In conclusion, this research employs two complementary methods: GT and NA inspired methods. The family network approach developed in this doctoral research is relatively novel and it is both a method of sampling and analysing data.

The next chapter (Chapter 5) presents the NA of one specific family from the sample, while the subsequent three chapters (Chapters 6-8) are based on the GT analysis of individual transcripts.

## **5. Family Narrative Analysis**

### **5.1 Introduction**

The following chapter explores the illness narratives of one specific family from the sample, tackling a crucial interest of this thesis, namely, the interrelation between individual and familial experience of illness. It will focus on how the experience and relevance given to risk change over an individual's lifecourse and how these changes can be significantly linked to family dynamics (Parsons and Atkinson 1992; Cox and McKellin 1999).

The impact of illness on family life has been widely acknowledged in research on the experience of chronic and genetic illness (Bury 1982; Finkler 2000). A probably less explored, but complementary object of investigation is the study of the ways in which the family setting (e.g. family dynamics, bonds and structure) can influence the understanding, experience and management of illness (Cox and McKellin 1999; Gregory 2005; Gaff and Bylund 2010).

In the previous chapters, I have discussed how genetic information has considerable familial implications. Risk and diagnoses can lead individuals to engage in disclosure and genetic responsibility behaviours with their family members (Hallowell 1999; Finkler 2000; Featherstone, Atkinson et al. 2006; Williams, Skirton et al. 2009).

Familial dynamics and environment can strongly influence the psycho-social impact that genetic conditions or genetic risk may exert on the identity and behaviours of individuals, playing a very important role in the way individuals come to terms with and manage genetic risk and diagnosis (Skirton 2001; Featherstone, Atkinson et al. 2006).

Family dynamics can therefore represent decisive biographical instances which shape

the multifaceted and variable individual response to genetic knowledge. Not only can the family setting shape (in many indirect and direct ways) one's life and illness experience, but it is even more critical in relation to the management and experience of genetic disorders, given the familial implications of genetic information.

Focusing on illness narratives provides a valuable means for the exploration of the manifold interrelations of individual and familial dynamics on genetic disease and knowledge. Lifecourse foregrounding of genetic risk and family dynamics will also be the object of discussion in the next results chapters. However, despite my attempts to preserve the original flow and context of participants' accounts . by employing 'inclusive coding' (Strauss and Corbin 1990) and the family network analytical approach , in the next result chapters I will fragment and group participants discourses under common themes in order to give an overview of the results across the whole sample.

In this chapter, I focus instead on one family and present the narrative analysis of the family members interviewed from this network. In conducting the narrative analysis I have treated this family and the individuals who compose it as a unit. The aim was to reconstruct and present individuals' illness narratives in the light of the broader familial context (delineated by the juxtaposition of the family members' interviews) avoiding as much as possible to fragment their accounts (Hollway and Jefferson 2000). I have selected this specific family narrative from the sample as it offers a particularly rich example of the entanglement between familial narratives of illness, responsibility and other familial dynamics and individual experience of self, disease and genetic knowledge. This is probably also due to the fact that this is the network I have explored

the most - I have interviewed five individuals from this network. Moreover, these interviewees were particularly eloquent and articulate.

In presenting the findings, I will draw on research on illness narrative and use in particular Frank's narrative types (1995). In line with narrative thinking (see Chapter 4), Frank's tenet is that stories of illness experience are composed of individual elements structured around main storylines or narrative types i.e. cultural models available in our society (Frank 1994; Frank 1995). Through an original combination of literary and sociological approaches, Frank has analysed the content and structure of various illness accounts and has distilled them into three main storylines: restitution, quest and chaos (Frank 1995). In the restitution narrative a person is ill and after seeking treatment gets better and/or goes back to the 'pre-illness' self. In the quest narrative the person does not necessarily get better, but the experience of dealing with the illness has an overall positive transformative effect on the person's life. In the chaos narrative the person does not get better and is overwhelmed by the illness. Chaos narratives are characterised by resignation and fatalism towards the illness and loss of control over life as a whole. They neither have a 'happy ending' nor a real narrative structure. They are antithetic to the restitution narrative.

In accounts of illness experience, these three narrative types are often intertwined (Frank 1995; 1994). Both restitution and quest narratives underlie the resolution of the majority of illness accounts which are also available in the media and other resources, whilst chaos narratives typically represent the moment of tension/climax which is (or will be) overcome and dissolved into a final restitution or quest.

Frank's storylines provide a very useful means for the analysis of narratives as they allow to reflect both on the form and content of accounts of illness experience. Having

already an awareness of these basic plot sequences (Sandelowski 1991) allows the listener to concentrate their energy on appreciating more deeply nuanced meanings which may otherwise be lost. This awareness can also help to easily detect and unpack the ways in which the story conveys or allude to the relationships between the affected storyteller and the outside world (Frank 2010).

Unlike other analytical frameworks which have been developed to explore illness narratives,<sup>13</sup> Frank's three storylines have the advantage of being easily accessible not only to social and cultural theorists, but also to healthcare providers as well as patients and families (Thomas-MacLean 2004).

For all these reasons, Frank's work has been fruitfully employed in both cultural and clinical studies on chronic illness, particularly of cancer and disability (Thomas-MacLean 2004; Willig 2011).

The adoption of Frank narrative types can help to increase our listening skills and thereby our grasping of the needs and coping patterns of affected individuals. This in turn can benefit professional and patient relationships, leading to an improvement of the quality of life of affected individuals and their family members (or others who take care of them).

In this chapter Frank's storylines will be used to show that genetic illness narrative are not just individual, but familial. The majority of narrative studies – including the work of Frank – ultimately imply a tension (and mutual influence) between the individual and society (or culture). I will add to this dyad the family. The family can be seen as occupying an important middle ground and filter in between the individual and society.

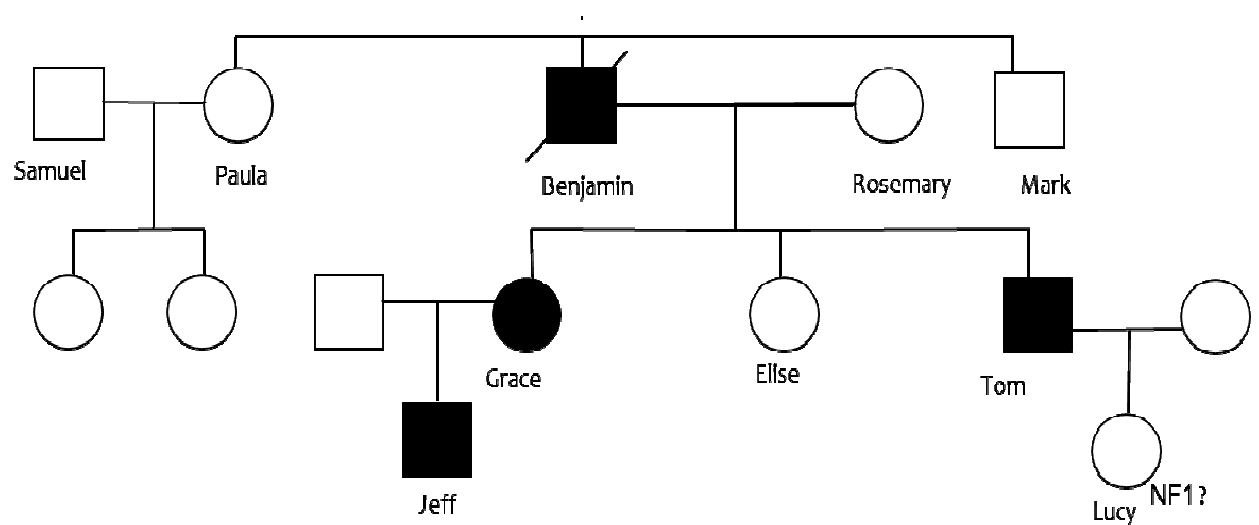
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<sup>13</sup> See for example Bury's (2001) theoretically acute, but potentially cumbersome distinction between 'contingent', 'moral' and 'core' narratives. The first addresses beliefs about the aetiology of a disease; the second the links between the disease and the individual sense of identity and social relations. The latter the affinity of individual accounts of illness to cultural genres and meaning attached to illness (e.g. heroic, comic, regressive narratives).

As concerns genetic disease, I will show that the family can significantly shape and influence individual perception and responses to genetic knowledge and disease.

This chapter begins with contextual information about the family and the recruiting method. The familial experience with NF1 is reconstructed by focusing on the stories of two individuals from this family. The exploration of these narratives will illuminate significant instances related to the infiltration of familial relations, feelings and duties and health management discourses and practices towards the syndrome. The chapter ends with a discussion of the relevance of family dynamics in researching patients' experience with genetic diseases.

## 5.2 The Family



## **Biographical details**

The family members interviewed were living in Greater London and Cornwall. All respondents were white, highly educated, middle-class individuals. Below, biographical details of the respondents and of the other family members they often referred to in their interviews are provided. All the names are fictional.

### ***Grace*** (interviewed family member)

Grace was a lively and educated woman in her late thirties. She has NF1, but was almost unaffected (main symptoms: café-au-lait marks, UBOs<sup>14</sup>). She worked as a journalist and has written for major newspapers. At the time of the interview she was also working on a book proposal. Grace had a son, Jeff, who was in his early teens and had NF1. Jeff's father left when he was five years old. Grace reported that Jeff has been looked after regularly by professionals and seemed to have some mild cognitive and learning problems.

### ***Elise*** (interviewed family member)

Grace's sister; in her mid-thirties, with dyslexia. She worked as a restaurant manager. In a relationship, she did not have any children at the time of the interview.

### ***Tom*** (non-interviewed family member)

Brother of Grace and Elise, in his early thirties. He had NF1 and was mildly affected; he was described as having a few non visible neurofibromas and dyslexia. He experienced episodes of epilepsy in his teenage-early adulthood. He had a daughter Lucy, around two years old and undiagnosed at the time of the interviews.

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<sup>14</sup> Unidentified Bright Objects. These objects are shown by cranial MRI scans in patients with NF1. They tend to disappear with time and have rarely been found in individuals with NF1 over the threshold of 20 years. Some researchers have suggested connection between UBOs and lower IQ, language skill, visual-spatial functioning and academic achievement, but this hypothesis is still controversial (see Chapter 2).

***Benjamin*** (non-interviewed family member)

Father of Grace, Elise and Tom, with NF1 and seriously affected, he died approximately ten years ago of pneumonia. Benjamin was reported as being the first diagnosed case of NF1 in the family. He was the middle child; the two other siblings are Paula, the eldest, and Mark. Benjamin suffered from external neurofibromas, but the main complications stemmed from internal neurofibromas in the spine, that made him become progressively quadriplegic. A very active and bright man, he was a successful oil engineer and got involved in many charity initiatives to raise money for NF1 and to promote workability for people with impairments. All the individuals interviewed in the family described his incredible determination and exceptional coping skills with his disorder. However, Benjamin was also reported to experience a five year long very critical phase characterised by anger and denial of his condition. This period was difficult to manage for his family, especially for his wife Rosemary, who at some point left him. This phase was overcome once Benjamin- thanks also to the help of a specialist nurse- managed to accept his NF1 and quadriplegia.

***Rosemary*** (interviewed family member)

Very energetic and clever woman. In her early 60s, mother of Grace, Elise and Tom and wife of Benjamin. She was a nurse and came from a medical family. Her father was a GP, and her uncles were all surgeons. She was Benjamin's wife and his full-time carer.

***Paula*** (interviewed family member)

Sister of Benjamin and Mark, in her 60s, she used to be a teacher. Very lively woman. She was married to Samuel and they had two daughters.



*Samuel* (interviewed family member)

Husband of Paula. In his 60's, former PE instructor, he attended high school with Benjamin, but they were not in the same year. Frank, Paula and Rosemary were on good terms and still in touch at the time of the interviews.

### **5.3 Recruiting Method**

This family has not been recruited through the department of clinical genetics, but using snow ball sampling technique. First contact was made with Grace (recruited through acquaintances of the researcher) and an interview was arranged at her home. At the end of the interview, when asked about the possibility of interviewing other members of the family, Grace suggested her sister Elise. In the subsequent days, after checking the availability and willingness of her sister, Grace emailed me Elise's contact details. I contacted Elise and agreed to interview her at her house. After this interview Elise phoned, while I was still present, both her mother Rosemary and her aunt Paula, asking them if they were willing to participate in the study; they both replied positively. I contacted Rosemary first since she was living near to Elise and managed to interview her the day after Elise's interview. As decided by Rosemary, this interview took place on a bench in a public park. Rosemary suggested I could interview her son Tom, and was willing to contact him first. There has not been a follow up from this, but I preferred not to ask further in order not to bother the family who already responded so generously and enthusiastically to my study. I also suspect that - as Grace suggested during her interview - Tom was probably experiencing a delicate moment in his life due to the birth of his daughter who may have NF1. I was able to interview Paula only a few months after Rosemary's interview. This interview took place at her house and when I asked Paula about the possibility of interviewing other family members she promptly

asked her husband, Frank, who was also at home and I managed to interview him too on the same day.

#### **5.4 Grace's Narrative Arc**

*Grace: It [Grace's experiences with NF1] has absolutely and definitely changed over time, it kind of went from an experience of a child and her father, to a teenager with a diagnosis she didn't want and refused to accept, to a mother accepting her own diagnosis and actually being quite proud of it (my emphasis)*

Grace - a young woman with NF1 - gives in the above passage a very clear summary of how her experience with NF1 has significantly changed throughout her life. These changes underlie different attitudes towards genetic knowledge. Within these fluctuations which constitute the arc (the trajectory) of her illness narrative, it is possible to isolate some decisive phases that - as I will try to show below - are strongly entangled with familial dynamics.

##### **“A child and her father”**

Benjamin and Rosemary decided to have children notwithstanding Benjamin's diagnosis. Before Grace - the first child - was born, Benjamin already had the first laminectomies<sup>15</sup> and Rosemary, given her nursing background, had already decided to dedicate the majority of her time to look after him.

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<sup>15</sup> Surgical operations of the spine.

Hence, NF1 had been introduced to Grace within the familial environment. She stated several times that in her household there was a culture of normalization (Bury 2001; Sanders, Campbell et al 2007 see below § 5.5) towards NF1.

*Grace: I grew up knowing that my dad had neurofibromatosis and it was always treated as something very normal and interesting in the family*

Although Benjamin was already starting to experience the initial serious signs of weakness and lack of control over his body, he maintained a positive attitude towards his syndrome.

*Grace: he [Benjamin]’d say “oh I am special; all the consultants want to talk to me. Whenever I am in the room everybody... they always will ask everyone to come and see me”. He framed it as something special*

On the contrary, her mother Rosemary, coming from a medical background, tended to deploy a colder, detached medical approach towards Benjamin’s condition.

When Grace was a child, Rosemary and Benjamin, notwithstanding their frequent contact with health care providers, had an “*incorrect idea*” about the inheritance pattern of the syndrome; they believed that the disorder could be only inherited through the male line. Thus, in her infancy and early teenage years, Grace was used to the fact that the father had the condition and - even when she started developing café-au-lait spots - she did not consider the possibility of having the syndrome as well. But Rosemary and Benjamin discovered in the subsequent years that NF1 can be inherited equally by both sexes.

**“A teenager with a diagnosis she didn’t want and refused to accept”**

Suddenly, one morning, when she was 16 years old, Grace was woken up by her mother who counted her café-au-lait marks all over her body to see whether she had NF1. Grace recounted this event as a very traumatic one for her; she described herself as being extremely annoyed by Rosemary’s ‘diagnosis’ which, in her opinion, was performed without sensitivity. Grace suggested that particularly on this occasion she suffered the ‘medical detachment’ (i.e. the cold and pragmatic attitude) of her mother towards NF1, experiencing it as inappropriate.

*Grace: so I didn’t think I had it [NF1] until I was 16...and I had this kind of very bizarre experience of being woken up by my mum one day, who is not the most sensitive of individuals...looking at me, looking at my body and counting...because I have the café-au-lait marks and counting them...and I was like “what are you doing?” and obviously, you know, I was a teenager I was quite sensitive and...she said “oh well I am just checking, if you’ve got more than 5 café-au-lait marks then you might have neurofibromatosis as well!” And I was really...I was really annoyed, and offended actually, and upset. Kind of by the way it’d been presented to me and kind of by the whole thing. My mum comes from a medical family, she is a nurse, her father was a GP, her uncles were all surgeons...so she was very matter of fact about it. That wasn’t the way I wanted that information presented to me*

Subsequently, all the three siblings were referred to the department of clinical genetics to be inspected; two of them, Tom and Grace, were found to have NF1 and Elise did not. Grace recalled being very upset, shocked and traumatised; she also pointed out

during the interview she did not have any emotional and psychological support around her diagnosis during that period.

Following this traumatic diagnosis - performed by the mother first and then confirmed by clinicians - Grace refused to accept and come to terms with her own NF1. This period in Grace's life, which strongly overlaps with her teenage years, represents a significant critical juncture of her illness narrative (Parsons and Atkinson 1992; Cox and McKellin 1999; Petersen 2006): she became "*a teenager with a diagnosis she didn't want and refused to accept*" This stage of firm denial towards the diagnosis is very well captured in the following passage:

*Grace: personally I totally shut off from it [NF1], I refused to talk about it I refused to even accept it to myself, and if anyone in my family said to me "oh you've got it as well haven't you?" I would say "No. I don't know what you are talking about" and would completely block any further discussion on the matter (my emphasis)*

Grace's awareness about her personal diagnosis was not accompanied by a desire to change her identity and possibly engage in behaviour geared to monitor and minimize risk. Rather, Grace's relation with NF1 was more stratified. Similarly to many studies discussed in Chapter 3 (Cox and Starzomski 2004; Klitzman 2009), Grace received and interpreted new medical information in the light of already existing experiences and value systems. In particular, Grace's awareness of, and relation with, NF1 was mediated by familial factors.

Rosemary, the mother, who deployed a medical, detached attitude towards NF1 and performed the unpleasant diagnosis, appears to be considerably linked to Grace's lack of acceptance towards her NF1 diagnosis. Benjamin, the father, who was seriously

affected by NF1, but deployed a positive attitude towards it, represents instead a significant point of contact for Grace and her diagnosis. In fact, during the ‘negation’ period towards her NF1, Grace was living with, helping and supporting her affected father. Arguably, Grace was experiencing her own diagnosis predominantly in an indirect fashion, through the father. This intricate and contradictory experience of refusal and acceptance of NF1 clearly emerges in the following passage:

*Grace: probably until my mid 20s I pretty much refused to think about it, talk about it or engage with it in any shape or form...for me. In terms of my dad, that was a completely different subject, because we were living with him and supporting him [...] my perceptions of NF with my dad were quite proud and if anyone made any comments about him, ‘cause he gradually looked more and more disabled, I’d be very protective and proud of him and what he was and what he had. But when it came to me and my diagnosis I took a lot of, of getting used to it (my emphasis)*

Grace acknowledged and engaged with the NF1 of the father, Benjamin, whereas she left her own diagnosis out of her sight. Grace’s relation and experience with NF1 could reflect Benjamin’s ‘normalising’ and original way of living with his condition. The father’s and familial culture of normalization towards NF1 may have influenced Grace’s tendency not to engage with her diagnosis or, at least, not to live it in an anxious way. On the other hand, it could also be possible to argue that Benjamin’s severe NF1 which required significant care and attention (from all the family) represented a further cause for Grace’s dismissal of her own milder form of the disorder. Thus, the seriousness of Benjamin’s symptoms may have hindered Grace from engaging with her own diagnosis.

Alongside herself, Grace seemed to remove also the mother from her personal familial 'NF1 experiential picture'. After been asked about Rosemary's experience with NF1, Grace hesitated, as she realised she had never considered the mother to be part of this picture:

*Grace: Oh yeah, oh well yeah, there's her too... umm... well... [...] her experience of it I suspect would be quite different because she was a medical person... [...]... what was her experience of it? I don't know actually..... I really don't know... I should ask her shouldn't I?... (laugh) I don't know, I really don't know actually..... [...] I don't know... that's quite... I don't know... I guess, she experienced it medically and in quite a negative way personally... I don't know if I can answer that... you've stumped me... (my emphasis)*

Grace saw NF1 as being "Benjamin's thing". Despite Rosemary's 'obvious' and substantial involvement with the syndrome, not only as a mother and wife, but also as a full-time carer of Benjamin, Grace tended to remove the mother from her personal NF1 familial experiential world and to avoid communicating and sharing with her any aspects related to the condition. When asked whether Rosemary, given also her medical background, was in charge of the management of her NF1 Grace replied:

*Grace: I don't really discuss it [NF1] with her [Rosemary] because I kind of think it's not her... it's like my dad's thing, and I didn't want to talk about it with her very much...*

Grace tended to avoid talking about NF1 also with her siblings, including even the brother, Tom, who actually had NF1 as well, both in the past and at the time of the interview:

*Grace: With my brother, we don't really talk about it [NF1] actually, so maybe that's something in itself (my emphasis)*

These example of family communication are in line with the notion of practical kinship (Featherstone, Atkinson et al. 2006). Grace's narrative clearly illustrates how processes of disclosure, communication, bonds and sharing of illness experience within families are not simply dictated by biological closeness, but can vary according to the perspective of each family member. It is also worth noticing that problems and/or lack of communication about genetic disorders may not only occur between those in the family who have the condition and those who have not, but it can also take place between family members - like Tom and Grace - who actually share the same diagnosis. As Grace's story unfolds, there are other significant familial triggers that modify her relation towards NF1 and genetic knowledge, which are provoked by 'new' family members.

### **“A mother accepting her own diagnosis”**

The event that radically changed Grace's attitude towards her diagnosis was an unplanned pregnancy. Grace accepted her diagnosis only when she became the mother of Jeff, a child affected by NF1, who at the time of the interview was ten years old. Thus, in Grace's narrative there is a temporal leap of almost ten years between receiving the NF1 diagnosis when she is a young teenager and her full acceptance and management of it, which occurs only when she becomes a mother. Before the



pregnancy, Grace would only relate to and accept Benjamin's NF1, whilst firmly denying her own; but the new experience of motherhood completely overturned her denial.

At this point in Grace's narrative the 'adult mother' let in her NF1 perceptual-experiential world Benjamin's, Jeff's and finally her own diagnosis.

This clear link in Grace's narrative between the experience of motherhood and the acceptance and engagement with her genetic condition, resonates with other research on parenthood and genetic responsibility already discussed in Chapter 3 (especially section 3.2) (Graham 1979; Hallowell 1999; Novas and Rose 2000; Hallowell, Arden-Jones et al. 2005; Arribas-Ayllon, Sarangi et al. 2008). Particularly significant is the parallel with Hallowell's study (Hallowell 1999),<sup>16</sup> of women who are identified as having the potential genetic risk of developing hereditary breast/ovarian cancer (HBOC). The women in this study underwent genetic testing since they perceived themselves as having a responsibility towards their kin (including past, present and future generations) in order to ascertain the genetic risk of the rest of their families. In relation to Grace's case, it is significant that these women's feelings of genetic responsibility towards their kin prevailed over their right not to know about their genetic risk. Furthermore, these women presented their identity as being relational i.e. in-relation to others (prevalently family members). They tended to prioritise other family members' needs rather than their own. It was common for mothers in particular to justify the decision to monitor their genetic risk by referring to their obligations and duty of care towards their children. This same tendency was also expressed by Grace:

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<sup>16</sup> Grace's and Hallowell's women scenarios are not completely identical. Grace has a diagnosis of a genetic condition, whilst the women in Hallowell's study are identified as being genetically at risk for HBOC.

*Grace: I only started dealing with it [the NF1, diagnosis] actually when I became pregnant with my son and so that's when I was 24? Or 25...mid-twenties...so that's the thing that made me call up, go to my GP and say "I need to be referred to the genetics clinic"*

Hence, this juncture in Grace's narrative about her experience with NF1 mirrors the stories told by many other women interviewed by Hallowell who reported that it was not until they had the responsibility of caring for children that they started worrying about and managing their own genetic risk. The experience of parenthood and the consequent feeling of responsibility towards the offspring can generate or awaken parts of one's identity that were inexistent or dormant. Grace knew she had NF1 since she was 16 years old, but the acceptance of her genetic condition and her entrance into the medical system occurred only when she has her son. Hallowell also observed that genetic responsibility can be trans-generational (it can be directed to present, past, future generations) (Hallowell 1999). With reference to this aspect, it is also worth recalling that before being pregnant Grace ignored her own NF1, but at the same time she fully acknowledged her father's condition and looked after him.

Grace's illness narrative strongly revolves around familial elements, such as the seriously affected and interesting figure of the father, the traumatic diagnosis performed by the mother and the pregnancy. In line with the ideas of lifecourse foregrounding of genetic risk and practical kinship already discussed in Chapter 3, Grace's example shows once again how individuals can respond to genetic diagnoses in fluid and not necessarily consistent ways; such responses are significantly influenced by family dynamics and relations. This strong interrelation of Grace's NF1 identity discourses

with her experience of the syndrome in the family context surfaces several times throughout the interview.

A further example of the pervasiveness of family elements in Grace's illness discourses can be found in the articulation of her position against prenatal screening for NF1.

Recalling whether she was offered prenatal testing for Jeff she stated:

*Grace: I do feel really strongly about that because say if I'd been offered it, it had been positive and I'd had made a decision to not continue with the pregnancy, well like that's what a disrespect to my father and to my brother and to myself, that's like saying we don't deserve to live, we don't deserve to be born.. we are more of a problem than we are opportunity*

As the passage shows, Grace deployed once again a relational-familial NF1 identity to justify her position against any kind of prenatal screening for NF1 (Hallowell 1999; Arribas-Ayllon, Sarangi et al. 2008). It is significant to point out that she did not talk about the variability and different degrees of severity of the syndrome.

Grace's position of not favouring PND as disrespectful to other family members represents a common theme in the literature on disability right and prenatal genetic testing (Parens and Asch 1999; Raz 2004).

### **"...and actually being quite proud of it"**

The experience of motherhood led Grace not only to come to terms with her diagnosis, but also to assign a positive value to it. The 'adult mother' Grace presented NF1 as being part of herself. In line with Frank (1995) quest narrative, she even described the syndrome as something that has crucially and positively shaped her personality. In fact,

during the interview, Grace often stated that NF1 has made her an interesting person; the following passage is just an example:

*Grace: I am quite proud of who I am and what I am, and I believe that what's different about me is what makes me an interesting person. I think I probably would have been a much less interesting person had I not being different from other people in that way*

Touching upon the issue of the ambiguity between identity, body and illness (Kelly 2005; Landsman 2009) Grace has incorporated NF1 within her personhood. In her own words, NF1 has given her '*a very interesting insight into the world*'. The journey - or quest - of the acceptance of her own diagnosis that Grace has undergone, has given her a positive attitude towards life. She stated to have achieved a deep and more authentic conception of what is normal, what is desirable, and to be more able to value difference. Grace's narrative of illness and self-discovery is, as already shown, not only personal, but relational; it is strongly interrelated to familial dynamics. I would argue that for Grace not only accepting NF1, but also framing it as something interesting was functional in allowing her to properly exert her duty of the caring parent. This implies that both Grace's discourses of awareness and pride towards her NF1 were significantly linked to her son Jeff.

Grace presented herself as being a scrupulous and sensitive mother. She stated Jeff has been monitored and looked after by her and the healthcare system from the moment he was born. Grace also confirmed that she has always presented NF1 to the son in a positive and reassuring way. For instance, Grace said that when she brings Jeff to health check-ups, she always informs the doctors about her own diagnosis as well, in order to make Jeff feel more comfortable and to normalise the syndrome.

*Grace: whenever he goes to the doctor for anything I always say “Jeff has Neurofibromatosis” just in case... but I always make sure that I say “So do I”*

Performing this role of the caring and attentive mother would have been much more difficult if - in the first place - Grace herself had not incorporated her condition in her identity in a positive way. So her NF1 identity is meaningful as a result of her role as mother and familial interaction.

Upon closer examination, Grace’s acceptance of, and positive attitude towards, her NF1 appears to be a more manifold process which still presents vestiges of her teenage lack of consideration towards her diagnosis. In fact, whilst being very articulate in describing the health problems, the health management of her son and the concerns about his future, Grace seemed to be much less interested in her personal health. This pattern mirrors her concern towards the father and simultaneous rejection of her own condition which characterised the teenager Grace. Thus, also the mother-Grace who has accepted her own diagnosis for the son still presented instances of rejection towards her personal diagnosis.

In the first place, Grace expressed several times her worries about Jeff’s future; the two passages below represent just some examples:

*Grace: I have a waiting game with that...he may end up having all sorts of things might end up happening to him and with him*

*Grace: this phase of the experience is being a mother of a child who has NF who has gone through probably like...has navigated the first tricky period well,*

*he doesn't appear to have any problems, but he is approaching his adolescence, he may go through, he may have some problems during that*

At the same time, Grace rarely expressed concerns towards her own health. When some aspects related to her personal health incidentally surfaced during the interview, she tended to dismiss them very quickly. An example of this tendency can be found in this passage where Grace discussed the presence of UBOs in her brain (Unidentified Bright Objects: a symptom related to NF1).

*Grace: It's so funny isn't it, unidentified bright objects, what a strange... in a way that makes me laugh and that's something I feel I can say... that's an interesting thing about me, I have unidentified bright objects in my brain*

Considering the possibility that UBOs may be linked to cognitive difficulties she quickly and ironically dismissed the problem:

*Grace: where's the cognitive problem in me? I don't really understand it really... I know that umm... I do I mean people find me quite frustrating sometimes because I'm often late for things and I can sort of be a bit scatty or.. I find organising things quite difficult but how's that different from many other people? Or like my mum who's just...she hasn't got NF at all but she's like much more NFish in terms of like learning difficulty things...*

Grace's tendency to simultaneously foreground Jeff's health and dismiss her own was also confirmed by other family members. For instance, in this passage her sister Elise commented:

*Elise: Grace's concern is more related to her son than it would be to herself*

Grace's tendency to respond promptly to the symptoms of other family members, but to be at the same time less scrupulous in monitoring their health is in line with the idea of genetic responsibility discussed above (Hallowell 1999), and with previous studies on family and illness (Graham 1979). Graham (1979) for example observed that women (especially mothers) are more reluctant to be ill because their being healthy is functional to perform their role and responsibility of carers.

The acceptance and positive attitude towards NF1 deployed by Grace which incorporates foregrounding and backgrounding elements, also echoes her teenage mode of engagement with the condition, when Grace whilst dismissing her diagnosis was concerned about Benjamin's condition.

There are also other links between Grace's narrative of acceptance of NF1 and of Benjamin's experience with the syndrome. There seems to be a common perceptual-emotional pattern in the way Benjamin and subsequently Grace frame their condition.

With the expression 'perceptual-emotional pattern' I refer to certain styles of experiencing and interpreting phenomena; in this specific case a disease. These patterns can be mutually constructed and shared by different family members and can therefore be common within a household.

The important link between Grace and Benjamin will be specifically explored in the next section (§ 5.5) where I will show how the story of Benjamin has become a perceptual-emotional familial experience toward NF1 of all the family members interviewed.

## 5.5 Familial Reconstruction of Benjamin's Story

This section presents Benjamin's experience with NF1. Benjamin's story is reconstructed using the interwoven voices of all the other family members interviewed as he is an absent figure (he died about ten years ago of pneumonia). Benjamin occupied a central place in the familial experience with the disorder; below I will argue that he represented the familial archetype of the syndrome, and a remarkable influence over the experiences and actions of the other individuals in the family.

### Benjamin and NF1

Benjamin was considered to be the first person in the family with NF1. He was diagnosed only when he was a university student, in his 20s. The diagnosis took place in the hospital where Benjamin decided to go once he began to realize that the upper part of his body was becoming weaker.

*Paula: he was at University and he couldn't lift his briefcase up...like that then he couldn't lift his pint up to his mouth and so he went to the doctor and the doctor says 'oh well we must investigate this'*

On this occasion, Benjamin not only was diagnosed with NF1, but he also had to undergo the first of a long series of laminectomies (surgical operations of the spine) due to the presence of internal neurofibromas on his back.

As Grace vividly summarised, from that moment Benjamin started “*a kind of lifelong conversation with his body and illness [...] a relationship with his physical decline*”.

Benjamin represented a rare and quite serious case of NF1. Throughout his life he developed some external neurofibromas, but the main complications stemmed from the growth of internal neurofibromas in the spine, that made him become slowly quadriplegic. To remove some of these neurofibromas, he underwent many



laminectomies during the course of his life. He died when he was 57; therefore he was under medical care for three quarters of his life.

*Paula: ...his back was like a railway track...all the way down [...] seems all his life...his adult, well his... from his 20's onwards that he was either waiting for surgery, having it, or recovering from it*

Benjamin with “*sheer determination*” (Elise) fought his progressively debilitating condition and tried his best to lead a life which was as ‘normal’ as possible.

Given her medical background, Rosemary decided to become the full time carer of her husband. Over the years, despite the laminectomies, Benjamin’s upper and lower limbs continued to lose their sensitivity and motility. Nonetheless, he kept ignoring his debilitating condition and struggled to maintain his independence, continuing to exercise and walk, even years after he was already supposed to use a wheelchair. For instance, he continued going to work by himself, despite the serious problems that this generated.

*Samuel: ...I have heard sometimes he would go home and couldn't get inside the house... 'cause he couldn't get his keys out of his pocket and his wife wasn't there...and he used to...like when he was on the train he used to ask complete strangers to take his wallet out to take his card out for the ticket collector, 'cause he couldn't use his hands and get the things out of his pockets and so on, because he lost the use of his arm*

All the family members interviewed described with admiration and pride Benjamin's attitude and coping skills in fighting his condition; he was unanimously depicted as an "*interesting*", "*brave*" and "*inspirational*" man.

This was also due to the fact that, alongside fighting NF1 and the loss of his body, Benjamin also decided to get involved in very important national and international charity initiatives, as well as in raising money and promoting workability for individuals with impairments. Although being severely affected, Benjamin tended to ignore and downplay his NF1, even deploying a positive attitude towards it.

The familial account of Benjamin's illness experience encapsulates concepts which have been widely reported in the literature on illness narratives for diverse chronic conditions - namely- the idea of normalization of disease (Bury 2001) and the idea of fighting the disease.

Although chronic illness can disrupt a person's body and life itself (Bury 1982; Charmaz 1983), it can be construed in many ways and its impact can vary throughout their life. Lives of adult middle-class working people (like Benjamin) can be particularly disrupted (Bury 1988; Robinson 1988; Charmaz 1991); however, younger and middle aged people are also more keen to make efforts to manage their illness (Charmaz 2000).

Individuals often tend to normalise and reduce their symptoms for as long as possible (Johnson 1991; Stewart and Sullivan 1982; Charmaz 2000).

They may try to normalise illness by attempting to preserve their 'pre-illness' lifestyle and identity. This strategy can involve the maintenance of as many 'pre-illness'

activities as possible and can include the disguising or minimisation of symptoms in accounts and behaviours (Bury 2001).

Normalization discourses may also imply the integration of illness into the patient new lifestyle. In this case, illness is not ignored or hidden, but is represented as a constitutive part of the everyday life of the patient (Bury 1982; Kelleher 1988; Kelly 1992; Sanders Campbell et al 2007). Illness and the changes it can bring are made routine and treated as ordinary (Wiener 1984; Charmaz 2000b).

Fighting the illness refers to accounts in which illness is presented as a challenge that can give new meanings to the sufferer's life (Pinder 1988; Bury 1991; Williams 1993). The experience of fighting with a disorder can become an opportunity for 'biographical reinforcement' (Bury 1982) and lead to new 'reconstituted identities' (Charmaz 1983; Charmaz 1983; Herzlich and Pierret 1987; Radley 1993).

Benjamin's story blends elements of both normalization and fight. Despite the progressive impairment, he was still struggling to preserve his pre-illness lifestyle (for example by continuing to walk and go to work by himself). However, possibly also because the progressive impairment could not be completely ignored, Benjamin incorporated as well the illness into his life, transforming it into a personal challenge and into an opportunity to get involved in diverse enriching and socially valuable initiatives. This story of fight and enrichment can be seen as being reminiscent of Frank's quest narrative (1995).

Narrative analysis research has illustrated how people's ways of managing illness are influenced by sociocultural expectations and models (Frank 1995; Atkinson 1997;

Frank 1997; Thomas 2010). Our modalities of being healthy and sick mirror, and are modelled upon, ideological and social practices (Radley 1993).

Frank argues that in our society there is pressure for being ‘successfully ill’ (Frank 1997:117). Whilst restitution and quest narratives are very common and appreciated, chaos narratives – because of their lack of structure and ‘heavy’ content - are much more difficult to hear and can be even stigmatised or censored (Willing 2011). This social pressure towards reacting, fighting, making sense of diagnoses, and finding a meaning in illness is in line with our current neoliberal political climate which encourages health and productivity by placing emphasis on individual autonomy, control and empowerment (see Chapter 3). However, the expectation of being successful and in control even in the face of illness can cover up and simplify the situations of many patients and families.

Benjamin’s fight and rejection of his disabling condition seemed also to elicit feelings of anger and frustration in him. These feelings were described as being more pervasive as Benjamin’s health worsened and consequently it became very problematic for him to ignore and minimize his condition. This increasingly emerging ‘dark side’ of Benjamin’s attitude towards NF1 which follows the loss of his body is very clearly described in the following passages from Elise and Rosemary.

*Elise: he fought the decline of his disease, so it took a lot of effort for him to still walk, he was probably walking for 5, 6 years after they said there was no way he could walk. He was still climbing the stairs he was still he’d put so much energy into keeping those muscles working, ‘cause in his view keeping those muscles active would prevent it and you know and just by sheer determination, and I think that brought up a side of his personality that was quite explosive, because he had to put that sort of ferocious energy*

*Rosemary: from maybe the mid 80s when he had to go into hospital more often...to have operations and then got him out of and he'd go downhill quicker...coping with his upset over...to his mind, councils being terrible, not maintaining the pavements...so Benjamin fell over. Now, I knew it wasn't the councils. Benjamin couldn't lift his legs up and he was falling, he was blaming everything inside and he became very very aggressive*

Over the years, Benjamin's health worsened dramatically and it was no longer possible to surgically remove neurofibromas from his overly compromised spine. Benjamin became completely insensitive and paralysed from the neck downwards. His attempts to lead a 'normal', 'independent' life, refusing his NF1 and impairment were negated by the complete loss of his body. Although Benjamin continued to work very successfully (he was so important that his company taxied him to and from work) he had to become totally dependent on other people and needed fulltime daily care. Not only Rosemary and home carers, but also Elise, Grace and Tom had to look after Benjamin on a daily basis much more than before.

Nonetheless, Benjamin did not appear to be willing to accept his condition.

*Elise: he refused to accept that he was disabled, even when he was paralyzed to the neck downwards*

Benjamin's denial of his NF1 collided violently with the ineluctability of his impairment. Consequently, his "sheer determination" exploded in pure grief and anger which were described as become a prevailing trait of his personality.

*Elise: his character wasn't there anymore because it was blinded by pain, emotional pain 'cause he didn't feel physical pain, I suppose, which was a blessing*

Since Benjamin's body was paralysed, the voice remained the only vehicle to express his feelings. The anger and frustration towards himself and the world outside him was manifested almost exclusively within the familial household. Benjamin's sister Paula for example said she had never seen the "*nastiest side*" of Benjamin.

*Paula: you know, your nearest and dearest, you know you let them see the nastiest sides of you and maybe he let Rosemary see the nastiest side, whereas he didn't let us... 'cause I mean I would say things to my husband that I wouldn't dream to say to anybody else (laugh)*

Behind Benjamin's quest story, behind his fight and remarkable struggle to remain virtuous in the face of adversity (Williams 1993), lurked chaos.

Benjamin was putting an incredible amount of effort in trying not to be (and possibly look) too affected by his disorder and to maintain the role of the successfully ill person (Frank 1997). The familial household became the only 'safe' space where his suffering could be genuinely expressed. Letting out the chaos within the familial environment could be interpreted as being functional (or at least an aid) in maintain a strong identity, especially outside the household. But this chaos seriously affected the family life (McLean 2004; Smith and Sparkes 2008).

During this very critical phase that was described as having lasted for about five years, Benjamin lost not only his body, but also his family. In fact, Rosemary and the children found it increasingly impossible to cope with Benjamin's constant anger. They recalled

that they were overwhelmed by a mixture of sadness for Benjamin's poignant condition and of stark distress caused by his furious negation of it. Communicating and living with Benjamin became more and more arduous for his family members. The following domestic scene evoked by Elise (coloured by a retrospective sense of guilt and empathy towards her father), is a vivid snapshot of this process of stark isolation of Benjamin from his close family.

*Elise: So the poor man would come in from work, you know he struggled he'd still going up to town, still working, comes back, tries to you know, have a conversation with his family, but because anger was just boiling in him, we would be like "ah tititdi" and then we'd all be laughing, there'd be me, mum, Grace, Tom in the kitchen laughing, you know and we didn't even realize that we were doing it. And the poor man was sitting there on his own. So there was quite a... he got pushed out 'cause his anger just pushed lot of us away. So it was tough, it was tough for him, it was very tough*

Experiencing chronic illnesses can destabilise and alter patients' prior meanings (Charmaz 2000); challenging the way patients engage with and make sense of events in their daily lives. These new meanings may not be shared by others. Consequently, social interactions - most notably familial interactions - can become more difficult. For example, patients' calls for sympathy may be rejected by family members, whilst attempts to normalise their disorder may be considered inappropriate (Bury 1991; McLean 2004).

The wife and carer Rosemary found it profoundly difficult to cope with her husband's "very frightening personality". Her relationship with Benjamin was seriously

compromised up to the point that she had to leave him. An intense suffering and sense of guilt was expressed by Rosemary too in this passage about Benjamin's critical phase.

*Rosemary: he couldn't cope with his body...and he was a very upset aggressive angry man. It affected the children...after a certain level where Benjamin was becoming aggressive and it was upsetting for them. He would react quite horrendously towards friends being in, to them coming in at certain times of the day or night...he did become a very very frightening personality. It affected me as a wife, I have been carer and wife quite happily so, but I couldn't cope with the aggression and I did leave in the end...before he was dying...*

Whilst admitting to have experienced unbearable pain during this time, Rosemary appeared nonetheless to be sorry and to feel guilty for not having been able at the time to cope with Benjamin and for leaving him in a time of difficulty.

Although Benjamin remained alone, he was never completely abandoned by the family. Rosemary moved out, but continued for a few years to regularly check on his status helping, especially when the children and the home carers were not available.

In their study based on a series of in-depth interviews with an individual with spinal cord injury (SCI) who, like Benjamin, became completely paralysed, Smith and Sparkes draw on Frank's work to reflect on how in our western society we tend to avoid suffering bodies and the potentially tragic stories they carry with them (Smith and Sparkes 2008). Like Benjamin, the individual with SCI lost his family and at the time of the interview he was almost completely alone and severely depressed. Smith and Sparkes point out that certain types of illness stories which imply recovery (and thus a



happy ending) are more dominant and acceptable narratives in our culture (Frank 1995; Lupton 2003; Norrick 2005). On the contrary, when the possibility of recovery is effaced by the chronicity and severity of a disorder, affected individuals may bear the 'chaotic' marks of unavoidable and unbearable pain and suffering. Irrespective of the way they cope with their condition, their very presence may be received as being frightening or uncanny and their narratives may become less tellable and acceptable. Within this family, the potential disruption and sense of isolation caused by severe and chronic conditions do not simply pertain to the affected individual. As happened with Benjamin's family, it is also reflected in the experiences and lives of other family members who are related with the sufferer and thus also with their pain. The fact that family members may share this pain through their complex bonds, may represent an additional hindrance which can make the suffering individual and their narratives unbearable within the household.

Therefore, this chaos can trigger the emergence of self-perpetuating negative loops which can seriously poison family relations adding to the suffering of the affected person and her family members. Family members may be unable to deal with the ailments *and* the 'chaotic' coping mechanism of a loved one. In turn, the affected person may be unable to deal with their ailment *and* the reactions that other family members may have towards their 'chaotic' coping mechanism (McLean 2004; Smith and Sparkes 2008).

Benjamin's very lonely and depressed phase was overcome after he found a particularly good carer who helped him to accept and come to terms with his condition. The family members interviewed converged in claiming that this carer was particularly successful, especially because he was able to handle and block Benjamin's aggressiveness. The carer helped Benjamin accept his situation by also engaging him in diverse activities

from going to the pub, to jumping from airplanes, showing him that he could still have a life once he accepted his condition.

Benjamin's final 'reconciliation' with his NF1 and related quadriplegia was considered a crucial point of his experience with NF1. His family members converged in saying that, after accepting his impairment, he eventually got his life back and became able to deal with his body in a more serene way. In Grace's parlance, Benjamin experienced a "second wind in his life".

*Elise: because there was that anger and that anger sort of escalated the more his body deteriorated, until he finally accepted that, yes he was paralyzed from the neck downwards and actually went: "Ok! I'm in a chair! I need help!...Let's have fun!"*

The family found Benjamin again and also Rosemary was willing to come back and live with him, but, unfortunately, this could not happen.

*Rosemary: By the time he was actually dying I had said "look, I think you've gone through this phase, I still love you I want to remain your wife or come back into your household" but...he died so (laugh) that never came to being*

### **Familial Archetypes: Benjamin and the Familial Experience with NF1**

Benjamin's journey with NF1 interrelated in many significant ways with the family experience of the condition including both individuals with and without the syndrome. Benjamin seemed to have absorbed a considerable amount of energy and attention from his family members, whilst at the same time greatly inspiring them.

I would argue that Benjamin has strongly influenced the familial experience with NF1 becoming a perceptual-emotional familial archetype (i.e. model) of the syndrome. First of all, all the family members interviewed converged in claiming that Benjamin gained something very important from his illness; they positively integrated his experience with NF1 to his identity. In fact, it was repeatedly stated that Benjamin, as a result of his condition, became a more interesting and inspirational person. These passages reported below provide just some different examples of this link between Benjamin's identity and his illness experience:

*Grace: he was a tough kind of guy anyway but it really toughened him up and he was a very tenacious person, and I suspect that the reason he succeeded in his latter life was because of his kind of fight he had to go through*

*Elise: Oh I think...in certain aspects it's been very very positive, for dad, when he finally came around to finding himself again and finding joy and happiness. I think it created a man in him who was very very caring and very insightful. Because he had to get over the loss of body*

*Paula: how much admiration I have for him really, as a result of his condition*

Secondly, as I will show below, the tragic and inspirational story of Benjamin underpinned other family members' narratives of their personal experience with NF1, irrespective of whether they had a diagnosis or not.

### **Benjamin- Grace**

Grace's narrative arc of her relation with NF1 appears to display some significant similarities with Benjamin's case. Echoing the thrust of Benjamin's story, the adult mother Grace has positively integrated NF1 with her personality, considering it as something that makes her a more valuable and interesting person. Thus, following this common familial quest narrative, both Grace and Benjamin have been fortified by their illness experience.

There are some interesting structural similarities in the accounts of their experiences with NF1. Like her father, Grace did not accept her diagnosis and because of this she spent a critical period (although of a different intensity) until she came to terms with it, like Benjamin did.

Furthermore, Benjamin's denial and minimization of his condition could also represent part of the reasons which influenced the teenager Grace to adopt a similar behaviour towards her own diagnosis. These examples illustrate only some aspects of the deep relation between the figure of Benjamin and Grace's account of her personal experience with NF1.

### **Benjamin-Family**

The relevance of the figure of Benjamin is not only limited to Grace, but is also extended to the rest of the family as a whole. In fact, the positive link described above between Benjamin and Grace's identities and their illness experience was also absorbed by other family members and transformed into a collective familial illness identity narrative. The family as a whole has faced tragic and poignant periods because of (Benjamin's) NF1 without -though-being completely defeated. This idea which is reminiscent of Frank's quest narrative - is clearly expressed in the following passages from Grace and Elise.

*Grace: I feel proud that as a family we've had a challenge and we've all kind of stepped up to it (my emphasis)*

*Elise: we always knew that they loved us and that we were loved and that we would always be a family. They were very very strong in that, we just knew it, it was not even something that was questioned. Our parents were our friends and if we were in trouble, we knew we could turn to them and they would be there to support us. Which even going into everything that they went through, was I think quite an amazing fit really (my emphasis)*

*Elise: but we have always been quite a supportive family even though we've had anger and whatever. I think we have all been quite self-sufficient in a way, because we had this very strong face (my emphasis)*

The above quotations indicate that, according to Grace and Elise, traversing the difficulties generated by Benjamin's NF1 without completely succumbing, has revealed the strength of this family and at the same time it has fortified it. Benjamin's illness story with its negative and positive moments has become part of a collective familial experience i.e. part of the family's collective perceptual-emotional attitude towards NF1. This stands independent from how severely family members are affected and whether they have NF1 or not.

Elise for example, who does not have a NF1 diagnosis, has incorporated this familial NF1 quest narrative within her self; in the following passage she shows how it has influenced her personal attitude towards difference:

*Elise: because you look at life very differently [...] it changes your perspective and it gives you a far less judgmental outlook on society*

Looking at Benjamin's story in its entirety, his very angry isolated phase and his death can be seen as muddling or even undermining the quest narrative, shifting the balance more toward the realm of tragedy and chaos. Nevertheless, the familial reconstruction of the story is undoubtedly permeated by elements of quest. It is revealing that the family members interviewed describe Benjamin *and the family as a whole* as being overall enriched by the struggle and fight with his illness.

### **Benjamin-NF1**

Another meaningful aspect of the relevance of the figure of Benjamin is that the family members tended to deal with *his personal* NF1 and not with NF1 as such. In other words, in their account they tended to claim they did not experience the syndrome NF1, but *Benjamin* having the syndrome. This also implies that they did not seek to obtain a comprehensive medical genetic understanding for example of the overall nature of Benjamin's syndrome and its symptomatology. On the contrary, in line with what emerged in other studies presented in Chapter 3, the attitude adopted by the family members was more practical and limited to the management of the everyday and most pressing problems. In Benjamin's case, these problems were, for example, the surgical operations, the progressive quadriplegia and his anger stemming from the refusal of his condition. Other aspects of the syndrome did not seem to be generally taken into account, but became relevant only when necessary. This tendency to foreground Benjamin whilst backgrounding NF1 as such, surfaced abundantly within the accounts of all the family members interviewed. This is just an example from Rosemary:

*Rosemary: but that wasn't, again, it wasn't the disease as much as Benjamin with the disease and the way we worked through it*

In the following passage Rosemary made a further related important point:

*Rosemary: I personally didn't find it frightening, it was something Benjamin had got and we were going to have to work through it..... and I am glad I didn't have quite honestly all the backup, in inverted commas, because it would have probably made me a bit more scared than I was, I just took it as a disease he'd got and there was nothing we could do to rectify other than maybe keep him less stressed...which is an almost impossible task, ...and you would just live and hopefully do the right thing (my emphasis)*

Ignorance about the medical definition of NF1 was functional in keeping her calm enough to look after Benjamin. Concentrating on the daily instances of Benjamin's NF1- rather than getting information about the syndrome as such- made the task of looking after her husband easier. The fact that Rosemary had a medical background makes her statements even more meaningful.

A similar attitude towards NF1 appears to be also employed by Paula and Benjamin's parents. Talking about her knowledge of the syndrome, Paula states:

*Paula: I suppose in one way I have buried my head in the sand about it a bit, but ...yeah [...] I have never thought about that before...but I think now I am talking to you, perhaps I thought ...I don't wanna know too much about it because, it may be too hard for me to deal with*

Whilst possibly displaying signs of retrospective guilt towards Benjamin for not having sought all the relevant information on NF1, Paula admitted that keeping a certain degree of ignorance about the syndrome was an effective, if not necessary, coping strategy.

In the following passages, Paula explained how also her other sibling Mark and her parents preferred not to seek information on Benjamin's genetic syndrome in order to be able to better cope with him.

*Paula: perhaps we should all have thought about it [being more knowledgeable about NF1] more really...I think, but you know you are so drawn and look after him and help him, but you don't really wanna know too much about it, which is a bit...peculiar really (laugh)[...]mum and dad, were probably a bit like that probably they didn't want to know..[...] dad didn't like anything to do with medical things, he didn't want to know about blood, or conditions or anything...(laugh), mum would probably have been, more interested, but she didn't want to think...*

A more medically nuanced understanding of the condition was seen as unnecessary and potentially very harmful as it could scare the family members and decrease their ability to look after and support Benjamin.

### **NF1= Benjamin, but not Grace and Tom**

Finally, it is also worth noticing that the figure of Benjamin represented such a pervasive and important aspect of the familial experience that other individuals who actually had NF1 in the family, for example Grace and Tom, were always in the background and almost considered as not having the condition. This passage from Paula



where she referred to Grace and Tom as not having NF1, but only '*the potential*' or the '*possibility*' for the syndrome is very revealing:

*Paula: I believe that out of the 3 children...that Elise...they have all been tested and Elise hasn't got it at all...Tom could, has the possibility to have it, that's the son...and Grace...had the potential*

Probably, this discourse about the potentialities and potentials for NF1 is linked to the fact that both Grace and Tom have not presented with serious manifestations of the condition or with symptoms that are serious enough to be compared with Benjamin's. Not considering Grace and Tom as actually having NF1 could also be a consequence of the aforementioned coping strategy adopted with Benjamin, based on dealing with the everyday and most pressing problems and not on gaining a comprehensive understanding of the syndrome as such. In other words, since Grace and Tom have not manifested serious (enough) symptoms it may not be necessary to worry too much about their NF1, thus they can even pragmatically be considered as not having the syndrome. Therefore, family dynamics and not only medical genetic diagnoses can determine who is and who is not considered to be affected by a condition within the household. Furthermore, as the case of Benjamin shows, one individual in particular within the family can become the 'standard' archetype of the condition with which the other affected family members are compared. The passage below from Rosemary seems to follow this same idea. Referring to Grace and Tom she states

*Rosemary: I didn't have any worries about them, I wish, you know it'd have been nice if they didn't have a disease...but I... I always used to say to them you would be very very unlucky to have anything at the level dad had*

## 5.6 Concluding Remarks

In this chapter, I have delved into some aspects of the illness narratives of a family from the sample to present instances of how the family setting can significantly shape and influence individual responses to genetic knowledge and diseases.

I would argue that the family can represent a significant heuristic and experiential 'model'.

I coin the notion of 'family models', to refer to the mutual influence between familial dynamics and individual experience of self, disease and genetic knowledge. Family models can be viewed as a perceptual emotional template, which results in members of a certain family relating to a disease, to each other in relation to the disease and to 'the external world' in ways that are similar. The idea of family models helps to highlight the presence of familial meanings in the fabric of individual narratives about their disease and health management behaviours. Family models can influence which aspects of a genetic condition individuals let in and out of their illness experience and identity.

Biographical and other contextual reasons can lead families to consider certain symptoms of a genetic disorder over others (which may nonetheless be important from a medical point of view) and possibly to associate the syndrome mainly with these salient symptoms. Moreover, family models can influence the way medical information is received by individuals and family members (Graham 1979; Hallowell 1999; Featherstone, Atkinson et al. 2006). An example of this aspect can be an emotional-perceptual model that frames a condition like NF1 as "*interesting*". Using Frank's types I identified a common model of quest narrative in Grace and Benjamin's family.

As I have highlighted throughout this thesis (see for example Chapter 3), exploring family constructs of genetic disorders and knowledge can shed light on important aspects about disease experience and can ultimately benefit patients. Alongside the

notion of practical kinship, the idea of family models can represent a rich heuristic tool for exploring these aspects.

The analysis presented in this chapter offers also the possibility to further reflect on the notion of practical kinship (Featherstone, Atkinson et al. 2006) in the light of the ideas of chaos and tellability of illness narratives. Drawing on the work of Frank (1995) Smith and Sparkes, in the concluding reflections of their study mentioned above, state that a possible way for the individual with SCI to overcome his highly depressed and isolated condition would be to find somebody to whom he could safely and freely express his suffering and ‘chaotic’ feelings. They point out that in order to be transcended, chaos narratives need to be told (expressed) and listened to i.e. sufferers need to be granted narratibility. The authors observe that this may imply that, for instance, especially those who are in contact with this person with SCI (family members and, possibly, close friends) should have a higher tolerance towards his narratives and his state. People who care about this person with SCI should welcome his words and pain and acknowledge their existence and worthiness; by doing this, they would create a space for the possibility of subsequent changes of attitude in this affected person.

Smith and Sparkes’ paper can be read as a lucid invitation to increase- as individuals, family members and part of this society- our threshold of tolerance towards tragic illness narratives, namely, our capability to listen to and accept suffering and pain. Following in the wake of Frank’s exploration of illness narratives the authors acutely observe that ‘chaos narratives’ have also a lot to teach us (Frank 1995).

Benjamin’s family members, for example claimed to have learned a lot from their experience with Benjamin and NF1.

Hence, increasing the boundaries of tellability of illness narratives would certainly benefit patients, families and ultimately our society as a whole (Frank 1995; 2010). Nevertheless, there are diverse reasons that make the hearing of these stories very difficult. First, narratives of intense suffering may often lack coherence and a proper plot. Secondly, the narrative and the narrator demeanour may be very vivid, their bodies and words may be charged with intensely overwhelming knots of pain. Finally, suffering, disease, impairment are still viewed as taboo in many societies. Hence, those who are considered or consider themselves 'normal' may prefer to avoid contact with those who are in pain for many reasons, but principally because by avoiding the 'anomalous' (even when something is just slightly anomalous) they affirm and strengthen their 'normality' (Douglas 1966). Individuals tend to avoid be reminded about their their mortality and the fact they can easily become ill.

Unravelling and reflecting on the 'chaotic elements' in Benjamin's story highlights that within the familial household, it can be very difficult to manage diseases, precisely because family members, especially those who live in the same household, may be particularly vulnerable, as they are already linked by complex biological and affective bonds (McLean 2004). The notion of practical kinship then adds an important perspective to illness narratives and the idea of a threshold of tellability within the familial environment. The pre-existence of compound family relations may lower the threshold of tellability within families. In other words, family members may not be able to hear the untellable as it is part of their own personal stories too.

Moreover, individuals who become carers of affected family members may experience a mismatch between the status of the family prior to and after the occurrence of a serious disease. In a study on the experiences of families with HD, Williams et al. used the concept of 'cognitive dissonance' to describe this clash i.e. how the introduction of an

illness within the household can alter family roles and relations (Williams, Skirton et al. 2009). In this study, spousal carers reported to have experienced significant difficulties in adjusting to their new role and in accepting that their relation with their affected spouse was not based anymore on reciprocity, but on sheer dependence. The distressing psychological impact of cognitive dissonance could persist in time. In order to deal with this experience and to continue to be carers, family carers adopted diverse strategies, for example seeking comfort from selected family members or ‘falling out of love’ with their affected spouse (Williams, Skirton et al. 2009).

As shown with Benjamin’s family, not only the affected individuals, but also their family members may suffer because of the ailment and may not be able to listen to each other and live together. The family can be viewed as a first source of practical and emotional support, but at the same time it can also become a source of amplification of distress because of the very nature of family life (characterised by sharing space, emotions, problems, complex bonds).

These considerations about practical kinship, chaos narrative and the boundaries of tellability can support the idea (that will be further discussed in the next chapters, especially Chapter 8) that an available external source of help, like for instance a specialist advisor may benefit patients and families. Genetic disorders like NF1 pose problems that go beyond the medical management of the individual patient. Their effects extend to the family and household.

I would argue that it is important to offer the possibility to patients and families of expressing, if they have a need and wish to, their anxiety and suffering to an external advisor. This could be granted by a social community model of care which addresses the ill person’s situation -rather than only the symptoms and disease within the patient (Charmaz 2000).

The case of Benjamin's family is illustrative of this. Benjamin and his family manage to overcome their intense crisis period also due to the help of a specialist home carer. Raising our awareness of the complexity of familial relations can help us think about ways to make available more support that can better meet patients and family's needs.

## **6. NF1 Genetic Subjecthood**

### **6.1 Introduction**

This chapter presents the first part of the results of the analysis of patients and families interviews. It principally addresses the first research aim of this thesis, namely, how genetic knowledge influences individual experiences with NF1, how it can be used to define self, in interpersonal management and in structural support such as support groups.

In section 6.2 I will present and discuss patients' NF1 identity discourses and genetic subjecthood trying, where possible, to contextualise their accounts within the network of their familial dynamics and relations. The section 6.3 will further explore individual and familial implications of this syndrome by analysing participants' understanding of its inheritance and their reproductive behaviours. Finally, in section 6.4 I will tackle the broader collective implication of genetic information by examining participants' engagement with and views on NF1 genetic support groups, in particular the British Neurofibromatosis association, currently named Neuro Foundation.

The findings reported within this chapter have implications for the notions of genetic subjecthood, genetic responsibility and genetic citizenship explored in the literature review chapter (Chapter 3). In the concluding remarks of this chapter I will discuss some of these implications (§ 6.5).

## 6.2 Genetic Subjecthood

### Downplaying Attitudes

The vast majority of the interviewees, when asked about their experience with NF1, but also spontaneously during the course of the interviews, tended to reject invalidating NF1 identities, suggesting directly or indirectly that the syndrome did not influence their (and /or their family members') lives. On the contrary, they described their lives in as close to a normal manner as possible, conveying a sense of control over NF1 by putting the syndrome in perspective and considering it “*just an illness*”, “*nothing to worry about*”, “*just one of those things*” and something “*manageable*”, extending at the same time the state of ‘defectiveness’ and ‘imperfection’ to society as a whole. Rather than isolating and identifying themselves or their family as ill and/or with impairments, respondents deployed an attitude of acceptance of the fact that “nobody is perfect and every family and individual have or can experience problems sooner or later”.

Dylan was an adult who had NF1 with some cognitive, visual and motor complications. He was diagnosed when he was a child and since then he has needed periodical assistance; at the time of the interview he was working as a sales assistant and living with his parents. In the following passage (as in the whole interview) Dylan, whilst recognising the difficulties caused by NF1, minimised the overall impact that the condition has exerted on his life.

*Dylan: I think I live quite normally. I don't think about it every single day. I don't let it bother me, I don't let it affect me, I try and do the best I can every single day. It's just not something I think about every day. Obviously the visual impairment is something I live with every day and I know about.*

*With work it means I check it all a bit more carefully than my colleagues,  
but apart from that I don't let it affect me*

Tim had NF1 with bone lesions and his body was covered with some neurofibromas. Like other individuals with visible symptoms, Tim tended to minimize the cosmetic significance of his disorder by adopting a downplaying attitude *in context* (when facing society) as a way to manage actual or potential stigma.

*Tim: I've noticed some people, like if you're in a swimming pool and that, some people do glance at you, but I tend not to – I just shrug it off and carry on myself with what I'm doing*

### **Being fit**

Edward had NF1 with café-au-lait spots, learning difficulties, visual problems for which he underwent several operations, and some other mild physical and cognitive complications. He was diagnosed when he was in his early 30s. Many individuals in his family network- including his partner and two of his three children- had a NF1 diagnosis as well, presenting very variable physical and cognitive-behavioural degrees of severity. At the time of the interview, his partner Amber, who was found to have two internal neurofibromas in her head, was probably the most severely affected individual in the family. These neurofibromas were causing her serious headaches and were probably linked to other physical and cognitive difficulties she presented with. To monitor the development of the neurofibromas, Amber required at least one MRI every six months. Nevertheless, throughout his interview, Edward tended to minimise the impact that NF1 had had on his life and the life of his family; the following extract provides an example of this :



*Edward: we live a normal family life, there is nothing really different... [...] to be honest I don't really think about having NF [...] just a few patches in your arms, I am a lot better off on lot of people, that's the way I look at it*

Edward's discourses on his personal experience with NF1 were also particularly interesting. As the following passage shows, he appeared to downplay the influence that NF1 exerted on his life and identity by portraying himself as being a very fit and determined sportsman.

*Edward: when I first got diagnosed with NF, people said to me "oh you will never be able to do this, you will never be able to do that", if you know what I mean, this and that and things...and I was good to run marathons, I have run 48 marathons now so...I am pretty fit, so I think it is kind of rubbish when they say "you can't do that sort of stuff" which you can do. [...] When people got NF or any other sort of disability and people tell you "you can't do things" or "I don't think you'll be able to do thing" I think to myself "I can do it and I will do it"*

Edward's downplaying discourse appears to rest upon a common social belief that being fit presupposes being healthy. It also seems that keeping and deploying a fit image of the self, allowed Edward to resist the pressure exerted by other people who, drawing on another common belief about the impact of disease on one's life, were telling him that having a disease necessarily implies hindrances and limitation of possibilities.

## **Not knowing NF1**

For respondents who presented with less severe forms of NF1 than Dylan, Tim or Edward, it was probably even simpler to ‘distance’ themselves from their disorder, relegating it to the very background of their lives.

Martha was an adult woman who was diagnosed with NF1 when she was a child. She presented with a very mild form of NF1; she had café-au-lait spots and she developed one neurofibroma on her leg that was surgically removed. Throughout her interview Martha tended to downplay NF1, strongly emphasising how she had personally not been affected by the condition. Her answer- reported below- to the question about how she would explain NF1 to somebody is indicative of her downplaying discourse.

*Martha: I probably would have to research it a bit again, even though I know what I've got, and what it is, I probably would still have to read through my notes again to sort of know exactly what it is. I just know that it is a benign tumour that breaks off in the nerve endings that's probably...but I would reassure them to say like, I've managed to sort of lead a normal life and things, and that probably there isn't anything to worry about*

Martha portrayed herself as being distant from the disorder and as possessing a low level of knowledge and personal engagement with it. Martha's discourse provides an example of interpersonal management of her (illness) identity, aimed at presenting herself as being normal. This is -once again- indicative of the links between respondents' downplaying discourses and their impression management work.

Pamela, a young woman with some café-au-lait marks, a small subcutaneous neurofibroma on her hip and mild behavioural problems, did not attribute particular importance to her NF1, even forgetting the name of the condition.

*Pamela: It [NF1] don't bother me... don't mind it being this...yeah ... don't know the name how is it called? [...] because I forget what it's called, so I just called it...I just don't know how it's called so I just say "skin thing" ...*

In this passage and in similar ones of other respondents, the distancing from the disorder appears to also have the purpose of protecting from distress and upset related to it.

Pamela's way of referring to NF1 as "*skin thing*" is particularly meaningful as it was linked to the only aspect of the condition that appeared to bother her. In fact, because of the presence of café-au-lait spots on her body, she was unsure about whether she could have a tattoo. Notably, her concerns were not strictly related to the medical or psychological aspect of the condition.

*Pamela: ...there is one question, can I ask one question? [...] do you know if I can have a tattoo done on this skin condition thing?*

### **Family members' downplaying testimonies**

Downplaying discourses were also common among undiagnosed family members, irrespectively of the severity of the condition they experienced.

Alex, wife's, Nicole had NF1 and her body was completely covered by neurofibromas.

At the time of the interview they were both in their mid-50s. In describing his experience with the condition Alex stated:

*Alex: it is just something we live with, I mean, it's... I don't think either of us thinks about it very often...I don't think about it very much at all...*

Janice, the wife of Eric who had NF1 with some serious complications including a plexiform neurofibroma on one of his eyes, similarly stated:

*Janice: I don't think it affected me that much, it was just something, you know we were together and that was...you know it was just an illness isn't it?*

### **More articulated versions of downplaying discourses**

Other respondents provided more detailed and articulated versions of downplaying discourses. As already discussed above, Gabrielle's son Dylan had NF1 with some cognitive, visual and motor complications which required periodical monitoring and assistance. Gabrielle asserted the uniqueness of her son Dylan whilst rejecting the diagnostic label.

Furthermore, she stressed that keeping a light and positive attitude, focusing more on Dylan's general wellbeing than on his diagnosis, was an actively sought strategy that allowed the family to manage the condition on a daily basis. Moreover other important and practical familial aspects, like the presence of undiagnosed siblings, could also influence the day-to-day familial downplaying strategy:

*Gabrielle: you'd rather have children that didn't have NF, but if they did have NF then I think you can only try and look at it from a very positive point of view, and we try and do that. He [Dylan] was very young when he was diagnosed. We had another child as well who was incredibly active and was very noisy and always in the frame as well. So you've got two children to look after so you just need to get on with it and just...Dylan is Dylan and I would rather Dylan be thought of as Dylan than somebody with NF. How he feels and his wellbeing and his whole self - his confidence - is about him, and not about any diagnosis he might have. If that makes sense*

Gabrielle also gave to the familial downplaying strategy a deeper existential justification. She normalised NF1 reinterpreting the disorder as being simply one among the many different medical problems which *all* human beings encounter at some point in life.

*Gabrielle: as I have explained to Dylan on more than one occasion, nearly everyone in the world has something that needs attention medically and all of us are unique, and all of us have got different things to deal with, and we have to deal with them the best that we can*

Another interesting, although more atypical, example of illness identity discourse is the one provided by Grace. Grace's experience with NF1 was extremely varied. She had a mild form of NF1, whereas her father, for example, developed among other things internal neurofibromas in his spine that made him become quadriplegic (see Chapter 5). Whilst reinterpreting NF1 as being just an overt manifestation of "*the things that can go wrong in life*", Grace paradoxically argued that a NF1 diagnosis can be useful and thus serve a positive function.

*Grace: having awareness about the things that can go wrong in your life can be quite useful in terms of how you navigate your life or how you engage with our life. Because, instead of being these unknown potential problems, they're known potential problems*

The thrust of Grace's argument seems to be that diagnoses can be empowering, since they provide individuals with more information about the actual or potential symptoms and complications they may develop. Because of this amount of information about themselves, individuals who receive a diagnosis are then placed in a better position to

conduct their lives than ‘undiagnosed individuals’, whose medical present and future is more unknown.

Grace turned upside down the common belief that diseases limit the possibilities and somehow diminish the value of a person. On the contrary, the awareness of the limits of one’s ‘value’ is transformed into the recognition of the ‘value’ of these limits for one’s life. A practical corollary of her argument is that having a syndrome makes individuals more likely to be in the healthcare system (medicalized). This can be advantageous since it gives individuals (provided they can and want to follow medical recommendations) the possibility to have full regular annual health checks (e.g. blood pressure) and it is more likely that any health problem is individuated and treated promptly.

*Grace: one other good thing about having a syndrome that you know about means that you’re more likely to be in the healthcare system anyway, so I’m very used to going to doctors and if I have a problem, I’m going to go see a doctor, and so as part of my process of being in the system, it means I get my blood pressure checked, I get this checked., any other normal problems that would be happening would be picked up nice and early, because of the process of having a full healthcare check once a year*

## **Problems**

Data analysis also highlighted interesting links between downplaying discourses and the experience and management of the uncertainty of NF1. As it will be further discussed in the ‘Uncertainty and Family’ section in the next chapter, the unpredictability of NF1 represented a significant source of concern for individuals and families. Normalising NF1, concentrating on the positive or more urgent aspects of daily lives or not thinking

about the syndrome figured among the diverse downplaying practices employed by respondents to manage these concerns. Thus, feelings of worry and the disposition to downplay NF1 could be intertwined in respondents' discourses.

### **Practical Utility of Downplaying**

Avoiding thinking about present and future health problems could be also justified by respondents as a necessary life strategy to keep a job assuring their own or their family's economic stability.

One of the most illustrative cases was that of Julia's, a widow and a mother of two children, one of whom was going to start at University. Julia had NF1 with some serious complications including neurofibromas, scoliosis and pseudoarthrosis which were causing her, among other things, pain and discomfort. Knowing that she was required to have regular monitoring- since she could develop further complications and possibly to undergo surgical operations- she preferred, nevertheless, to avoid thinking about her health condition as much as possible. Although being worried about her health and possible complications, she explained she could not afford to take many days of absence from work to attend all the hospital appointments or undergo surgical operations and that she preferred to rely on tablets to manage her pain on a daily basis. In her parlance, downplaying NF1 was functional to her need to find "*the fine balance*" i.e. a compromise between taking care of herself as well as taking care of her family.

*Julia: there are some things that can be quite, you know, like tumours and stuff that you can get, sometimes it's best not to know... I don't want anything sort of down myself, and I thought I've got the bumps and lumps going in the spine, but if I couldn't sort of maintain, sort of pain management programme, 'cause I mean it's sort of finding the fine balance really...ok there might be*

*some sort of surgical options, but at the end of the day it's a risk to go through that and it's easier just when I've got to work, because I am a widow, just sort of taking the tablets! You know and just maintaining a sort of lifestyle and being able to work, because obviously I have to work*

Moreover, Julia vividly highlighted that downplaying NF1 and keeping a positive attitude was necessary in order to be accepted by the rest of society and avoid isolation.

The following passage from her interview provides a further example of how respondents' downplaying attitudes can be triggered by social contexts and be functional to their impression management work.

*Julia: you just got to make the best of it, no good wallowing in self-pity, because at the end of the day you just like piss people off sort of, excuse my language...it does because they say 'oh gosh...' I just got to sort of think what I have got and I haven't*

### **Downplaying & unpredictability**

In different contexts, for example vis-à-vis the actual uncertainty and unpredictability of NF1, respondents' downplaying strategies were undermined and, thereby, the syndrome could be foregrounded.

Seth's son, Adrian, was found to have NF1 in his early childhood; during his life he presented with dyslexia and external neurofibromas. He died in his early 30s, straight after the sudden occurrence of a malignant triton tumour (MTT), a rare complication of NF1. Seth highlighted several times the lack of consideration given by the son and the family to NF1, neurofibromas and dyslexia notwithstanding, until the appearance of the MTT.



*Seth: well at that time...there wasn't very much experience...he [Adrian] ...he had this things around his waist around his chest, but it didn't seem to interfere with his life very much.[...] Until he developed one on his neck causing a lot of pain we really didn't pay a great deal of attention to it. Frankly. [...] You know, from my son's point of view until he got this thing on his neck he just assume he would have had a minor inconvenience that would go with him, but that it wouldn't be any more than a minor inconvenience*

In talking about Adrian's experience with NF1, her sister Wendy made similar observations. Seth and Wendy's accounts represent a vivid testimony of the potential conflictual interrelations between downplaying practices and uncertainty:

*Wendy: He just didn't bother about it at all and I thought "good on you really". But it wasn't until when it actually got on his nerves that he bothered doing anything about it, sort of things until it sort of got in the way. [...] I definitely saw lumps, they got a lot worse as he got older because they weren't any that noticeable they were almost sudden. It sort of got bigger*

Acute anxiety towards past, present and future NF1 complications is intertwined, in John's interview, with timid attempts to manage and downplay the condition. At the time of the interview, John was a man with NF1 in his late 50s. Since adolescence, his face and body had been increasingly covered by external neurofibromas. John recounted that the cosmetic implications of this symptomatology, alongside the direct experience of stigmatising behaviours, had left vivid psychological marks, leading him to have periods of serious depression and suicidal thoughts. Despite his attempts to "forget" about his NF1, focusing for example on the positive events in his life, the syndrome was always there "at the back of his mind".

*John: in the earlier stages it just made you feel down, depressed but...life goes on you just got that will to live and that's what keeps you going and that's it and that's it really, it's just like another day tomorrow so you gonna earn another loaf and bread sort of thing...[...] you fight it all the time...and then if things are going good for you, you forget it and life carries on you just completely forget it, but at the back of your mind it's always there[...] I quite often worry, should a lump block off an artery? And it stops the blood from going wherever you know that worries me, since it got worse over the years*

Within Claire's family NF1 "is there all the time" or, in her husband's (Tim) words, "you can't go away from it". In Claire's account, the experience of cancer amplified the presence of NF1 dramatically, making its unpredictability a source of constant concern and anxieties for the whole family. Claire's son, Leonard, was a teenager with NF1 who was suffering from serious complications, among which was malignancy. The physical and emotional impact exerted by NF1 had seriously compromised the functionality and relations of the whole family which at the time of the interview was also undergoing psychological therapy. In Claire's quotations reported below, the uncertainty related to NF1 and malignancy in particular is so significant that it seems to leave no space for happiness and serenity within the family life.

*Claire: Yes, it's a concern all the time, yeah – it's just – because you know it's there all the time, and you've just got to live; just go through and do what you got to do, especially with this diagnosis as well; all the time something is there. You can't feel – like before – yeah, happy, yeah – but now you're scared. One thing, when we got told it was ok, we were partly scared because we know something else might happen. My daughter was worried about that and then*

*what happened was it did; it turned out he had another lump, like, you know.*

*We're just wondering all the time now what's going to happen next*

Therefore, there were very different examples of foregrounding among respondents. In particular, acute episodes (e.g. malignancy) *forced* individuals to engage with their genetic diagnosis or at least temporarily foreground it.

### **Familial Contextualization of Downplaying**

There were cases in which the testimony of family members directly or indirectly questioned the downplaying practices of affected individuals. Eric had NF1 with some serious complications among which was a plexiform neurofibromas on his eye. In discussing whether NF1 had influenced his life he stated:

*Eric: it hasn't at all, I have not let it into my life I've tried to ignore it as much as I possibly can...and just get on with life as it comes [...] I have accepted and don't pay any real attention to it*

Throughout his interview Eric, having mentioned the medical problems related to his NF1, tended to downplay its cosmetic and psychological significance, presenting himself as serene and confident as possible. On another occasion he also affirmed:

*Eric: if you got NF and other people don't like it, they've got the problem, you haven't*

However, the subsequent interview with his wife Janice brought into light new aspects of Eric's experience with NF1 which gave more depth to his past and present relation with the condition:

*Janice: Eric was very very self-conscious...about meeting people, and I think ...he went, when we were married first , if any of my family came into the room or anything he ...disappear<sup>17</sup> ... like I remember, because I think he was just self-conscious...but I think he is not very good at making friends...because I think that's one of the things [...] I think gradually because my family being... would support him really 'cause they haven't taken any notice of it, he was just one of the family and that's it...but I think when you are in the shop sometimes...and you see children looking at him and things like that, but I don't know whether he is noticing them, or not , but one time he would and then disappeared*

A similar dynamic is echoed in the confrontation of Edward's downplaying discourses towards his NF1 and the perception of his mother Paige:

*Edward: it [NF1] didn't bother me to be honest...I knew it wasn't anything dangerous and nothing like that [...] it doesn't bother me, I can do what everyone else does so*

*Paige: Edward has difficulties with coordination he has difficulties and he is frustrated when he can't do things and, he had problems at school*

It is interesting to compare Edward's depiction of himself as a capable person not limited by his NF1 with Paige's description of his school, coordination problems and his frustrations.

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<sup>17</sup> After interviewing Eric and Janice, Janice kindly offered me tea and biscuits. Both of us were dining at the table while Eric was standing by the window of the adjacent living room a bit far from us, looking outside. Eric's behaviour seemed to mirror Janice's description captured in the above passage. Eric joined us only after my direct invitation.

Downplaying attitudes are also problematized in Wendy's account of her brother Adrian's experience with NF1

*Wendy: He was trying to be very confident on the outside, but for me he wasn't really, so he was not as confident as he came over. He bluffed it well; he joked about the way he looked*

These passages provide a further example of the richness of data that can be obtained by exploring family networks. As already mentioned in the methodology chapter, collecting the voices of different family members allows for a better contextualization of participants' accounts and, consequently, to consider more details and contradictions in their discourses. Therefore, the familial contextualization can strengthen the analytic process, helping the researcher to go beyond mere description and rationalization of participants' discourses. In this case the family method approach has allowed us to explore how each family member can have a different experience and perspective of the same issue.

### **6.3 Genetic Responsibility**

Although participants were not directly asked about the inheritance of NF1 and their reproductive decisions, these topics emerged as very common sources of concern for individuals with NF1 and their family members independently from the severity of the condition and whether there was a family history. All the respondents displayed, in different modalities and degrees, anxiety towards the possibility of younger or future generations inheriting the syndrome. Expressing concern for the health of future children was commonly presented as a moral parental duty of thinking about what is right for the child.

Inheritance and reproductive decisions, alongside uncertainty, were important aspects of the experience of NF1 of patients and family members. These were among the principal contexts that made NF1 salient, leading individuals to (temporarily) abandon their downplaying discourses and engage with the genetic nature of NF1.

Martha was a mother with a very mild form of NF1. Passages like hers, in which individuals were minimizing the impact that NF1 had had on their lives, but expressing at the same time some form of concern over younger generations in their family presenting the condition, were extremely frequent. This familial dynamic was shown both from respondents with and without NF1 and independently from the severity experienced.

*Martha: the only thing I really worry about is my girls like having it [...] but other than that really to be honest, it hasn't... I mean I've had one little lump removed at the back of my legs, but it hasn't really bothered me, it hasn't stopped me from doing anything or whatever*

Alice did not have a NF1 diagnosis, while her brother Robert had the condition with some physical complications and mild cognitive-behavioural difficulties. She similarly stated:

*Alice: I felt initially afraid of having my own children, you know, would that skip to them, but I have been reassured that's not going to happen...so, actually, really it is not something I think about a lot, to be honest*

Like Alice, many other respondents, also expressed some form of apprehension towards the possibility and the implications of their NF1 family members having children.

*Alice: he [Robert] has got a daughter...and she's been tested and she is alright, so that had been a big fear*

Paige had a very mild form of NF1. During her interview she admitted to having been worried about the health of her future grandchild, due to the fact that both her son Edward and his partner Amber had NF1. The fact that both parents had the disorder seemed to be a factor that increased her worry.

*Paige: Amber, my son's partner, she has NF, Edward has NF...now when Amber fell pregnant we were really worried that the baby would be badly affected with NF, but she hasn't got any sign of NF at all*

Ethan, in talking about the possibility of his son with NF1- Dylan- having children, clearly explained the familial relevance and implications of the inheritance aspects of the disorder.

*Ethan: I think the genetic side of it is a fact – it's likely to be passed on, so obviously if Dylan ever did marry, there's no doubt about it, he's going to have to have some genetic counselling to help pave the way really, and that's determined by whether he has children or not. Dylan's made it quite clear he's not interested, but you never know. You never know what comes along, so the genetic side revolves very much around that, and it's actually the future of the family tree if you like*

Inheritance and reproductive aspects of NF1 were not only important threads in the fabric of individual and familial experiences with the disorder, but they significantly permeated and influenced respondents' courses of action.

Some respondents could start engaging (or engaging more significantly) with their own condition and enter into the medical system because of the event of parenthood. In line with the idea of genetic responsibility conceived as a familial relational experience, the experience of parenthood could in fact represent a point in individuals' lifecourses in which NF1 became salient. Similar dynamics could also be shown by family members of individuals with NF1. Wendy did not have a NF1 diagnosis and was the sister of Adrian who died prematurely in his 30s from serious NF1 complications. In the passage below, she vividly described her experience with the disorder, showing again how genetic risk could become salient in relation to reproductive decisions.

*Wendy: you know...it's really alarming to hear about the family, it's kind of wow, I was kind of frightened actually, so I suppose I shied away from it a bit a sort of, you know ignorance is a bliss kind of thing, but I got pregnant and I thought kind of I might pass it on to my children...*

It is interesting to point out that Wendy appeared to be worried enough about her skin pigmentation to refer herself to healthcare specialists only as a result of her pregnancy, but not earlier in her life or for example after her brother died because of NF1.

*Wendy: I was worried that I had it [NF1] because I have the brown marks so I was worried about me passing on, maybe to my children. So I asked to be seen, I told my midwife and I got seen*

The awareness of having a condition that is inheritable could also significantly influence the reproductive decisions, marital life and sexual relationships of individuals with NF1, potentially impacting as well the lives of their partners.



As already mentioned above, Nicole was a woman in her mid-50s whose body and face were completely covered with neurofibromas. Nicole discovered that she had NF1 after the growth of the first neurofibromas when she was a teenager; it appeared very likely she was a de novo mutation in the family. In her personal account about her experience with NF1, the event of the diagnosis coincided with her inflexible choice not to have children.

*Nicole: It was when we were undoing our bicycles, when my mother said to me “when you’re thinking of having children, you must go back and talk to him” ... I knew then, it’s genetic and there’s no way I’m passing this on. That was at 16, I made a decision there and then... I can still remember as I’m unlocking my bicycle lock from the railings outside the hospital. I made the decisions there and then, no children; that means no children...*

As Nicole highlighted below, her decision of not having children had significant implications on her marital life:

*Nicole: I had to be very careful about the man I chose to get married to. I was dead lucky, I found a really good one... but, I mean, it had to be a man who particularly did not want children.*

However, Nicole’s statements notwithstanding, her husband Alex appeared to be quite affected by the absence of children in his life. In fact, several times throughout his interview, Alex mentioned their reproductive decisions, clearly stating that renouncing the possibility of having a family was the most difficult aspect of his experience with NF1.

*D: what do you find are or have been the most difficult aspects of your life in relation to NF?*

*Alex: well it's probably the decision whether or not to have a family! Really that's the fundamental thing about NF...*

In two families individuals with NF1 underwent surgical procedures like vasectomies to prevent the birth of other children with NF1.

Claire and Tim had two children one of which, Leonard, with a serious NF1 profile.

After Leonard's diagnosis, Tim was discovered to have NF1 as well and – consequently - to have passed it to the son. Knowing about the inheritability of NF1, Tim decided to undergo a vasectomy to avoid having other children with the condition.

*Claire: we thought, well, two's enough anyway, and we didn't want to risk anybody else having to have the NF like, you know, [...] my husband's had the snip to stop us having any more*

In line with parental discourses of genetic responsibility (Raspberry and Skinner 2011), both Claire and Tim stressed in their interviews to be glad and grateful for having Leonard, but Claire also mentioned her husband could not avoid feeling responsible and guilty for being the cause of their son's NF1.

*Claire: my husband [...] he passed it on to Leonard, he feels guilty for it, even though I tell him he's not – you know it's not his fault. It's just one of those things*

John recognised as well in his feelings of guilt and responsibility for passing on NF1 to his children the main reason for his choice to undergo a vasectomy. NF1 was quite

widespread in John's family; his brother and children presented with different manifestations of the condition and at the time of the interview his first newly born grandchild was still undiagnosed. John, whose body and face were completely covered by neurofibromas, represented the most serious case in the family.

*John: I wasn't responsible for me brother was I? So we are both like it but in fact I had a vasectomy for that reason. I mean I love me children and I have three children and the main reason for having a vasectomy, is not because I didn't want any more children, I didn't want no more children coming into the world with what I have got...and it took me a long time to realise, it took me a long time being a bit thick (laugh) it took me a long time to realise 'bloody hell it's me that's passing on this....this disease!' and on those grounds that's why I went to my GP. I would have been quite happy to have a couple more of kids, I mean I love my children, but that wasn't, I thought, I seriously thought about it: "bloody hell it's me that's passing these genes down to my...grandchildren or my children"*

It is interesting to observe that respondents' reproductive behaviours could also be interpreted as attempts to establish some control over the uncertainty of NF1. Choices like Nicole and Alex's not to have children or John and Tim's decision to undergo vasectomies might be indicative of their attempts to reaffirm their agency and gain some control over the unpredictable nature of the condition, or also to control it in future generations (Petersen 2006; Kelly 2009; Raspberry and Skinner 2011).

## **Ambivalent Use of the Variability of NF1**

The extreme variability of NF1 appeared to be ambivalently interpreted by different respondents as a factor that could justify the decision of having or not having children. Some individuals tended to characterise NF1 as being an overall mild condition which would rarely cause problems to future offspring. On the other hand, for other respondents the fact that NF1 could potentially cause serious complications represented a sufficient reason for not having children. Whether the emphasis was put on the mild or the extreme end of the variability spectrum was independent from the severity of NF1 experienced by individuals and their family members.

Nicole and Alex gave articulate accounts of how the possibility that NF1 could cause severe physical and cognitive complications, justified their choice of not having children. They mainly drew upon their previous experience as workers for the NF Association which allowed them to meet many individuals with NF1 and consequently be directly exposed to different degrees of severity of the condition, including the most extreme end of the spectrum.

*Nicole: It [working with other people with NF1] confirmed my decision not to have children as being absolutely the right one...because I've known people who haven't known they've got it... to people who've died from it, people who are very severely deformed, the face or the leg, people who've got lumps and bumps, and... people who were blind with it, so... children with problems, learning problems, 60% they say, it's very high... behavioural learning problems [...]so having seen all that, now I think "well thank God I didn't have children"... that's all I can think now... because I don't think I could have coped with having a child with NF who's severely affected and knowing that I've given it to that child... I'm very glad I didn't...*

*Alex: Through the association we have both seen people who were severely affected by it in one way or another and we were very glad Nicole wasn't this way and our decision not to have children, we are quite happy with it, because we could have brought a severely handicapped child into the world and perhaps that wasn't the right thing to do*

Nicole assigned so much importance to the negative aspects of the variability of NF1 to even advise other individuals with the disorder who were facing reproductive dilemmas to “*know the worst*” before making their decisions

*Nicole: I could remember people when I was in the West group down here... and people used to say to me “I don't know whether to have children or not” and I would say “well it's your decision [...] find out everything about NF, know the worst before you make the decision, you need to know what it can do...” and that's what I used to say to them, and I said “I'm not advising you not to have children, that's your decision, but know the worst, know what it could do and then make the decision”*

Paige had a very mild form of NF1, but a significant part of her family network presented with the disorder in much more severe forms. Her son and daughter in law, for example, suffered from diverse physical and cognitive complications. Paige claimed that potential parents should not fear having a child with NF1. Differently from Nicole and Alex, she supported her argument emphasising the mild end of the spectrum of variability of NF1 suggesting that NF1 is prevalently a mild condition:

*Paige: as far as I know, [NF1] doesn't severely affect a child who knows...I think I mean they are affected...but nothing that they can't have a good life*

Moreover, Paige suggested that meeting other people with NF1 could help potential

parents to understand how normal the condition typically is.

*Paige: it should be explained to them [potential parents of children with NF1], you know, that... or meet people with NF1 so that they know that that's not a severe...can be severe but on 9 times out of 10 it is not severe*

Paige's case is of particular interest because she also stated, as shown above, to have been worried about the health of her future grandchild after she discovered her daughter in law was pregnant.

### **Reproductive Technologies**

In Chapter 2 it has been observed that although prenatal molecular genetic testing (PND) and pre-implantation diagnosis (PGD) for NF1 are available for families in which the mutation has been identified, they are rarely employed in clinical settings. This seems to be mainly dependent on the variable and unpredictable nature of the condition as well as the reliability and cost of these technologies. Similarly, genetic testing is infrequently used as a diagnostic tool and individuals are normally diagnosed relying on the clinical diagnostic criteria.

Reflecting the scarce utilization of these molecular diagnostic tools in medical settings, all the respondents with NF1 recounted how they were diagnosed clinically and none of them employed any form of prenatal testing for the disorder. Notably, the vast majority of respondents, when asked about their view on new reproductive technologies (PND and PGD) often demonstrated vague or no knowledge of these technologies. Their views towards the idea of prenatal genetic testing appeared often to be unclear.

Only one individual expressed a clear cut positive attitude towards PGD and PND, considering termination in the eventuality of a positive result for PND to be advisable.

This was the case of Seth whose son Adam died (as discussed above) of malignancy and was the only person with NF1 in the family.

*Seth: Well...if somebody has it any child has a 50% chance of having it and it would seem sensible for somebody with the disease to have genetic testing; at the appropriate stage to ensure any children didn't have it. Whatever stage that was whether that was using pre-implantation or early abortion, but it does not seem wise to have children with it... to me. I think others would disagree with me on that!*

Interestingly, Seth's daughter-in-law (Adam's wife) had a child after Adam's death through IVF using the semen which Adam had previously deposited in a sperm bank and she decided not to use PGD.

*Seth: my daughter in law had a child using sperm from the sperm bank that my son had left before he started his cancer treatment and she didn't have any test, she simply said, I'll take my chance. And the child fortunately didn't have NF*

Almost half of the sample displayed anti termination and abortion positions, explicitly claiming they were not interested in prenatal testing; this group was predominantly composed of women and those who had experience of mild to severe forms of NF1. The line of argument of respondents within this group was based on reckoning NF1 to be overall a mild condition and considering prenatal testing to be driven by an increasing societal pressure to have perfect babies (in other words to be a form of 'eugenic normalization') or regarding abortion to be ethically wrong in itself.

*Julia: if testing would have been available I wouldn't have had it, because....when you think about it these days it's all like to make a perfect race, and everybody's got to be perfect, but you look at like in history the genius, like Mozart he had some sort of things, Beethoven had got something wrong with him and several other sort of really famous people had got genetic disorders or these days disorders that would be screened and that you could... and it is such a waste, you know, you still got a lot to offer and sometimes it goes too far with all this screening*

*Alice: this is a very ethical question. Because it wouldn't make any difference to me if I was pregnant already, what kind of deformities my child might have, because I wouldn't agree with abortion, so I'll continue with the pregnancy and have the baby, so I wouldn't have a test like this*

The remaining individuals in the sample either found it too difficult to have a clear opinion on prenatal testing or did not tackle the issue at all. This second group was equally composed of both genders and experienced as well mild to severe forms of NF1. A few individuals from this second group expressed the opinion that PND could be useful for allowing parents, in case of a positive result, to prepare for the birth of a child with NF1.

*Martha: if it was sort of like me, I would be reassured and made me then decide what my choice would be, so I do think that's a good idea now, [...] and I think if you decide then to go on and have it and it has got and at least you are prepared and it's not gonna be a shock when you have the baby*



Some individuals across these two groups seemed to be inclined to regard artificial insemination and PGD as a less morally problematic option than PND.

*Nora: I feel that if we were that naive, right, we wouldn't eat eggs, because we'd be eating baby chickens. Right? I feel that an embryo is an egg, isn't it? Before it's been fertilised it's an egg. And we lose eggs every month, I mean, hundreds and thousands of them. You don't think, "Oh, my baby's gone!" do you?... when you have a period? So I think that's quite a good thing*

Others were inclined to view reproductive genetic technologies as a possible way to eliminate NF1. Considering possible treatments for the condition Charles said:

*Charles: make it so that it is not passed on, if you know what I mean...sort of genetics, it's a genetic disorder it's something is wrong it's not passed on. Something is wrong with the DNA so passed on sort of thing making so that that doesn't happen*

Eric expressed a similar idea, but he also observed that genetic reproductive technologies would not eliminate NF1, due to the possibility of de novo or spontaneous mutations.

*Eric: preventing NF baby to be born would be one way to cure NF, but it is a genetic default, so it possible for it to spontaneously occur again in the future*

## **6.4 Genetic Citizenship and Support Groups**

As stated in Chapter 3, investigating whether NF1 is associated with a specific identity and community based on genetic knowledge, can help in the understanding the role of

genetic knowledge in individual and familial experiences with NF1. For this reason, respondents were asked about their relation with NF1 support groups. The experiences reported with NF1 support groups are related to the UK NFA in Kingston Upon Thames and its branch in Devon which used to exist up to a few years before the interviews were conducted.

### **Positive Accounts of UK NFA**

There were a few respondents who had some direct contact with the NFA and talked about the support group in positive terms. They and/or their family members all had NF1 with severe symptomatology.

Eric had NF1 with some serious complications including a plexiform neurofibroma on one in his eyes. He was a member of the NFA and actively participated for a few years before the time of interview in activities organised by the association. In the following extract, drawing upon his experience, he described how the association helped different individuals with NF1 providing a space for interaction and representing a point of reference for emotional, psychological and medical support.

*Eric: that was good, because you could go there talk to other people and encourage them, because other people seem to be affected far more, and it seems to worry other people far more, and you give them an encouragement. [...] If they met people with the same problem as well people realise they are not alone and there is support out there if they want it [...] they could call, you could go and tell them where to go to get expert advice....sometimes they just wanted to talk; you talked to them and if they wanted I could tell them where to go to get medical advice*

Along the same line, Julia emphasised how by joining the NFA she had the chance to share her personal experience with pseudoarthrosis with somebody else presenting the same symptom.

*Julia: this is why having the support group is good because when I joined I... someone rang me out of the blue and they said “oh there’s a girl with pseudoarthrosis, do you mind talking to her?” and I said “oh that’s fine!” and we kept in contact for a few years [...]so she said it was very very helpful to talk to me. So I think that’s always good, when you’ve got the support of a group that you can then talk through your experiences and say “well that worked for me, that didn’t...have a think about...”*

Vera praised the aid received in the past from a NF advisor to help her grandson Douglas, who had NF1 and serious cognitive and behavioural difficulties, to obtain support at school:

*Vera: she [the NF advisor] was fantastic!...when Douglas started school first and he was getting problems, she was fantastic about going into the school and coordination with the school...she managed a little bit more than what we did, because it was like it was Douglas [...], but she [the NF advisor] understood it more and also maybe that’s because she was... had something, because she was...maybe more an authoritative figure?*

Charles who avowed not to “*have many friends*” and to normally “*finding it hard to talk about things*”, recalled participating in a NFA camp,<sup>18</sup> stating that on that occasion

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<sup>18</sup> NF camps are events (that can last for a few days) organised by the NF Foundation which provide a space for NF1 patients and families to meet and share experiences. Often these events host also research

he found “*it was sort of easier*” to relate to people. Charles had a serious NF1 profile presenting, among other things, cutaneous and plexiform neurofibromas as well as cognitive difficulties.

Joining one NFA camp utterly changed Edward and Amber’s life. Both of them presented diverse physical and cognitive symptoms. They both described having met there for the first time and subsequently deciding to get married. Notably, in this case a shared disease identity strongly influenced the lives of two people and their family; as Edward repeatedly emphasised, it was NF1 that put Amber and he together:

*Edward: I met Amber up in Scotland in a way; NF put Amber and me together because you know [...] I went up and that’s where I met Amber! If I didn’t go there I’d never have met Amber! So NF put us together*

*Amber- I met my husband on....in Scotland in an adult camp and ever since...  
[...] that’s the best thing (laugh)*

### **Lack of Support**

With the only exception of the positive instances just presented, the testimonies provided by the vast majority of respondents (including sometimes the same ones who reported the positive experiences) included a gamut of different elements that pose serious challenges to the actual and potential existence of a biosociality or biocitizenship for NF1.

A fifth of the respondents were not from Devon, but from Northern Ireland, Cornwall and Greater London. The participant from Northern Ireland was the only one in this

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meetings where leading NF clinicians and scientists illustrate the latest advances on the understanding of the syndrome.

non-Devonian group to explicitly complain about the lack of any medical and non-medical support around NF1.

The majority of the remaining participants, resident in Devon, highlighted the lack of local support for patients with NF1 and their family both in the past and at present.

Alice's family was from Northern Ireland and still living there. Alice repeatedly stressed that in that part of the country there was (and is) not support group available for her brother Robert who has NF1 nor for the rest of the family.

*Alice: apart from the consultants in the hospital. There's never been a support group*

*D: Still at the moment? Do you think nothing has changed?*

*Alice: No, not that I have heard of*

Edward complained in animated tones about the lack of a local group in Devon at present, bringing the issue back to more general political problems of the uneven distribution of resources throughout the country:<sup>19</sup>

*Edward: I think there isn't any association down on this county which is ridiculous. I have always said...every time there is something, it is always in London or it is always in Birmingham, or it is always Manchester and I said, and I blame the government for this. Anything south from Bristol...the government don't give a rat's ass about anything, north from Glasgow they don't care about, it is that middle belt between Bristol and say Glasgow anything north from Glasgow don't get anything, anything south from*

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<sup>19</sup> Interestingly, there is a NF special clinic in Plymouth, which is not a support group but still it can be a useful source of help. However even the majority of healthcare professionals I interviewed didn't know about it.

*Bristol don't get nothing...never done anything in this part that's ridiculous it is a big area; the southwest is a big area it is the biggest area in the country you know, nothing done here it's ridiculous, they can do something in Plymouth for instance, so you are halfway between Cornwall and...it would be very successful if they'd done something like that*

Paige, who used to attend NF1 events with her son Edward, also highlighted the present lack of support in Devon, attributing the cause to a lack of funding:

*Paige: because I know the funds are very very small, so... therefore... at the time they had more NF family support, that worked throughout the area, but now I think our nearest one is probably way up. I know we haven't got anyone down this way...*

Eleanor, Charles' mother, could not continue participating in the NF camps and other activities; due to her personal health and financial problems, she could not afford to pay the yearly subscription.

*Eleanor: because of my financial [situation], because I am not a worker and you get income support so I can't afford the yearly subscription*

Eleanor had a serious form of NF1 with cognitive and physical complications. She had experienced difficulties finding and keeping jobs conducting a life in a low socioeconomic status. After she developed MS (Multiple Sclerosis) her situation got worse; she was not able to work anymore relying solely on social income support.

## **NFA- NF1 Variability**

Joining the NFA (or just the idea of joining it) could expose individuals to the wide spectrum of variability of the condition and in particular to its most extreme end. This link between the NFA and the high variability of NF1 is very complex. Some individuals who participated in NFA events found the experience of meeting other people who were more seriously affected comforting, as they could put in perspective their own personal or familial situation. Others considered the same experience, or the idea of it, distressing, consequently preferring *a priori* or *a posteriori* not to join the support group.

It can be argued that respondents tended to display ambivalent positions towards the variability of the condition similarly to what has been noted about the use of variability to justify reproductive decisions. Thus, the variability of NF1 appeared to be interpreted by different respondents as a factor that could justify the decision of having and not having children and joining or not joining NF support groups.

Grace was very mildly affected. Apart from her family members, she claimed not to know other individuals with NF1. When asked about her willingness to meet other people with the condition she replied positively:

*Grace: well I'd love to meet some other people, that'd be great, that'd be cool... well I wouldn't necessarily want to hang out with them all the time, but I would love the opportunity to meet some other people*

Nevertheless, she did not want to join the NFA since she found its image to revolve around the more extreme and rare NF1 cases, leaving all the other individuals with NF1, who, like her, were mildly affected unrepresented. Thus, Grace suggested the NFA should mirror more faithfully the wide ranging nature of NF1.

*Grace: If you have got major disfigurement you probably want to find people who are like you, you want that kind of normalization of your situation and I understand that, but what if you have got NF and you don't have any problems with it? The only place you've got is full of people who look very different to you and they are so rare and unusual...how many people are there? You know, how many people in the country are there with the major facial disfigurements you see on documentaries? It's like a handful isn't it? So why is that the face on NF in terms of the online resources? I think it shouldn't be*

Tim had NF1 with physical and cognitive complications. He and his wife Claire, who was undiagnosed, participated in one NFA event. Talking about that experience, Claire claimed not to have had any problem in meeting other people with NF1. However, she noticed Tim seemed to feel uncomfortable especially around individuals with NF1 who were more severely affected than himself.

*Claire: I don't mind it. I'm not worried but I'm not sure about my husband and – I don't know if it's because, perhaps he feels that it's bringing it a bit – yes, you've got it – you know you see some people that are a bit more deformed and stuff [...] somebody came. I can't remember his name – he had a funny face. My husband stayed away from them. He was at the bar with me, but my mum and I were sat down talking to them, and he stayed at the bar*



On the other hand, for Eleanor, attending the NF camp and realising that there were people much worse than her helped her to better accept her condition and gain some confidence.

*Eleanor: I thought since the meeting of all these people “well, I’m lucky I only got it across my stomach, torso and all over my top-half of my body, which can be hidden” you know, just the odd one like that and people don’t really notice those too much [...] I saw these people with it and an awful lot worse [...] this made me think “wow... I am not, you know, not as bad as people make out by looking down their nose at you”*

Dylan who presented with visual, cognitive and physical complications similarly stated:

*Dylan: for my part in groups I have seen people with growths down the side of their legs, I saw a lady whose face had basically slid. Basically it can affect people in different ways. I’m still quite fortunate. I can still get myself about by myself whereas others can’t. They need 24 hour care and support. So I have got it quite mild compared to other people*

### **NFA: “A Negative Message”**

A few respondents, regardless of the severity of NF1 experienced, felt the NFA delivered a ‘negative message’ conveying an excessively invalidating image of NF1.

Grace pointed this out for example by criticising the layout and style of the NFA website:<sup>20</sup>

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<sup>20</sup> The interview took place in 2008 so Grace was referring to the website layout up to that year. It has to be noted that in 2009 the NFA website underwent significant changes in its layout.

*Grace: It doesn't look like a very appealing or a very inspirational place to go. There's nothing inspirational about it. [...] I'm saying that recognizing that I'm possibly being a bit snobbish about it but there's an absence of aspiration about it... instead of saying "let's celebrate some successful people" ... or something... there's a lot about fundraising on the website, "Let's celebrate someone who's jumped out of a plane" woohoo... brilliant well done, whatever, I don't really care (laugh)*

Rosemary, Grace's mother, explained that when the NFA was founded her husband Benjamin- who was very severely affected and had an extensive medical experience due to long periods of hospitalization- decided to join to use his testimony to help other people with the condition. Nonetheless, Rosemary recalled that Benjamin was unhappy about attending the NFA events; he *'didn't ever look forward...he didn't go regularly...but he didn't look forward to it'*. He used to complain about the excessively negative atmosphere he always found at the meetings expressing the belief that more than providing support the association was frightening people with NF1.

*Rosemary: I'm pretty certain it was when that society first started up. He [Benjamin] decided that, as having the disease... he looked at it and he felt they were over-exaggerating things. He thought 'well no I've got it. I've had a lot of input from NHS...maybe my joining could help other people who have it' [...] but he always came back upset, he didn't like what he found there. That they were almost saying: "well you got leprosy". I know... I may be over-exaggerating, I am not using words he used, but he didn't like a lot of their information, he felt it was frightening people who had it*

Gabrielle and Ethan, Dylan's parents, tried to join the support group when Dylan was a child, thinking it may be beneficial for him. However they both decided to abandon the group very quickly since they found it to be too negative. Gabrielle suggested for example that NFA members had such a negative attitude towards NF1 that they believed that individuals who know to have the disorder should not have children. In fact, Gabrielle recounted feeling that NFA members were judgmental over the fact she had another baby after Dylan. This attitude can be interpreted as an example of a sense of guilt and responsibility for genetic futures of children.

Gabrielle also noticed that the support group tended in general to overemphasise individuals' NF1 disease identity and she thought this was deleterious for her young child Dylan.

*Gabrielle: I thought it would be useful for Dylan possibly later. I thought he would meet other people, [...] I went along and it was fine. We didn't mind meeting people with NF, we didn't mind joining in with things, but I was constantly introduced as "the mother whose son – this is Dylan who has NF, and this is me – and they have another son, but of course when they had the second son they didn't know that there was NF in the family". And it was almost saying to me, "you shouldn't have had more children because if you had known there was NF in the family, maybe you shouldn't have had more children". That was the message that came across and I didn't like it. [...] I also felt – Dylan was being introduced as someone with NF and people were labelling him and I didn't want at five, six, seven years old I wanted him to be him and not "Johnny down the road who has NF"*

The following episode gives a more vivid example of Gabrielle's conflict with a certain negative outlook on NF1 that permeated the activities of the NFA she attended:

*Gabrielle: when they went to do a collection they wanted me to wear a t-shirt that said, 'NF ruins young lives', and I didn't feel that that was appropriate [...] I felt there was a really negative message and as a mum I didn't want my children to be reading that because I didn't want them – a) I didn't want them to think that, and b) I don't think it does – I think it changes, but not ruins, and I thought it was too powerful a message*

Ethan also gave a very explicit sketch of his perception of the negativity that permeated the NFA:

*Ethan: I think the people's negativity there was so – almost like they were on a crusade to make people unhappy – one or two of them – and it was very much, and it wasn't very much a sharing experience, it was almost like they were there to try and get as much as they could and not put much back into it*

Edward participated in one NF camp successfully engaging in the NF camp physical activities. He recounted that the organisers and other's participants were astonished over his performance and the fact somebody with NF1 could be fit, even doubting he had the disorder.

*Edward: I was pretty fit, there were always activities. As you know a lot of people with NF have difficulties moving around and things like that, you know, and I got told back on from anybody, and right I can do this, all these things like that "you can't .." no problems I could shoot up that wall I was as fast climbing as the instructor was and he realised "are you sure you have got NF?" "yes I have got NF" he said "I have never seen someone*

*that fit”...there was a rowing machine and there were competitions, you have got a rope 2000 meters you go as fast as possible... and the fastest time was about 16 minutes and 6 seconds...I got there in 6.45! You know they couldn't believe that... “How could you? You are really fit!” you know ...they could be as fit as I am... I have always trained I have always done running I have always done cricket, football, whatever I have always kept fit*

This story told by Edward can represent a further experience of a biased/prejudicial and invalidating attitude towards NF1 held by the NFA.

### **Engagement with Associations Related to Specific Pressing Symptoms**

A few respondents did not seek help from the NFA, but from other associations, internet websites and support groups related to the specific pressing symptoms or concerns they were experiencing. This was also indicative of the fact that individuals' disease identity and health management practises could revolve around pressing symptoms or concerns, rather than the whole genetic syndrome.

Seth, whose son Adrian had NF1 and died of malignancy, stated that although he was a member of the NFA he and the family never attended NFA events nor asked the association for any support: *“it is not that the NF association wasn't supportive, it's that we weren't asking for their support”*. Seth explained that the lack of engagement with the NFA was mainly due to the fact that Adrian's pressing concern was malignancy (and not NF1 as such), so the family sought the help of an online support group on soft tissue sarcoma.

*Seth: ...the NF association...I was simply a member. I received the newsletters, I read them, I never went to a meeting I never participated in any way! [...] Because...partly because when he was really ill, [...] his concern was not NF, but cancer. That was his pressing concern and we did get support from an internet...group around soft tissue sarcoma*

Grace was undergoing a delicate and complex period around the birth of her son Jeff. Some of the difficulties she was facing were related to the fact that, even before Jeff was diagnosed, she felt he had inherited NF1 from her; furthermore she found it difficult to disclose information about the disorder to her partner and his family. During this intense period she looked for help and comfort on an online forum of mothers who were undergoing similar problems related to pregnancy and disclosure.

*Grace: there was a forum that I used during this time and it was quite difficult in terms of Jeff and my feelings about that, there was a forum that I found online where I met some people that I talked to for a bit, and that was really useful, and it was quite mind-blowing ...because I'd never met anybody else like me, I've never met anybody else with neurofibromatosis. [...] Nobody's ever been that useful emotionally I suppose, apart from the couple of people I met on this forum. One of whom had a son who's a bit older than me, my son, so she'd been through that process and helped me a bit in that process of me telling people, like telling my son's paternal grandparents and trying to protect the information, so that it kind of went out correctly*

The online forum represented for Grace a protected environment in which she could share feelings and emotions with a reasonable possibility of preserving confidentiality.

Grace provides a very illustrative example of how other familial and biographical instances (e.g. motherhood, disclosure, responsibility) can strongly shape individual experience with and management of genetic disorders. Grace sought support from a specific forum of mothers of children with disability and not from a general NF support group.

Because of Dylan's NF1 related visual complications Gabrielle and Ethan decided to enrol him in a primary school for the partially sighted. Both parents openly admitted Dylan and themselves found decisive support from this school, rather than the NFA. Notably, in their discourses the contrast between the abovementioned negativity of the NFA and the motivational and empowering attitude of the school is quite clear. Whilst the NFA was viewed as being oppressive, inefficient and potentially dangerous, the school seemed to possess the right characteristics to support Dylan in facing and managing his NF1. The school worked as a support group for this family.

*Gabrielle: going to the school for the partially sighted I think was absolutely paramount to how he [Dylan] is now because they made him feel that there was absolutely nothing he couldn't do, or couldn't try and they saw him as a whole person [...] as a complete person and homed in on his strengths and built up his weaknesses.. We've met some really nice people – through schooling. [...] We've met people who have been really positive and made it – not everything is negative about stuff. The schooling has been brilliant [...] and it was lovely to see people who believed in him and invested in him regardless of his situation, so I think that's really great and you get to meet some really great people*

*Ethan: the support they gave to parents there was spot on. It was very very good, there were lots of sort of interactions with the parents throughout the*

*year, and I got more from that, than from a specific organisation which has an association with NF. The school was there much, very much – you know a whole lot of children have a whole range of disabilities. I don't think they had anybody else there with NF. Dylan was quite unique, but they had a lot of kids there with all sorts of hereditary genetic neurological disorders which affected sight, and the school were great in supporting that – so that's where the support really came from*

### **Management Strategies “Not Thinking about NF1 and not Being Group People”**

The already discussed widespread tendency to downplay, ignore NF1 or focus on positive or more urgent aspects of daily lives, could be also interpreted as another important factor that prevented individuals from engaging in any NF1 association or support group. Obviously rejecting strong NF1 identities implies avoiding any form of group which defines itself around the syndrome.

Nicole's testimony is very vivid as well as particularly interesting being internal to the NFA. Nicole had been working for a few years for the NFA and as such she observed that some individuals with NF1 prefer to avoid thinking about their disorder as much as they can. She even affirmed that “*the people who cope, actually forget about it most of the time*”. Notably, Nicole herself despite being diagnosed when she was 16- and since then having an increasing number of neurofibromas all over her body and face -only started seeking information about her condition when she was 40. She eventually decided to join the NFA, in her early 50s, only two years after she went into job retirement.



*Nicole: There are some people who say “I don’t wanna know”, which is fine. [...] I understand that exactly because I didn’t get involved until I was 40, and absolutely fine, I understand why you don’t want to, you want to live a normal life, you don’t want NF stuffed down your throat all the time [...] the people who I think cope actually forget about it most of the time. Unless they’re brought up short, then you tend just to live and think “well you can’t spend all your time thinking about it” and get on and live*

Ross considered himself as not seriously affected by NF1 and therefore not in need of joining any support groups. He repeatedly highlighted how inessential it would be for him to join the NFA, stating that because of his very mild form of NF1 he would not even be able to properly help other people with the condition.

*Ross: I think ...Dr X did say there are support groups out there. Because I feel...because it doesn’t really affect me...I feel at the moment there is no need to join it...if you see what I mean....I guess if I had more time I could join it, to help others...but I don’t know if it will be beneficial or not because there’s nothing...because I haven’t got it severe, I don’t know how I could help*

Other respondents like Vera openly declared they were not interested in joining support associations of any sort simply because they were not “group people”. This is indicative of the fact that individuals may not like to participate in group activities and having a genetic condition does not necessarily change their dispositions. Although some members of her family attended NFA events (e.g. her sister Paige and nephew Edward)

or benefited from the help of NF family advisor<sup>21</sup>s (e.g. her grandson Douglas), Vera stated she never got personally involved in any NF group activity due to her solitary nature.

*Vera: I haven't joined any groups. I am not a groups- joiner. I don't join groups of any kind I am not a...I don't like joining groups , no, no I am very very.....I like to be on my own a lot, I don't like joining in with people a lot...I am not that kind of person*

It was also not uncommon for respondents to say they never consider joining a NF1 support group because they were too busy taking care of the aspects of daily life. This was, for example, the case with Martha:

*Martha: to be honest I haven't really thought about it [joining a support group] , I suppose, because I work fulltime and I've got children and I am always busy, I just haven't really...because I haven't got the time to do things, I haven't really thought about it*

## **Familial Engagement**

Claire provided an example of engagement with NF1 and in NF1 related activities on a familial more than support group level. Although Claire's family joined the NFA they did not get involved in meetings and events organised by the association:

*Claire: well – things came along, they [The NFA] would invite you to go up to meetings and things, but we've never had done. I suppose it's just one of*

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<sup>21</sup> Family advisors are NF specialist advisors who work within hospitals and give support to anyone affected by NF1 (see Chapter 8).

*those things we never – we decided, no we can't get up there, or no, we haven't got time to, or – so, yeah...*

Their way of helping consisted instead of organising- relying upon the help of family and relatives- fetes and other social events in their village to raise funds for research on NF1. Thus, rather than “joining” the NFA to *ask for help*, Claire’s family preferred to *give help* to the association and other people with NF1 by organising their own charity events.

*Claire: we joined the association; the neurofibromatosis association, and started doing fundraising and things for it, and that way, so that helped us cope with it a bit more because we were, you know, helping other people as well, and helping them try to do the research to find helping, helping, to find what's the cause of it, to help it along the way.[...] - my aunt started it by doing a cream tea, then after that we done a general sale, disco's and different things..[...] Yeah, draws, raffles, things like that, all that different village fete stuff – stuff like that. My dad passed away six years ago as well, and they do a darts thing, memory, every Christmas, you know, a darts competition, and the money goes towards NF*

### **“(Bio)Social Service” (Helping Other People with NF1)**

Generally, the idea of helping other people with NF1 was mentioned by respondents as another possible modality of engagement with their disease identities.

Concerning NF1 support groups, some respondents affirmed the only way they could possibly “join” a support group would be to help other people with NF1 by using their experience with the condition. This is for example the case of John:

*John: I feel a support group won't help me...that's the way I am, [...] I don't know how I could help anyone with a similar problem, by talking to them, you know I just...I don't know, I don't know if it affects people mentally more than physically how I can....say someone was suicidal, over similar to what I've got and I went talk to him.....maybe...by talking to them.....about the complaint... I might help him getting over it*

John's body and face were completely covered by neurofibromas. He stated he could hypothetically enter into a support group to help other individuals who were facing similar problems by sharing his personal experience. John had been depressed and suicidal because of the discomfort caused by his neurofibromas and he may have benefited from some help. However he never sought help from any NF1 support group, preferring to manage his problems by ignoring his condition as much as possible. Joining a support group, as he explained in the following passage, would have amplified the reality of his NF1 and being antithetic to his management strategy:

*John: I'd have to be into it [NF1] more than what I already am and I don't want that*

## **6.5 Concluding Remarks**

This chapter has argued that, regardless of the severity of NF1 experienced, the majority of respondents rejected NF1 identities, employing diverse downplaying strategies such as normalising the condition, avoiding thinking about it or focusing on the positive or more urgent aspects of daily life.

Nevertheless, despite the widespread downplaying attitudes, there were moments in participants' lives in which NF1 could become salient. This was mainly in relation to the emergence of serious symptoms and the familial implications of genetic information. Participants demonstrated, in particular, 'genetic responsibility' behaviours concerning reproductive choices and the management of genetic risk and information. Overall, NF1 did not appear to be associated with a strong disease identity or community. The identities of individuals with NF1 were fragmented around specific symptoms. Mildly affected individuals often did not want to identify themselves (or be identified) with the more seriously affected, and vice versa.

These findings have important implications for the discussion on the notion of genetic subjecthood, responsibility and citizenship. At first glance, as envisaged by many social theorists analysed in Chapter 3 (Lippman 1992; Lippman 1998; Finkler 2000; Kerr and Shakespeare 2002; Kerr 2004; Rose 2006) NF1 and its genetic nature could actually be relevant for the patients and families interviewed. However, the salience of genetic information was not a stable, life-long phenomenon, but tended to vary within and between individuals and to surface at certain critical junctures in the life courses of the participants, who, otherwise, deployed downplaying and minimising discourses towards their disorders. These critical junctures tended to be mainly related to the aforementioned severe symptomatology of NF1 and familial instances. The discourses deployed by the participants of my study were more in line with other empirical research on individuals' constructions of genetic disorders (reviewed in Chapter 3) which highlighted the heterogeneity and situatedness of disease experience (see for example (Parsons and Atkinson 1992; Cox and McKellin 1999; Hallowell, Arden-Jones et al. 2005; Petersen 2006; Sanders, Campbell et al. 2007; Klitzman 2009)).

## **Normalization**

Normalization strategies, notwithstanding the presence and seriousness of a disorder, have been widely reported in literatures on chronic illness and disability which have focused on various conditions with or without a genetic basis (see also Chapter 5, § 5.5).

The vast majority of these studies are often based on qualitative interviews with affected individuals or parents of affected children. One of the main overarching themes which traverses these literatures is that responses to - and experiences of - illness are not solely determined by the biological symptoms, but are also shaped and mediated by biographical and wider socio-cultural factors and expectations (see also Chapter 3, § 3.3).

In the classic text 'Chronic illness and the Quality of Life' Strauss and Glaser (1975) explored the implications of living with various specific conditions and identified normalization tactics as being basic coping strategies. They observed how, alongside managing symptoms, trying to live as normally as possible despite the disease was a main concern of chronically ill people (and families) (Strauss and Glaser 1975).

In Kelly's (1992) study of individual experiences of ulcerative colitis, patients were also engaged in impression management exercises apt at presenting a relatively normal self. However, normalization strategies were jeopardised by the unstable nature of the condition. Impression management often required the help and support of relatives and partners, especially when affected people were facing society (Kelly 1992).

Kayama-singer found that the vast majority of respondents (both men and women) in USA who were diagnosed with cancer presented themselves as being healthy (Kayawa-Singer 1993). Their comments such as "*I am very healthy. I just have this problem*" (Kayawa-Singer 1993: 295) bare strong similarities to those of the NF1 respondents in my study.

Age and life-course are also important biographical and social factors that can mediate the impact of illnesses. In a study on osteoarthritis, older people normalised their symptoms by incorporating them into the natural process of ageing (Sanders, Donovan et al. 2002). Along the same lines, old people who had strokes did not consider the event to be a major disruption in their lives (Pound, Gompertz et al. 1998). Stroke was construed as a 'normal' and inevitable aspect of ageing and one of the many problems experienced in a lifetime.

In line with these findings, the other strategies reported by the respondents in my study (e.g. being fit, preferring not to know about NF1) can be seen as further attempts to normalise or ignore symptoms for as long as possible (Stewart and Sullivan 1982; Johnson 1991) in order to maintain the maximum relative normality in the face of impairment and stigmatization (Locker 1983; Kelleher 1988; Pinder 1988; Gerhardt 1989) and ultimately exert control over life (Charmaz 2000b).

Disability studies have also shown how affected individuals can claim disability as a positive identity (Oliver and Barnes 1998; Hughes 1999; Reeve 2002; Thomas 2007).

The ideas widely reported in disability literature of the value of difference (Reeve 2002), that there are positive aspect in living with (and being challenged by) a disability or in living with disabled individuals - are also in line with Frank's (1995) quest narrative (Chapter 5). Grace's argument presented in this chapter that having a condition can be empowering - since it gives more information about the actual or potential symptoms and complications the affected person can develop- represents an original embodiment of these ideas.

Olney and Kim's (2001) focus groups with university students with a variety of cognitive impairments showed that disability was positively integrated with their image of the self. Embracing and valuing difference, identity management, personal definition

of difficulties and limitations were among the main normalization and resistance strategies adopted (Olney and Kim 2001).

Green's (2007) quantitative and qualitative study on mothering children with a wide range of physical and intellectual disabilities illustrated that although mothers experienced socio-structural constraints in caring for children with complex needs, most of them found there were also valuable aspects in having a child with a disability (Green 2007). In a recent study, Landsman (2009) reported that mothers of disabled children experienced a diminished motherhood – especially at the onset of the disability.

Nevertheless, the experience of caring for disabled children lead mothers to change their views. Children were often described as gifts. Moreover, mothers resisted and criticised socio cultural consumerist models of the perfect and healthy child and consequently redefined 'normality' and 'difference' construing difference as a universal characteristic of human nature.

### **Universalization of Difference**

Discourses of universalization of difference were not uncommon among the NF1 individuals and families I interviewed (e.g. Gabrielle). They strongly echoed the ideas of 'ubiquity of human misery', 'unity of suffering', 'universality of disability and impairment' (Turner 2003; Hughes 2007) and that disablement is an intrinsic aspect of the human condition (Bichenbach, Chatterji et al. 1999) which permeate a lot of disability literature and which again can be seen as presenting elements of quest narratives (Frank 1995). Disability can have a transformative effect and lead the affected person and/or those who care for them to gain a different/deeper insight on life and the human condition (Larson 1998; Hughes 2007; Landsman 2009). In addition to serving as 'private' coping strategies, these universalization arguments have often been deployed to challenge the idea that disabled people are a minority, and have also been used to advocate the rights of disabled people (Shakespeare 1994).



## **Balancing**

However, the management of chronic illnesses and disability is often characterised by an uneasy equilibrium between normalization/resistance strategies and the manifestation (and potential exacerbation) of the disorder.

Balancing these two aspects is paramount to affected individuals and families.

This chapter has shown that a common downplaying strategy adopted by participants consisted on focusing on the most pressing symptoms when they emerged, rather than seeing themselves as having a chronic genetic disorder. This behaviour mirrors Charmaz's point that if acute episodes are followed by a semblance of normal life, affected individuals may interpret their chronic illness as series of acute episodes rather than a lifelong condition (Charmaz 1994). Moreover, as I will show in Chapter 8, this attitude can also be influenced by access to healthcare services.

Balancing is more complex when conditions are highly uncertain and unpredictable (Pinder 1988; Ablon 1999). This can be reflected in uncertainty of behaviours (Bury 1982) in relation to the appropriate way to manage symptoms especially when there are not clear medical regimens and prescriptions available.

As I have shown in this chapter (e.g. Seth), uncertainty could significantly undermine individual downplaying discourses also because the unpredictability of NF1 was often not 'solved' or managed by medical and genetic knowledge.

Moreover, by exploring accounts of different family members, my study has also illustrated how normalization strategies present complex and sometimes contradictory elements if contextualised within the familial environment. Different family members, whether with or without NF1, can have a different experience of normalization and uncertainty (e.g. Eric and Janice).

### **Genetic Responsibility**

The family had also an important role in relation to reproductive choices and the management of genetic risk and information. These findings about respondents' genetic responsibility behaviours are mostly in line with other research on NF1 (Benjamin et al 1993; Ponder 1998; Ablon 1999) and other genetic conditions (See also Chapter 3).

In all these studies, NF1 was found to influence individuals' reproductive decisions.

Unlike my research, in Ponder, Murton et al (1998) and Ablon (1999) NF1 individuals' reproductive choices could also be influenced by the severity of the condition experienced by prospective parents or seen in other family members.

The findings in this chapter are more akin to Benjamin, Colley et al (1993) study which reported there was no significant link between participants' perception of their own severity, medical classification of severity and reproductive decisions. Another point of commonality was that affected individuals who had children with NF1 showed a relatively higher knowledge of the condition than those without a child. A greater knowledge of the disorder was also found in those who stated that the presence of NF1 had influenced their reproductive decisions Benjamin, Colley et al (1993). This is in line with the link found in the participants to my study between fluctuating relevance of genetic risk and genetic responsibility behaviours (Petersen 2006).

As highlighted in Chapter 3, Parenting and reproductive decisions are important familial aspects that encourage individuals to seek more information about genetic risk and disease (Fretz, Duivenvoorden et al. 1991).

Furthermore, as shown in my thesis, Benjamin, Colley et al (1993) respondents reported that the variability of NF1 was one of the main factors which complicated reproductive decision making. The great variability in prognosis was also linked with the low uptake of prenatal diagnosis for NF1. Similar links between variability and low uptake of prenatal testing have also been reported other conditions (e.g. neurological disorders) (Binedell, Soldan et al. 1998; Downing 2005).

In my study the vast majority of respondents were not knowledgeable about reproductive technologies available. This supports Ponder, Murton et al (1998) findings that NF1 individuals reproductive choices were influenced by the timing and poor access of genetic counselling (see also Chapter 8).

Another important finding, i.e. the complexity of enactments of genetic responsibility behaviours, resonates with literature on a variety of genetic conditions.

In Chapter 3 I have shown the complexity of individuals' forms of engagement with genetic risk and responsibility. Both men and women can draw on discourses of responsibility, care, guilt determinism to affirm their identity as individuals and family members (Hallowell 1999; Hallowell, Arden-Jones et al. 2006; Weiner 2010; Arribas-Ayllon, Featherstone et al. 2011; Arribas-Ayllon, Sarangi et al. 2011).

Raspberry Skinner (2011) showed how women carriers for FXS reproductive decisions were complex and shaped by lived experience. The authors propose to expand the notion of genetic responsibility. They argue that not only the accounts of women who chose not to have children but also those of women who purposely became pregnant or continued an unplanned pregnancy presented elements of genetic responsibility which included negotiation of religious beliefs, personal aspirations, family values and diversity (e.g. parental love and respect towards diversity and children). This is in line with the complex account given by my participants (e.g. Nicole, Paige ) about their choices or views about having children.

### **Biocitizenship and Biosociality**

What has been pointed out so far can explain as well why NF1 resists the idea of biosociality and biocitizenship *vis-à-vis* support groups. NF1's extreme variability and individuals' management strategies, which do not necessarily entail joining support groups or adopting strong NF1 identities, emerged as important mitigating factors.

Participants' reluctance to join NF1 support groups was also justified by contextual-historical characteristics of the UK NF1 association (for example the lack of structural support or the way the association portrayed NF1).

Although this doctoral thesis presents just one case study, the findings in this chapter suggest that scholars like Novas, Rose and Rabinow may overemphasise the significance of biosociality. The findings confirm Ablon's points discussed in Chapters 2 and 3 about NF1 support groups (Ablon 1999). They are also in line with Benjamin, Colley et al (1993) study with NF1 individual conducted in the UK. Although a considerable part of the participants were members of the NF1 support group - at the time of the study called LINK (see Chapter 2) - they also stated that medical professionals were their main source of help. Participants reported to have joined the group more to get information about the condition rather than to use it as a social networking service (Benjamin, Colley et al 1993).

Therefore, it is possible to extend the scope of these findings and claim that currently in the UK, as well as in the US a few decades ago, the variability of NF1 can represent a considerable obstacle towards a unified patient identity and sense of community.

In addition to Ablon's argument, my study found in patients' downplaying strategies another crucial hindrance to a NF1-biosociality. Ablon's interviewees dwelled on the stigma experienced and the general negative impact that the syndrome had on their life. Along the same lines, the genetic nature of the condition tended to indicate the presence of something rogue or defective which was deeply entrenched in their body (Ablon 1999).

On the contrary, the participants in my study -whilst still mentioning instances of stigma and distress- were much more prone to minimize the impact of the condition on their lives and to distance themselves from it. Another novelty in my findings is represented

by patients' engagement with support groups related to specific symptoms, rather than to NF1.

The variability and unpredictability of NF1 present important mitigating factors against a unified identity and thereby a sense of biosociality.

As discussed in Chapter 3 the notions of biosociality and biocitizenship have been grounded mainly on studies which have explored the experiences of living with late onset disorders - like HD and HBOC- which have known and predictable phenotypes (Rose 2006 ). NF1 is instead characterised by highly uncertainty phenotype.

There is surely a degree of uncertainty in HD and HBOC. HD is variable in terms of age of onset and severity. HBOC can cast a spectre over individuals' lives for decades as they may not know whether or not they will get cancer.

However, NF1 is a syndrome and has much more variable and unpredictable manifestations. It can cause different physical and cognitive symptoms (e.g. bone lesions, benign tumours, malignant life-threatening tumours, visual impairment, learning difficulties etc.). Moreover, it can be life-threatening to a tiny minority, highly disfiguring and stigmatising to others, it can develop in unpredictable ways during patients' life courses, but it can also be very mild.

As Shakespeare (1996) pointed out in relation to the idea of disability identity, the experience of disability is also affected by the nature of the impairment. Not all disabled people experience the same degree of disabling barriers and discriminations. For example, individuals with visual impairments may have a different experience of disability to those with mobility impairment (Shakespeare 1994; Shakespeare 1996; Hughes and Paterson 1997). NF1 can cause a variety of impairments and therefore a variety of different experiences of disability ad illness. A unified symptomatic identity for such a variable and unpredictable condition like NF1 is less likely to emerge.

There are also more general considerations about the very idea of illness identity that can mitigate the relevance of biosociality.

First, considering illness identity as an important or prevailing trait in affected individuals and families' lives and subjecthood may lead to overlook other identities such as gender, ethnicity sexuality, age and other relevant biographical elements.

Moreover, the same socio-political context- which underlies the notions of biosociality and biocitizenship - characterised by public health discourses about responsible patients-citizens and a strong connection between health, responsibility, productivity (see Chapter 3) can be seen as potentially undermining these same notions.

In such socio- political climate, the inability to achieve, maintain or restore health tends to be viewed as a weakness of the person and ultimately as a failure to fulfil the obligations of the responsible patient/ citizen (Novas and Rose 2000). These considerations can help to explain the tendency of ill and disabled people to reject identities based on illness and disability and to seek as much as possible to re-establish a sense of legitimacy and self outside the realm of illness (Charmaz, 2000).

Therefore, individuals who are diagnosed with a disease, rather than endorsing a disease identity may prefer to separate it from the self as much as possible, downplaying it and/or adopting a variety of normalization strategies. This tendency mitigates the idea of biocitizenship and biosociality (see also Chapters 8 and 9).

The lack of biosociality for NF1 is not only theoretically important, but can also have important clinical implications. Healthcare providers may refer patients to specialized disease specific support groups (especially when the NHS does not possess the structural capacity to manage certain complex or rare disorders), assuming that they will join and find the help and information they need. I would argue it is important for healthcare providers to know that with conditions like NF1 (and possibly other

multisystem variable disorders) this may not necessarily be the case.

The notions of biocitizenship and biosociality may be more relevant for other disorders with a clearer profile and less variability (like HD).

Further research may help in clarifying the kinds of conditions to which these notions are more evident.

## **7. NF1 and Familial Dynamics**

### **7.1 Introduction**

This chapter presents the second part of the analysis of patients and family interviews. It principally focuses on family dynamics, in particular on individual and familial meaning making practices around uncertainty, which is the second research aim of this thesis.

The chapter provides diverse examples of how familial dynamics (like parenting, family bonds and patterns of communication) can significantly affect individuals' construction of and experience with genetic disorders.

In section 7.2 I will illustrate this link by reflecting on the different modalities in which the individuals interviewed engaged in practices of surveillance of their family members as a significant part of their process of dealing with the uncertainty of NF1.

Further examples of the conflation of family dynamics and the construction of genetic disorders will be shown by the exploration of communication and relations amongst family members, representing the main themes in section 7.3.

In section 7.4 I will discuss the dimension of parenting, providing some examples of how parents attempted to deal with the uncertainty of their children's NF1 by blurring the pathological with more 'common' challenges related to bringing up a child.

The implications of these findings to the notions of practical kinship (Featherstone, Atkinson et al. 2006) and medicalization of the family (Finkler 2000; Finkler 2005) will be further discussed in the concluding remarks (§ 7.5).

## **7.2 Uncertainty & Family**

As already discussed in Chapter 2, NF1's physical, cognitive symptomatology and complications are highly variable and difficult to predict. Dealing with the uncertainty caused by the unpredictability of NF1 represented an important aspect of participants' accounts of their experiences with this syndrome. In fact, all the participants in the study reported to have, or at least to have had at some point in their lives, concern over the possible developments of their own NF1 and or the NF1 of other family members.

Although NF1 displays high variability and unpredictability both within and between individuals, surveillance of other family members with the condition emerged as an important and powerful tool used by respondents in managing these concerns. In the absence of a general role model for NF1 (Ablon 1999) and clear prognostic pathways, the family was an accessible and powerful resource for many respondents to come to terms with the gap between their present and an unknown future.

Family members with NF1 were often explicitly or implicitly mentioned by individuals as benchmarks/models in attempting to make sense of different aspects of uncertainty, for example, how to manage one's condition and how it may develop. This resonates with the findings from the narrative chapter.

Ross was an adult with a relatively mild form of NF1, but a family history with more serious cases. Despite the generally mild manifestation of his condition, Ross developed in his late teenage years a small, but visible, neurofibroma on his face. As this



neurofibroma was causing him cosmetic discomfort, he underwent a surgical operation to remove it. Healthcare professionals made him aware about the possibility that removed neurofibromas can grow back again. The following passage illustrates Ross's reliance on the example of his father, John, who had already undergone several surgical operations due to the same problem, as a way to attempt to overcome his feelings of uncertainty related to the possibility of his removed neurofibromas growing back.

*Ross: yah...but they [the doctors] did say they [the neurofibromas] could come back, there is a chance they can come back through it...and there's no signs just yet...but my dad has had lumps removed and when he's had them removed they haven't grown back, but he's had them removed a good few years ago now...*

It is worth pointing out that Ross used his father as a model despite a considerable difference in their symptomatology. Ross was very mildly affected by NF1 presenting only one visible neurofibroma on his face, whilst his father's body and face were completely covered by them.

Observing the broader family history also appeared to be a powerful sense making strategy around uncertainty. Reflecting on what was known or supposed about occurrences of NF1 in past and present generations was a common way employed by respondents to try and predict the symptomatology and progression of their NF1 or the NF1 of other family members. The invocation of family members as benchmarks to visualize possible future trajectories of the disorder was common among respondents;

*Ross: my father had it, so...my dad had it and his brother's got it and their dad had it and that's...my granddad originally, from it, so it's come from*

*there really...but I think as it's gone down it slightly got slightly less, my dad is the worst case scenario...my uncle, which is my dad's brother has got it...but he's got a slight less than my dad and my sister's got it worse than me, but she's younger than me... (my emphasis)*

By observing the trend of manifestations of NF1 through generations, Ross ventured to formulate the hypothesis that the severity of NF1 decreased through generations. Similarly, Claire attempted to use the example of other affected relatives to gather possible indications concerning the future of her teenage NF1 son Leonard:

*Claire: sometimes I wonder because I know they – my husband's parents, well his mum passed away early and so did his granddad – 66 his granddad was, something like that, so I think are they still going to be going on – it's hard to know*

The above passages, like the ones of many other respondents with a NF1 family history, are examples of how the family can be a resource drawn upon by individuals to fulfil their need to explain and manage their own NF1 and its modality of manifestation within the rest of the family.

Respondents were also driven by their personal and familial experience with NF1 to assess whether other family members had the condition and, if so, how they could develop it. Once again, these models of surveillance and diagnostic practices did not necessarily follow biomedical knowledge, but could be strongly driven by their personal and familial experiences and beliefs.

The following passage from John, Ross's father, clearly illustrates these points. Because of his personal and familial experience with NF1, John drew the conclusion that Ben, his two year old, yet undiagnosed grandson (Ross's son), had NF1. Despite the fact that birthmarks are not sufficient diagnostic criteria for NF1, their presence in the grandson represented for John a significant diagnostic proof. The following passage shows that behind this form of surveillance of younger generations, there could also be feelings of guilt and responsibility for passing down the condition.

*John: In my experience, because I've got it, I look at him and I see this mark, and I know that he has got some form of NF. That's how I feel. I just know he has, he's got it. But it's like me, I think up until I was 15, [...] up until I was 15/16-ish, I was beautiful skin, perfect, no problem apart from the birth marks - the what I call birth marks. And our birth marks are exactly the same. And big brother's got them, and Rowan's got them. Ruth, my daughter, has got this mark*

For many respondents, comparing their symptomatology with that of other family members did not only serve as an attempt to shed more light on their future health, but became also a way to make sense and better accept their present condition. This phenomenon could take place for example when other family members were perceived to be more seriously affected.

In thinking about the possible future developments of her NF1, Julia looked at the figure of her father who also had the condition. However, after considering that her father's scoliosis got worse over time and that he also developed malignancy, she tried to reinterpret his example as a reason to feel glad for not being affected in the same way, rather than as a prognostic model:

*Julia: I don't know a few years along the line what it will happen, as I have got the scoliosis whether is going to get worse, I mean me dad did get worse, but it never runs through...what would happened with him might not happen with me! [...] You know you just got to accept that you've got the condition and it could be a lot worse...I have got to be glad I haven't got like, serious tumours or life- threatening lumps and bumps like me dad had*

Examples similar to those shown above were very common across the respondents who had a family history with NF1, but also between affected and unaffected family members. Family surveillance practices are also common in other genetic disorders (Cox and McKellin 1999; d'Agincourt-Canning 2005).

It is important to point out that these surveillance practices could be used by respondents in original and diverse ways. Overall, these practices appeared to be triggered by their attempt to make sense of the syndrome and deal with the spectre of its uncertainty.

### **7.3 Family Communication and Relations**

As discussed above, mutual surveillance between family members was a very common and important part of individual and familial experiences with NF1 and other genetic disorders (Cox and McKellin 1999; Clarke, Richards et al. 2005; Gaff and Bylund 2010). However, respondents seemed to suggest it was not accompanied by much direct communication. The majority of respondents appeared to relate to other family members mainly in a 'silent way' i.e. by observing them, but without directly speaking to each other. The scarcity of familial dialogue over NF1 was often explicitly or implicitly presented by respondents as a sign that their family was reasonably well adjusted to the condition, hence free from any arguments or any

blame and guilt issues. Within this portrait of familial daily life, NF1 could enter into everyday conversations on particular occasions, for example if one presented problems related to the conditions that were thought to be serious. This familial experiential picture was common among participants.

For instance, Alex, the husband of Nicole, a woman with NF1 whose body and face were covered by neurofibromas, commented:

*Alex: no...she ...is not something she mentions very often...no....sometimes if one of them hurts or if I bump into her or something...she goes 'uh' and realise there is something there but it's not sort of you know, we don't sort of discuss it every day...or anything like that...*

Eric had NF1 and a family history of the condition. In his adult age he started developing a plexiform neurofibroma on his eye. The following passage from Janice, Eric's wife, provides another example of a lack of discussion in her family and in Eric's family about NF1:

*Janice: I think after we were married, well when I met Eric first, he had a slight problem, not very much...just noticeable that there was something wrong with his eye and I asked his mum and she said 'oh he had an operation as a child', tonsils I think, and it was to do with that...I didn't take any more notice of it, but as time went on, it got worse and worse [...] I didn't know a lot about it to be honest, until ...the doctors...Eric's mother never talked about it...*

The example of Ross' family is particularly revealing as it also clearly shows that the perception of seriousness can vary among different family members. Even in the presence of NF1 related problems, the conversational flow within family members may not be always automatic. In fact, whilst the family discussed the removal of some of the neurofibromas from John (Ross's father) Ross instead had to tell John about his surgical operation because John did not notice it.

*Ross: as a family we really just got on with it [NF1] ...we don't really talk about it that much, apart from my dad just recently because he's had one of them removed, but for months we haven't talked about it...and I have my lump removed and my dad didn't even notice it (laugh) so I had to tell him...we really just got on with it...it hasn't affected us as a family, you know we still love each other and we get on with it. [...] we have never had any arguments or anything like that at all...(my emphasis)*

A closer look at respondents' accounts and their contextualisation in relation to the testimonies of other family members, revealed however that the diffuse lack of dialogue about NF1 between family members could also be the result of (and a way of hiding) complex and delicate problems of communication. Family dynamics could interfere with everyday conversation, disclosure and health management practices. Moreover, talking about NF1 could be for example distressing for many family members who may prefer to downplay, ignore or even not accept at all the condition.

Returning to the example of Ross's family, Ross stated in the above quotation that NF1 did not usually enter into familial discussions, suggesting that this reflected the lack of problems related to the syndrome in his family. However, in the light of John (Ross's

father) and Nora's (John's wife) interviews, it is possible to understand that the lack of dialogue over NF1 in the family was also the result of tensions between family members. John's concern about the possibility that his two year old grandson Ben, Ross's son, had NF1 has already been introduced in the last paragraph. Nora observed Ben as well, and given her experience as an auxiliary nurse, community carer and her general interest in medicine, she strongly suspected he may have not only NF1, but also autism. Nora and John manifested throughout their interviews serious worries over Ben's present and future health and wanted Ben to have a proper health check in order to make sure nothing was wrong with him or, if some health problem was found, to obtain prompt intervention. Nora and John's anxiety was aggravated by encountering hindrances when expressing their concerns to Ross and his wife Meredith, who appeared instead to think Ben did not have any problem and did not require any particular health assessment.

*Nora: they [Ross and Meredith] 're not cooperative. If I said to them we think Ben's got autism, I think you ought to go and have him seen they'd cut me off.  
Bang. Just like that*

*John: I said to Ross and Meredith not so long ago, 'has Ben got any tell-tale signs of NF?' They says, "No, he's as clear as a bell", but he's not. He's got the brown spots, the brown mark like a birth mark [...] and that's a tell-tale sign of NF at that early stage (my emphasis)*

Nora and John expressed the belief that one of the main causes of this barrier to communication resided in a 'clash of parenthoods'. They described Ross and Meredith as young and novice parents lacking the necessary knowledge and experience (and

consequently any adequate system of reference) to be able to control Ben's development . Moreover, according to Nora and John, this limited experience led Ross and Meredith to be more protective towards Ben and closed towards external suggestions. Therefore, Ross and Meredith's parenthood irremediably clashed with the more experienced one of Nora and John, resulting in a 'forced' silence that is actually full of tensions.

*Nora: every parent feels that their child is perfect and what they're doing is normal, what the child's doing is normal, [...] Ross and Meredith see Ben as normal because that's what they've seen Ben do all his life. They don't know any different because it's their only child*

*John: They're very protective of their son, obviously, and it's very difficult to talk to them. [...] Because we've got our views, because we're older and more experienced, but they're young people and they've got their own views, and they think they're doing everything right. But we can see maybe that's not the way to go*

It is also interesting to point out that whilst Ross never mentioned this problem he and Meredith were having with Nora and John over Ben's health, he actually expressed his personal concern about Ben having NF1, stating this represented the only real worry related to the syndrome he had:

*Ross: it [NF1] doesn't affect me that much really, but my concern is, will my... because we've got a little boy, will my boy catch it? you know? Or inherit it? [...] everybody in my family now would probably have it, if it's in the gene... it all depends how serious it gets I suppose, so if I have my*



*boy done, they would probably tell me “yeah yeah he has got it” I am guessing so...*

It is beyond the scope of this analysis to further investigate the reasons why Ross was concerned about the possibility of his son having NF1, but at the same time did not get any referral and chose to hide his concerns from his father John, possibly Nora and himself too. What I would like to emphasise instead is that the exploration of this family has offered the possibility to shed more light on the complex interrelations between sense making practices around uncertainty and communication within families.

Combining the interviews of different family members has shown that the lack of discussion about NF1, rather than representing a sign of a family well-adjusted to the condition, can also denote a situation in which family members are all worried about a new-born member of the family having NF1, but are not able or willing to openly discuss these worries because of different strategies to manage uncertainty. The space and management of uncertainty and of communication within families can be interpreted differently by different members; there's no one role of this 'space'. This diversity can be connected with what has already been observed at the end of the previous section of this chapter (§7.2) concerning the diverse modalities of familial surveillance activities.

Lack of communication about NF1 or related problems between family members could also be the result of strategic parenting choices. Rachel was a mother with NF1 who preferred not to explain in clear terms to her daughter Pamela, to whom she passed the condition, what NF1 implies. She was afraid of the negative influence that the information would have on Pamela, especially when she was younger.

*Rachel: I have never explained to Pamela, because I have never said too much about it, because if I say too much, when she was a child it might play on...so I have never said nothing, I have never said to her like...this letter we had...sent to her what we need to look after I have never told her, because if I say to her “look you’ve got, if you get this you got a..” she might play on it*

This phenomenon of limited disclosure described by Rachel also shows how family members- in this case parents- by adopting certain attitudes and behaviours in relation to NF1, can significantly shape children’s experience of the syndrome.

Managing individuals with NF1 who developed complications within the network of familial bonds could pose many difficulties and appeared to be a significant cause for problematic –or even lacking- familial communication. Individuals with NF1 presenting for example cognitive problems or malignancy could be challenging for other family members to deal with.

Monica was the oldest of seven siblings the youngest of which, Robert, had NF1 with some physical complications as well as mild cognitive-behavioural difficulties; Robert was the only person in the family with a NF1 diagnosis. The leitmotif of her interview was the difficult relation with Robert which resulted in unsolvable feelings of frustration and guilt she (and her siblings) felt towards him. Monica stated that, since he was a child, Robert had been complaining about the pain caused by neurofibromas, but her siblings and she had found it difficult to have a deeper and authentic dialogue with him about his feelings towards NF1 and its unpredictability.

*Monica: every so often he [Robert] ’ll say “ouch my lump!” this sort of thing...but he doesn’t actually talk about his fears with it. I mean he doesn’t*

*talk to you about for example if he is scared it is getting worse. He doesn't speak about things*

Robert's NF1 related mild cognitive and behavioural difficulties could be listed as part of the reasons why his siblings could not properly relate to him. However, this lack of dialogue was also aggravated by uneasy relations due to the fact Monica and her siblings always thought Robert complained about his NF1 to attract attention and they tended to express their frustration towards him. Moreover, at the same time, Monica and her siblings also felt sense of guilt for reacting negatively towards Robert's behaviours, since they were also aware that after all he had NF1 and -being the youngest brother in a numerous family- it was understandable he was seeking for attention.

*Monica: he sort of you know brought it back and he was always looking for attention really, well he's the youngest in the family; and I supposed he used this constantly you know "I've got this terrible... disease" and I do really think we...as older brothers and sisters gave him all the sympathy that he deserved possibly, you know [...] he did talk a lot about how painful it was, but we weren't we ...most of us really didn't believe him actually because we thought he was looking for attention. We were cruel brothers and sisters*

In describing the lack and problems of communication between family members due to NF1 complications, instances of survivor guilt<sup>22</sup> (Murakami, Gondo et al. 2001) were also evoked by respondents.

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<sup>22</sup> Survivor guilt in medical genetics: an individual who is not at risk for a genetic condition running in the family, may evoke feelings of guilt for not having inherited the condition that is affecting other family members, and/or feeling excluded by the 'family subset' composed by the individuals in the family who have the condition.

In the following passage, Claire referred to the poignant crisis her family and in particular her two children Leonard and Anna were undergoing because of NF1. Leonard was a teenager with NF1 suffering from serious complications, among which malignancy. Anna, who did not have an NF1 diagnosis, was strongly affected emotionally by the health of her brother and was feeling guilty for not having the syndrome. The physical and emotional impact exerted by NF1 had seriously compromised the everyday life and relations of the whole family. To manage to traverse this sharply critical period, Anna- and subsequently the whole family- was receiving psychological therapy.

*Claire: Well yes, her and Leonard were going through a few emotional problems there due to NF and because of his diagnosis, so she was a very mixed up emotional little girl – well big girl now; she’s 16, but you know she found it hard. She sometimes wished why does he have it and not her – NF – stuff like that you know[...] we’re seeing a psychiatrist because of her – all the problems through it all, because of his cancer side of it and that*

Almost none of the participants made any direct or indirect mention about younger generations blaming their parents for inheriting NF1. Having the condition was usually framed in fatalistic terms as “*having just one of those things*” (Hallowell, Arden-Jones et al. 2006; Keeley, Wright et al. 2009) and family relations did not appear to be compromised by blaming practices around inheritance. The only exception was the case of a mother, Julia who reported that one of her daughters Maya had accused her, only for a certain period, for passing to her the condition.

*Julia: well....that's Maya....Maya got quite cross with me and I said to her "it is not my fault!" you know it is just like a genetic thing, people inherit things...but she is alright about it now*

The space and management of uncertainty and of communication within families can be interpreted differently by different members. This diversity can be connected with what has already been observed at the end of the previous section of this chapter (§ 7.2) concerning the diverse modalities of familial surveillance activities.

### **Perceptions of Marginalization in Families**

Some respondents reported feeling marginalised in their family due to their condition.

Jasmine had NF1 with some cutaneous and subcutaneous neurofibromas, mild cognitive difficulties and recently, in her early 50s, she also developed MS (Multiple Sclerosis). She was diagnosed with NF1 when she was a child and was the first and only person in her family to receive the diagnosis. Talking about the way NF1 was experienced by her family she stated:

*Jasmine: well....my mum has never really accepted it we just don't talk about...you know, because my nan her mum used to say "oh it's.." not tramps disease, she called it something that make it sound like it was a very lower class in-a-gutter person's like aids is now, then ...it was an infectious disease sort of thing then, that was my nan because she was in the 1890-something you know. When I developed she was in her 80s so she didn't accept it, which it made it so my mum hasn't really accepted it*

The above extract seems to suggest that courtesy stigma<sup>23</sup> (Goffman 1968) could be one of the reasons why the mother and grandmother were marginalising Jasmine.

Furthermore, it is also possible to hypothesise a familial link between Jasmine's mother and grandmother's lack of acceptance of the condition. Jasmine suggested not only that NF1 has always been a taboo subject in the family, but she also recalled always being left alone to deal with her condition, even after she became the single mother of a child seriously affected by NF1 who required medical and psychological support. In Jasmine's account the brother seemed to be the only person in her family who had occasionally provided some help; after Jasmine developed ambulatory difficulties due to the MS, he started to accompany her to some hospital appointments. Nevertheless, in Jasmine's experience the brother seemed to have kept the general familial stigmatising attitude towards Jasmine. In fact, the experience of her brother with NF1 was described by Jasmine in these terms:

*Jasmine: he says "well I haven't got it...so what?", you know? So he just supported me recently when I had appointments for the scan for the brain and that, but other than that you know, he doesn't really know anything about it really. I think if I had a problem with one, if it if they turned cancerous or anything, then he would be... caring, but...because they are just lumps... "so what?... you have got lumps on your skin, I have got a beard, you have lumps on your skin" you know...*

As the above passage shows, the stigmatization undergone by Jasmine rested also on a familial value-system of seriousness according to which, for example, neurofibromas became worthy of notice only if cancerous.

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<sup>23</sup> A stigma acquired as a result of being related to a person with a stigma.

It is significant that the familial negative attitude towards NF1 appeared to have been absorbed by Jasmine as well, becoming an important part of her personal experience with the condition. The following passage is just one example of this:

*Jasmine: I have accepted it [NF1] ...and I can't really think positive things you know...yeah.....as I say it is just is I've got it, and I've got it and that was what my family always said " you got it"*

The lack of acceptance and discussion of NF1 within her family had probably had a significant role in the negative psychological impact that the condition had left on Jasmine. This aspect resonates with a study conducted by Bjarnason on the interconnection between parents' reactions when coming to terms with parenting a child with disability, and the child's view of themselves and their prospects as a young adult (Bjarnason 2002). In her study, Bjarnason observes that the responses of parents towards the disability of their children could influence the way these young adults experienced their lives more than the disability label itself.

### **Family as a Shield from the Impact of the Outside World**

So far, predominantly negative examples of the role of family dynamics in individual experience with NF1 have been reported. However, the family was not only a gamut of complex and problematic relations, but could also represent a shield that provided protection from the outside world for individuals with NF1 who were experiencing problems because of their condition.

As already discussed, John's body and face were completely covered by neurofibromas. These symptoms, accompanied by a lifelong experience of societal stigmatising

behaviours, critically lowered John's self-confidence, leading him to undergo serious psychological problems, including periods of depression and suicidal thoughts.

Within this distressing picture John described his family- and above all his partner Nora- as having shielded him from the outside world, significantly helping him in regaining some confidence and serenity.

*John: I hate to think where I would have gone if I hadn't met Nora, because of that protection that you are on about (my emphasis)*

Particularly meaningful were also Nora's remarks about the fact she had always paid attention to John's personality, rather than his neurofibromas; in her parlance, she had always seen "John for John".

*Nora: When I met John he already had NF. I already knew it. Already knew he had NF, but I've never looked at his NF, I've always looked at John. To me, being his wife, he was just John [...] Because it's something you learn, you look at him first, you think, yes, bit different to everybody else, but such a lovely person, you just...To me, I'm blinded; I don't see his NF at all. Not even when he takes his clothes off, it just doesn't, I just don't see it*

As the passage below shows, for having married John, Nora acquired courtesy stigma (Goffman 1968). Nevertheless, she managed to fight societal and familial pressures, and probably it was also as a result of Nora's endurance that John managed to gain some confidence and security. The last two sentences of the passage below are indicative of Nora's resistance to familial and societal stigmatisation. The reverse logic



adopted by her to fight stigmatization i.e. “if other people do not like John’s appearance it’s their problem not ours” is a motif that appeared also in John’s own interview.

*Nora: A lot of my friends said to me, “God, what do you see in him?” I said, “the person”. And my mum was one of them. And I said to my mum, “You always taught me that beauty was in the eye of the beholder. And you might not see it, but I do.” And for a long time she wouldn’t talk to John. But that was her ignorance, her problem, not mine, not John’s. But I see it as other people’s problem. It’s not John’s problem, it’s their problem. If they can’t look at it, talk about it, whatever, that’s their problem, not ours*

The following passage from Nora encloses a particularly incisive domestic example of her attempt to debunk John’s fears and anxieties about his physical appearance:

*Nora: I say to him, when I get bored with you I’ll just join the dots, play dot-to-dot. I’m really wicked!*

### **NF1 Parents ‘Using’ their Experience to Try and Help their Children with the Condition**

Parents with NF1 had the tendency to (re)engage with their positive and negative experiences with the condition to try and help their children who inherited it. I would argue that because of the event of parenthood, individuals with NF1 could give new meanings to their experience with the condition and draw on it for example to advise and support their children in many ways.

Jasmine, a mother with NF1 whose case has already been presented above, highlighted on several occasions her commitment to support her son Charles throughout all the

physical and psychological difficulties his NF1 had been causing him. Jasmine described this commitment as stemming from her desire to give Charles all the emotional and practical support she did not receive from her own family when she was a child and throughout her life.

*Jasmine: I have gone everywhere and done everything with Charles, so he doesn't have to go through what I went to*

Tim was the father of Leonard; both father and son had NF1 and they both presented with cognitive difficulties. Because of all the problems he faced with the school system, Tim could understand and help his son who was starting to encounter similar difficulties. As for Jasmine, the commitment shown by Tim represented also a route that parents had to compensate for the lack of support they received when they were younger.

*Tim: ...what my son has been through – fighting these battles and that... through – since he's been at school and everything; and knowing my experience I could put that to the teachers and say no, hang on a minute, this is how it was and I know because I've been there and I've been left to my own devices, so I could tell them how to do it – not trying to be rude or anything like that – but put it across to them how I suffered like really*

It has however to be pointed out that it was not clear whether these NF1 parents were effective in managing their children with the syndrome. As Ablon pointed out in her research, parents with NF1 were not necessarily able to manage the condition of their children, especially when they themselves presented cognitive or physical difficulties

(Ablon 1999). These difficulties could prevent parents from being ready to promptly and effectively tackle the difficulties their diagnosed children may experience.

#### **7.4 Parenting, Normative Order of the Family and Uncertainty**

Adolescents and adult people with NF1 requiring medical and/or homecare assistance could be difficult to manage for their parents because, unlike younger children, they tended to display more independence and resistance to parental authority.

Gabrielle was the mother of Dylan, a 30 years old individual who had NF1 with some cognitive, visual and motor complications. At the time of the interview, Dylan was living with his parents. Gabrielle gave an account of the difficulties that she and her husband Matt were experiencing in trying to persuade their son to have health checks. The difficulties they encountered were not only dependent on Dylan's mild cognitive impairment, but were rather the result of a process of negotiation of relationships between parents and child. Gabrielle and Matt had to find a fine balance between managing their parental worries about Dylan's present and future health and not excessively invading his personal-emotional space, giving also the fact that- as Gabrielle said- "*he feels he's so well*". Gabrielle seemed to suggest that Dylan, despite his need for assistance, was also an independent adult who probably preferred external people, other than his parents, to guide him through the "*next part of his life*". For this reason, she suggested that the external authoritative point of view of a healthcare professional may sound more persuasive to Dylan.

*Gabrielle: My only thing now would be that he [Dylan] doesn't actually see anybody. He hasn't seen anybody for years. He's down to annual check-ups with the doctor which sometimes it's difficult to persuade him that he does need an annual check-up as he feels he's so well. But he's not really checked*

*and I think there's a lot of things possibly going on underneath that we [Gabrielle and Matt], perhaps, don't know about. He does go once a year, but it is under – we have to hint. [...] So we have persuaded that perhaps it would be a good idea for somebody to have a look at it, but I feel maybe now he needs input from - I don't know - a neurologist; somebody to guide him through the next part of his life as it's a long time since he's seen anybody*

By referring to different kinds of relational conflicts with their children with NF1, parents were also reinterpreting and representing the specific problems and hardships they had to face due to NF1 using more standard motifs. Adolescence for example is normally acknowledged as a period during which parents-child relations become more problematic; parents of an adolescent with NF1 could draw on this common knowledge to partially justify and make sense of the problems they were encountering. Therefore, the idiosyncrasies related to dealing with and managing an extremely variable and unpredictable multisystem condition like NF1 could be reinterpreted and brought back to more standard parenthood motifs, like the parent-teenager conflict, or parent-adult child relations.

Claire, the mother of Leonard, mentioned some of the hardship and frustrations arising from the management of the NF1 of the son. As already discussed above, the whole family was particularly hit by Leonard's development of malignancy. In this passage Claire highlighted that the frustration arising from the everyday management of NF1 usually flowed into open conflicts with Leonard and herself.

*Claire: We did find it hard sometimes, frustrating because all the hospital appointments, check-ups, chasing up things, and that does get tiring, like,*

*here, there and everywhere all the time, and sometimes me and him have big – me and Leonard have big arguments because he’s frustrated about something and I’m trying to get him to see something and he won’t understand it and it it’s all.....and we both clash, so yeah, we find that a bit....sometimes, and it might just be his age as well – being a teenager...*

However, the fact that Leonard was a teenager was also considered by her as a possible further explanation for the difficulties she was having with him. By commenting that “*it might just be his age as well – being a teenager*” Claire brought back problems linked to the management of NF1 to more standard motifs of parenthood. Like Gabrielle, Claire provides a significant example of parental attempts to blur pathology with ‘normal’ human experience and thereby dissolving the unpredictability of NF1 into the unpredictability of ‘normal’ growing up. The passage below nicely sketches this attempt to subsume the NF1-idiosyncrasies into the more standard motifs (Landsman 2009).

### **Parents’ Expectations towards their Children and the Limitations of NF1: Finding a Compromise**

Discourses of parental expectations concerning children with NF1 could equally present as well an interesting conflation of idiosyncratic and more standard motifs. Parents often needed to find new compromises between standard desires for their children to be as happy and realised as possible and the limitation that NF1 could pose to these expectations. Gabrielle’s balance of Dylan’s life was characterised by this dialectic between desired and real. Her example is remarkable as she also had to ‘adjust’ her expectations towards Dylan and put her maternal sorrow for the problems he had in perspective because she recognised that Dylan seemed to be happy anyway.

In relation to what has been observed in the previous paragraph, it is also interesting to notice that this discourse of ‘adjustment of expectations’ is very similar to what a mother would normally be expected to say anyway about their children, independently from whether they have any impairment.

*Gabrielle: I think it's sad how it has affected him with his sight and also with his other medical little difficulties as well, but I think we are quite forward thinking. He's well. He's happy. He enjoys what he does; he's done very well. He's done the things he wanted to do. At eight years old he wanted to go to university and we said whatever you want to do we will support you in every way that we possibly can to do that and I think he's fulfilled that. I wish he could drive. There's lots of things I wish were different for him but he's content so what more could a mother ask?*

Voysey, in her study on the experiences of parents of children with disability, observed that independently from the specific impairment of the child, parents' responses were strongly informed by the official morality of child-rearing and family life (Voysey 1975). According to her, paradoxically, parents' responses did not reveal anything about the experience of having a child with disability, but were rather indicative of other's people ideas of what it ought to be like. She argued that parents interviewed, despite the difference between their situation and that of other families, adopted this modality of response because they wanted to convey an appearance of respectability and normal family life.

This tendency in parents to link impairment to something which appeals to more standard motifs of family life and parenthood may not just reflect a parent's aim to look respectable to the rest of society and gain its approval. As showed above, by drawing on

more standard interpretative frameworks (e.g. teenage problems), parents may make sense of some of the problems they are facing with their children, including uncertainty. By invoking the standard motif of parenthood of teenager-parent conflict, Gabrielle for example brought back the idiosyncrasy and uncertainty of Dylan's NF1 to something more standard and which can be comforting and can work as a tool to partially explain or make sense of some of the NF1 related problems she experienced (Goffman 1971; Richardson, Ong et al. 2007).

## **7.5 Concluding Remarks**

### **Family Dynamics and Family Communication**

In this chapter I have shown that although NF1 displays high variability and unpredictability both within and between individuals, surveillance of other family members with the condition emerged as an important and powerful tool used by respondents to manage their concerns.

Similar results have also been observed in literature around the interpretation of hereditary risk within families. Studies on cancer, especially HBOC, and other inherited disorders have shown how individuals' beliefs and knowledge of risk are shaped by the family history and by the experience of witnessing other family members with the disease (Cox and McKellin 1999; Hallowell, Jacobs et al. 2001; Hamilton, Williams et al. 2009).

Familial patterns of inheritance, the experience of caring for others, the severity of their symptoms, the number of times cancer occurred in the same person, all constitute key 'lifeworld' (Mishler 1986) heuristics which significantly contribute in shaping the personal perception of risk and uncertainty (McAllister 2001; Kenen, Ardern-Jones et al. 2003).

Individuals draw on the experiences of affected family members to construct their own perceptions of whether, when and how a hereditary disease (Cox and McKellin 1999; Emslie, Hunt et al. 2003) might affect them. These widespread practices of family surveillance, for example, have been found to play an important role in individuals' estimation of the age of onset of their (and/or other family members') cancer. This family practice clearly resonates with the accounts presented in this chapter (e.g. Claire and Ross).

The direct example of other affected family members and/or the passing down through generations of cancer family stories and legacies can contribute in shaping meanings about inheritance and uncertainty, creating a burden of anticipation (Wexler 1979; Finkler, Skrzynia et al. 2003; d'Agincourt-Canning 2005; Rolland and Williams 2005). Consequently, the presence or absence of family history and the experience of affected family members can also significantly influence individuals' risk management decisions to undergo genetic testing and to opt for surgical - or other forms of - risk management. For example, the death of a family member (or close relative) who was affected by HBOC can trigger individuals' decision to undergo genetic testing and assess and monitor their risk status or – if they are already found to be at risk- to seek surgical intervention (e.g. mastectomy , oophorectomy) or other risk minimization strategies (Hallowell, Jacobs et al. 2001; Hamilton, Williams et al. 2009).

In line with this body of literature on family experience of risk, I have provided further examples of how risk perception melds biomedical knowledge, personal embodied experience, social discourses, but also familial dynamics and experiences.

However, by tapping family networks I showed that this space and management of uncertainty within families could be interpreted differently by different members; there was no one role of this 'space'. The fact that family members' disease experience could



be different, even within a shared context, could potentially set up tensions between individual experience and meaning and that of the family (Cox and McKellin 1999).

In the same vein as Ablon's contentions, in my study the unpredictability of the disorder appeared to have a considerable impact on affected individuals and their family members who were ultimately forced to struggle as well with a *condition without parameters* (Ablon 1996; Ablon 1999; Ablon 2000). Nevertheless, differently from Ablon's research, participants in my study relied much more on the family in their attempts to seek for parameters to map uncertainty (I was made aware of this due to my family interviews). It is important to recall that- as discussed in Chapter 3- these practices of mutual surveillance, are independent from biomedical knowledge and occurred also when (and despite the fact that) the genetic disorder vary considerably within families (Cox and McKellin 1999; Featherstone, Atkinson et al. 2006; Metcalfe, Coad et al. 2008).

The strength of familial knowledge can be also justified by the fact that biomedical knowledge cannot 'solve' the uncertainty related for example to the age of onset of HBOC or HD and, even more, the extreme phenotypic uncertainty which characterises NF1.

Despite these widespread instances of mutual surveillance, the vast majority of the respondents asserted that NF1 was rarely a point of discussion within their families. They also directly or indirectly suggested that this phenomenon was indicative of the fact that the family was well-adjusted to the syndrome. Yet, the juxtaposition of the accounts of different individuals from the same family revealed that the lack of communication about the disorder could also be the result of underlying tensions within the household.

Thus, as I pointed out concerning uncertainty, the space of communication within families was multifarious; it could conflate positive and negative instances and could be interpreted differently by different family members.

This gives also more meaning to the downplaying practices discussed in Chapters 6 and 3. In fact, the juxtaposition of interviews of different family members allowed to highlight that downplaying discourses and lack of communication about NF1 in the family could be also the result of relevant familial concerns towards the syndrome.

These results support research on family communication about genetic risk and disease (Gaff and Bylund 2010). Studies which focused on a variety of genetic disorders have observed that communication within families is not always straightforward, but is generally reported to be burdensome (Clarke, Richards et al. 2005; Seymour, Addington-Hall et al. 2010).

Although respondents may acknowledge the importance to inform other family members and relatives about relevant genetic information, communication is frequently hindered by complex family dynamics such as patterns of mutual surveillance for signs of disease, moral judgement and beliefs about inheritance and disease.

As described in this chapter, feelings of guilt and survivor guilt can affect the disclosure of genetic information (Claes, Evers-Kiebooms et al. 2003). Individuals who are found to be carriers for a mutation (e.g. BRCA1/2) may prefer not to disclose this information to their parents to prevent them from feeling guilty about passing on the mutation (Green, Richards et al. 1997; d'Agincourt-Canning 2005).

Along similar lines, parents' feelings of guilt for transmitting a disorder to their children may preclude discussion of genetics within the household (Fanos and Johnson 1995; Hallowell, Arden-Jones et al. 2005). Those in the kin who have escaped a positive mutation may hold feelings of survivor guilt towards affected family members (e.g.

siblings) and may find it very difficult to talk about the disease (Tibben, Vlis et al. 1992; Michie, Weinman et al. 1998).

Deploying fatalistic discourses and attitudes towards the disease (such as the tendency reported in this and the previous chapter of seeing the disease as “*having just one of those things*”) can also be interpreted as a strategy to minimize guilt and to relieve individual and family stress (Hallowell, Arden-Jones et al. 2005; Whitmarsh, Davis et al. 2007; Keeley, Wright et al. 2009).

Overall, the vast majority of research suggests that lack of communication about risk and disease is more often dependent on a desire to protect family members and relatives from potentially distressing and harmful information rather than on poor or problematic family relationships (Brownsword, Cornish et al. 1998; Clarke, Richards et al. 2005; Hallowell, Arden-Jones et al. 2005).

In a similar vein to what was observed in Chapters 5 and 6 in relation to normalization strategies, it is possible to argue that families tend to deal with genetic risk and disease in a way apt to maximize autonomy and connectedness within family members and to minimize relationship skews (Rolland and Williams 2005; Hamilton, Williams et al. 2009; Arribas-Ayllon, Sarangi et al. 2011).

Individuals’ reluctance to disclose information about genetic risk to family members and relatives or to talk in general about genetic disease may be justified by their desire to keep balanced and serene family ties and relationships. Individuals take into account the best interests of the potential recipient of the information. Foregrounding genetic knowledge and aspects of a disease may cause conflicts and anxiety, potentially putting at risk family relationships and jeopardising family life. Anxiety, blame and conflict within families may increase the overall distress already caused by the presence of

ailments. Therefore individuals may try, if possible, to carefully think about who to talk to in the family, what to say, how and when to say it (Stokes and Hewitt 1976).

Furthermore, in line also with Ablon, this chapter has shown that a supportive family environment can play an important role on individual experience with NF1 (Ablon 1999), for example shielding affected individuals from the outside world.

Overall, by highlighting the complexity, contradictions and idiosyncrasies that characterise individual and familial experience and management of genetic disorders and their uncertainty, these findings support the idea of practical kinship (Featherstone, Atkinson et al. 2006). This confirms the hypothesis -already discussed in Chapter 3- that scenarios like Finkler's medicalization of the family (Finkler 2000; Finkler, Skrzynia et al. 2003; Finkler 2005) which envisage a reinforcement of kinship bonds due to the sharing of genetic disorders and destinies, may end up becoming too schematic and may lose important aspects of patients' and families' constructions of genetic disorders.

## **8. NF1: the Structure of the Medical Service and Patients' Attitudes towards the Syndrome**

### **8.1 Introduction**

This chapter presents the third and final part of the analysis and is based on patients' and healthcare professionals' interviews. The main aim of the chapter is to explore the role played by the healthcare system in patients' and families' experiences with NF1, which represents the third research aim of this Ph.D. thesis.

The first part of the chapter is based on health care providers' interviews (for a description of the sample see fig. 7) and addresses important problems related to the medical management of NF1. I will show that the professionals interviewed suggested

(directly or indirectly) that the NHS service around NF1 is partial, fragmented and not NF1-centred. I will also discuss their proposals about how to improve the system.

The second part of the chapter reports on patients' and families' experiences with the medical system and illustrates that their accounts reflect healthcare providers concerns towards the current capacity of the NHS to effectively manage NF1.

In the discussion, I will argue that there is a mirroring between the structure of the medical service for NF1 and patients' constructions of the condition (for example patients' downplaying attitudes towards NF1 and the lack of biosociality or biocitizenship), tracing a link between this and the previous two result chapters.

Before presenting the results, I would like to clarify that, whilst I will highlight serious problems in the healthcare management of NF1, my critique does not entail a lack of respect towards the medical service, the work of healthcare providers and more specifically, the professionals I have interviewed. I genuinely appreciated the commitment towards patients shown by the professionals I interviewed and I believe that the fact they agreed to participate in my study and that they have been willing to discuss difficulties they encountered in their profession- and more general flaws in the NHS- is in itself a sign of their awareness that something more can/should be done for patients with NF1 and similar genetic disorders characterised by high variability and uncertainty.

### **Healthcare Professionals Sample**

- Genetic counsellor (n=1)
- Genetic medicine consultants (n=2)
- NFA staff member (n=1)
- Ophthalmologist (n=1)
- Paediatricians (n=3)
- Surgeon (n=2) (surgeon1: orthopaedic surgeon , surgeon2: plastic surgeon)
- Scientist (n=1) (medical geneticist)

Total: n=11

Number of NHS Trusts involved: 4

Healthcare Professionals who worked together: Genetic counsellor 1, Genetic

Medicine Consultant 1, Paediatricians 1

Healthcare professionals who worked in a NF1 specialist clinic: Genetic Medicine

Consultant 2

**FIGURE 7**

Healthcare Professionals' Sample

## **8.2 Healthcare System**

### **Problems of Coordination and Management of Care**

As already discussed in the literature review chapter (Chapter 2, section 'Management of Clinical Problems') NF1 is a multisystem lifelong disorder with high unpredictability and variability. Individuals with a NF1 diagnosis should undergo regular monitoring, even when they are mildly affected. The minimum recommended requirement is annual

blood pressure check, since people with NF1 often suffer from hypertension.

Furthermore, clinical recommendations also state that patients should be encouraged to seek advice on any unusual symptoms (Ferner, Huson et al. 2007; Schaefer, Bull et al. 2008). Since NF1 potentially involves all organ systems, its monitoring and treatment can spread across medical boundaries, requiring the coordinated efforts of many different medical specialties.

The most common problem raised by all the professionals interviewed revolved precisely around the difficulty of offering a coordinated and effective service for patients with NF1 and their families. All the professionals observed that - as with other lifelong complex multisystem conditions (Marfan syndrome, cerebral palsy, spina bifida were the most frequently mentioned) - NF1's symptoms are often addressed separately by different medical specialties with no-one taking a comprehensive view of the disorder and coordinating the service. The NHS was generally described as being predominantly target-driven, that is, more prone to treat specific isolated symptoms, rather than considering the syndrome as a whole. Referring to the doctor-patient relationship in the clinic one of the surgeons interviewed commented:

*We probably don't spend enough time with our patients to get to know them as well as they should... you are just trying...we now got to the point when you end up- certainly as a surgeon- treating the bit of the patient that you get presented with, but not the whole (Surgeon 2)*

It was pointed out that patients and families may spend a considerable amount of time waiting for and attending numerous hospital appointments with different specialists who do not communicate to each other and may not provide patients with entirely coherent information or treatment pathways. These problems were believed, among other things,

to increase the levels of stress of family life. The following passage from another surgeon interviewed probably represents the most emphatic formulation of this problem:

*It's a scandal for me if you have a life-threatening condition, if you assemble great notes like this of a whole load of professionals who don't communicate very much with each other, each expecting these families to trot up to see them once a year or something [...] I just see this as quite cruel and unnecessary (Surgeon 1)*

Moreover, it was reported that patients and families may struggle to achieve an underlying diagnosis or, when they obtain one, they may not receive adequate monitoring and follow up, easily falling through the gaps in the medical system. For instance, interviewees expressed concern that general paediatricians and GPs, who are considered to be pivotal in coordinating patients' healthcare management, may not have an adequate level of expertise to properly manage or recognise these complex disorders (Skirton, Lewis et al. 2010). Conditions like NF1 are quite rare from the perspective of a GP or a paediatrician. The NFA staff member interviewed, who had a life-long experience as a NF1 family advisor, recalled a relatively revealing episode that can illustrate this. One of her new GPs once, to make light conversation, asked about her occupation and upon hearing that she worked with people with NF1, commented to have never come across individuals with this disorder in his career. However, she knew this doctor was actually the GP of four NF1 patients with whom she was working. One of the paediatricians interviewed, referred to NF1 as being "*a specialists' area*". He observed that NF1 patients often have quite specific needs, and he believed not to possess a sufficiently updated and specialised knowledge to be always able to properly



manage their disorder and refer them to the right specialists, in his own words, “*not to be fully up to speed with some of the subtle specialties requirements they have*”. He recalled a case he encountered to illustrate this problem:

*I had a recent case where a child was... not growing well, he had a growth delay and I had no idea that children with NF1 can have growth hormone problems and may benefit from growth hormone treatment, and I wasn't aware of that, and it was only when I looked into it, I saw a publication in 2006 and it kind of flagged up to me how... you almost need to be really knowing what's happening, because that was a real issue for that child, you know... anyway it turned out he had this growth hormone checked and it was ok in the end and he didn't have treatment, but if he had required treatment I would have felt a bit bad... (Paediatrician 1)*

Like this paediatrician, the majority of the professionals considered clinical genetics to be the medical speciality with the broadest and most updated level of expertise and understanding of NF1. Clinical genetics was described as possessing a “*better treatment capacity*” (Paediatrician 3) which encompassed explaining the nature of the disorder to the patients, monitoring its development and, when appropriate, referring the patients to the right specialists. Therefore, this paediatrician and the other non-geneticist healthcare professionals interviewed tended to delegate the responsibility for the management of NF1 patients to clinical genetics units.

However- as highlighted also by the genetic counsellor and consultants interviewed - clinical genetics is mainly a consultation service which is normally involved in diagnosing, taking family history and informing patients and families about the condition and its inheritance. Genetic counsellors do not tend to get involved in on-

going counselling and genetic consultants do not check patients and are not normally aware about whether patients are followed up by other physicians. As the genetic counsellor interviewed observed, dealing with the condition on an everyday basis (which she considered to be “*the bulk of the concerns of families and patients*”) is the task of GPs, paediatricians or other physicians:

*Our role is a very peripheral role [...] we are there to help them [the patient] adjust to the diagnosis as best as they can and then we step back and then the paediatrician, the physician or whoever is caring for that particular condition then tends to take over* (Genetic Counsellor)

Similarly, one of the genetic consultants interviewed, in describing the general role of her discipline in the management of NF1 patients, stated:

*We are not really involved in the annual check, we see them (the patients), confirm the diagnosis, give them the information, but then tend to refer them to...if they are children the paediatrician will follow them up, if they are adults we would just recommend they go to their GP for annual blood pressure check and make sure they are aware of what are the symptoms to look out for. So we wouldn't chase them up, we wouldn't know in fact whether they have had blood pressure checked or not* (Genetic Consultant 1)

Thus, because of this organization of the healthcare system, NF1 patients and families may not be adequately managed and monitored. In fact, patients may end up finding themselves in a loop between GP, paediatricians or other physicians- who are supposed to be the main carers, but may not always have the sufficient and updated level of

expertise to perform this task- and clinical geneticists, who possess this expertise, but are not normally involved in the management of the disorder (see fig. 8).

Similar issues of fragmentation of care for NF1 patients have been found in other studies conducted in the UK (Ponder, Murton et al. 1998; Huson 1999; Evans 2011).

### **Multidisciplinary Clinic**

As a possible solution to these problems of coordination and management of care sketched above, all the professionals hypothesised that patients and families would be better managed by NF1 multidisciplinary clinics. In this setting, patients would be able to see a team of different specialists during the same day and in the same clinic.<sup>24</sup>

It was also observed that a similar model could work for other conditions that involve many specialties. Medical genetics, given its level of expertise about complex multisystem disorders, was recognised as being the specialty placed in the best position to organise these clinics and coordinate the team of specialists (a similar suggestion surfaced in other studies about NF1 and other genetic disorders, for example in (McAllister, Payne et al. 2007)). Nevertheless, the point was also made that the professional figures that could coordinate the other doctors in the clinics do not necessarily need to be geneticists, but could come from any medical background, provided that they had an interest in NF1.

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<sup>24</sup> At the time of this PhD thesis there are three multidisciplinary NF1 clinics in the whole UK: in London, Manchester, Plymouth. The one in London appear to be the largest in UK with around 600 patients: source <http://www.kch.nhs.uk/services/neurosciences/neurosurgery/neuro-oncology/?entryid2=5344> London and Manchester are the 2 national complex NF1 services in UK launched in April 2009 <http://www.specialisedservices.nhs.uk/service/complex-neurofibromatosis-type-1>.

## **Making a Business Case**

Some professionals reflected on the possible hindrances that the realization of NF1 specialists' clinics could encounter. One paediatrician interviewed, for example, considered the highly variable nature of NF1 to represent a possible obstacle for the realisation of this project. He observed that the fact that the management of the disorder requires many medical specialties may not "*make a business case for a coherent and all-embracing service*" (Paediatrician 3).

Moreover, he pointed out that specialists' clinics may introduce some form of tension within healthcare settings with respect to the management of other health problems. Focusing on one specific condition can, for example, take attention and resources away from other more common disorders like diabetes, cardiovascular disease and cancer, which are among the main NHS-set priorities.

The scientist interviewed was working within a NF1 specialist clinic which, in her view, has been closed, owing to a decrease in political interest in the condition: "*It is a political problem, there isn't any more interest in NF1, or there is interest for other conditions or problems*" (Scientist). Nevertheless, as she also observed, it is important to highlight that NF1 is a relatively common condition, and since individuals with NF1 can develop a variety of tumours, investing resources towards the research on this syndrome would ultimately benefit cancer research, which is one of the NHS set priorities:

*The first thing about NF1 is that you find lots of patients suffering from it, and the second thing is if you are working on tumours, you are working on cancer you can really gather access to multiple types of tumours from the same patient [...] so you can really get tumours which are related to different organs in the body at the same time, so you get lot of material for your research* (Scientist)

NF1's link to cancer research, as well as its commonality, may therefore help in terms of making a business case to government and funding bodies to support specialists' clinics, and other infrastructures for research into this condition (See also (Donnai 2009) based upon the importance that knowledge of monogenic disorders has for affected families, clinical care, biomedical research and the general public).

### **Early Diagnosis and Intervention**

Merging an awareness of patients' needs with an appreciation on the emphasis on financial savings in government, the NFA staff member interviewed stressed the crucial importance of early diagnosis and intervention.

Alongside supporting NF1 specialists' clinics, she suggested that managing to diagnose children within their first year of life and planning the specialists they will need to come across in the subsequent years could "*change the whole face of NF1.*" She argued that an early diagnosis accompanied by the right support, for example occupational or physiotherapeutic, would decrease the impact of the syndrome on patients, bolstering their health outcomes and quality of life. Moreover, in case of cognitive problems, securing appropriate educational support (e.g. one- to-one teaching, learning assistance) from the beginning schooling would significantly benefit children's development and future employment prospects. For these reasons, the enhancement of diagnostic and support tools, she claimed, could dramatically change the lives of NF1 patients and their families, not only saving on welfare and benefits, but for a much lower cost than, say, heart disease.

*If we could make a politician to take on board that good diagnosis and support of a child with NF would actually save the country millions and*

*millions and millions, because these people won't become dependent, they'll be independent and at the moment we have got lot of people have been assisted quite rightly through the educational system, but then get poor career advice and then started a job in which they should never got into in the first place, because they are not fast enough to be able to do that, because I mean most of the jobs that do not require high qualifications, tend to be very fast, the places like supermarkets or, McDonalds or whatever, they want quick responses, you don't get that from somebody with NF. So they might start to work but they actually don't finish much unless they have a really caring employer. So then they go into unemployment, then they go into long term unemployment, then they become just a drain on society, and I think if you could stop that you could save the country much more than heart disease costs (NFA Staff Member)*

Drawing also upon her long experience as a NF1 family advisor, the NFA staff member remembered having seen many “angry parents” who were worried about their children since the day of their birth, but had to wait several years before obtaining a diagnosis because their concerns were not validated by doctors. On average, she claimed that, unless the child is born with a serious characteristic symptomatology, some problems are only spotted by the teachers when the child begins primary school and this slowly leads to an assessment of the child and to a final diagnosis.

The scientist corroborated this experience, highlighting the lack of knowledge about NF1 of GPs and other physicians. She suggested that, in order to reduce these problems, there should be annual NF1 meetings in the UK to educate healthcare professionals and other health workers and update them in the new developments in the field.

## **Between Duty of Care and Patients' Autonomy**

Whilst considering the idea of introducing more NF1 specialist clinics, some professionals directly or indirectly challenged this proposal in the light of the principles of patients' autonomy and empowerment already evoked and discussed in this thesis (see chapter 3).

One of the genetic consultants interviewed suggested that an advisable way to enhance the service would be to “*hand over*” to the patients the responsibility for the management of their diseases.

*If a patient isn't having too many problems and they are educated about what to report, then they don't necessarily need to see anyone and I think there has been a lot of work nationally, although I don't know whether it has been rolled out yet, where patients have their own records with information, so I think that's a good way...hand over the responsibility to the patient for their own condition rather than they coming to see different registrars in the hospital every year and being given different information*

(Genetic Consultant 1)

In contrast to the idea of educating health care professionals, this consultant envisaged that it is the patient, rather than the professional, who needs to be more educated and updated about the disorder. In other words, the consultant's suggestion entails that the healthcare system's problems of uncoordinated and insufficient care can be tackled by giving responsibility to the patients and letting them increasingly become the new coordinators of the care received. Patients who are mildly affected- for example- may decide to directly step out of the medical system, avoiding potential stress caused by

attending a number of appointments with different doctors who may provide no coherent information or treatment pathways.

These remarks are strongly in line with NHS and broader public healthcare discourses which deploy the rhetoric of patient autonomy and empowerment.

One of the paediatricians interviewed pointed out that prior to offering NF1 specialist clinics the actual demand for it from the service users should be assessed. In fact, even if theoretically advisable, NF1 specialist clinics may not fit patients' needs. Agreeing with the genetic consultant quoted above, this paediatrician affirmed that patients should primarily be empowered with information on their disorders.

*We have to be very careful about sometimes imposing strict regimes on people when it doesn't necessarily fit their needs, they might argue the GP can check their blood pressure and if they feel... headache or something, you know, they know there can be risk for brain tumour... they will respond. We have to be very careful sometimes not to be too paternalistic... at the end of the day I think the parents become the experts, or should be really, so they should be empowered with all the information, so that they know exactly what it needs to be done; they can dip in and out if they wish to follow a clinic- I don't think they must come to it- and they can pick what they want to be done [...] you know what I mean? Going with them a bit more that way rather than stigmatise (Paediatrician 1)*

This way, patients would be placed in a better position to autonomously decide whether and how to manage their disorder in partnership with healthcare professionals. Thus, like the consultant, the paediatrician argued that informed patients, rather than attending a specific NF1 clinic every year, may prefer to simply ask GPs to check their blood



pressure. Furthermore, being educated about the possible complications of their condition, patients would be able to keep their bodies under surveillance, seeking the help of the right specialists when needed. For example, if they experienced a severe headache, knowing about their increased likelihood for developing brain tumours, they would seek the help of a specialist (possibly a neurologist). Offering NF1 clinics without checking the actual demand, would overrule the autonomy of the patient; in the paediatrician's terms, it would be "*paternalistic*".

The paediatrician also worried that requesting NF1 patients to undergo annual monitoring could represent a violation of their coping strategies. Patients may decide to downplay or ignore their disorder and consequently could feel the annual follow up schedule as a stigmatising imposition or a form of labelling. Remarkably, it appears that this paediatrician's worries were also linked to the fact that he did not believe he possessed a sufficient level of expertise on NF1 to justify to his patients the necessity of an annual health check-up.

*I have sensed a bit of stigmatization in that process as well, being dragged back for follow up every year [...] in the sense, they (NF1 patients) might not want to be followed up, they might prefer to ignore their problems...I think if you are going to drag them back every year you need to justify it, you need to have a very clear reason for doing it and a level of expertise that befits that (Paediatrician 1)*

The principles of patients' autonomy and empowerment touched upon by the consultant and paediatrician above resonate with general objectives in the NHS to move towards a more patient-centred service. These same principles also appear to be part of the crucial tenets of the last White Paper 'Equity and Excellence: Liberating the NHS' published

by the Department of Health in July 2010 (DoH 2010). In this document, the aim of enabling patients to be more involved in the management of their health is clearly expressed and considered to be of great importance.

The paragraph reported below from the section emblematically titled ‘Putting patients and the public first’ stated that the principle of ‘shared decision making’ - - should become the norm. This move, it is argued, would enhance the quality of care increasing the levels of patients’ compliance.

*2.3 We want the principle of “shared decision-making” to become the norm: no decision about me without me. International evidence shows that involving patients in their care and treatment improves their health outcomes, boosts their satisfaction with services received, and increases not just their knowledge and understanding of their health status but also their adherence to a chosen treatment.*

Like the consultant and paediatrician’s discourses, the aim of promoting the principle of shared decision making is intended to be achieved by giving to the patients more information about their disease and treatment pathways.

*2.6 The Government intends to bring about an NHS information revolution, to correct the imbalance in who knows what. Our aim is to give people access to comprehensive, trustworthy and easy to understand information from a range of sources on conditions, treatments, lifestyle choices and how to look after their own and their family’s health.*

However, there can be a deontological tension in clinical practice between the duty of care and respecting patients’ autonomy. The concern expressed above by the paediatrician about the possibility that NF1 patients may feel annual monitoring to be stigmatising provides a good example for discussing this tension. The autonomy of the patient is an important principle and doctors certainly cannot force NF1 patients to

undergo annual screening. Nevertheless, monitoring is in the interest of the patients, because it offers in many cases the opportunity to find symptoms and intervene at an early stage, preventing serious or damaging complications. Prevention and early intervention can be crucial for a syndrome that can cause, among other things, malignancy, bone lesions and cognitive difficulties. A similar point is made by the scientist who, referring to the need to monitor patients stated:

*It is important to get them back every year and check them, it allows to screen out abnormalities' 'if there is an indication of say optic glioma, then it can be diagnosed at an early stage and can be treated, but if you, if things are left too late, then this probably becomes quite damaging for the patient*  
(scientist)

According to the scientist, the responsibility for health management should primarily be the duty of healthcare professionals before being the duty of the patient. Therefore, the simple act of advising patients to undergo regular monitoring does not necessarily have to be characterised as an imposition, but simply the actualization of health care professionals' duty of care towards the patient. In particular, the scientist observed:

*Patients should appreciate that, because if nothing is found, this should be encouraging for patients and families, if something is found then it is treated at an early stage, which again is something positive for patients and families* (scientist)

Recalling the principle of shared decision making, it can be argued that empowering patients with more information about their disorders could at least partially resolve this

deontological tension that healthcare professionals may face between respecting patients' autonomy and their duty of care. In theory, through the '*information revolution*' envisaged in the White Paper and evoked by the paediatrician and consultant above, patients with NF1 themselves would be more aware of the importance of annual check-ups and the consequences of not monitoring their health. Thus, they would be placed in a better position to be able to responsibly choose whether and how to manage their disorders and health.

However, as already discussed in the literature review (Chapter 3), asking individuals to increasingly take responsibility for their health, whilst giving the appearance of empowerment, can also become an obligation. If patients have this information and have been taking responsibility for managing their health in relation to this information, then they are probably expected to do so and if they do not fulfil this expectation, society may consider them to be responsible for the condition they have and/or individuals themselves may feel guilt for developing a condition or not managing it properly (Petersen and Lupton 1996; Petersen and Lupton 1997; Hallowell 1999; Kerr and Shakespeare 2002; Petersen and Bunton 2002; Rose 2006).

Moreover, normative and deontological implications of these principles aside, their very establishment could encounter complex challenges. In NF1 and similar conditions patients and families may have cognitive and learning difficulties. Educating patients and increase their responsibility for the management of their health could be problematic.

There can be physical-cognitive and social obstacles that may prevent patients from understanding the information about their disorders and/or acting upon it. For example, as the NFA staff member observed, although patients receive letters from hospitals regarding appointments or follow ups, lots of them are not able to attend due to admitted or un-admitted illiteracy and other cognitive and physical barriers. It is likely

that many of these NF1 patients are missing important appointments and, unless there is a good collaboration between their GPs and other specialists to properly address these problems, the healthcare system may lose track of them.

*The levels of illiteracy amongst patients with NF1 admitted or un-admitted is really high. Hospitals send out written appointments to patients who then can't read, won't admit they can't read, don't attend or they have very poor short term memory, so they forget they've got a hospital appointment, so they do not attend the NF clinics [...] I would like somebody at some point to actually do an in-depth failure to attend NF clinics and see (NFA staff member)*

Thus, conditions like NF1, which can cause physical and cognitive difficulties, provide revealing examples of how the idea of patient empowerment and autonomy may be problematic.

### **Reaching the Patients: Team of Travelling Professionals**

One of the surgeons interviewed proposed a model for the enhancement of the delivery of care that might also reduce patients' failure to attend clinics. He suggested the creation of outreach services in which teams of specialised healthcare professionals' could travel to the patients, covering different local hospitals, rather than patients having to travel long distances to the specialists located in clinics:

*There is a tension between concentrating a lot of patients in one place and therefore getting a high level of expertise and specialization and the opportunity for research, and delivering care for the convenience of the*

*patients... and one way of, perhaps, of addressing that is to have the team of specialists who travel to be closer to the patients, [...]so that the patient can go to the local hospital, but they can receive high standards specialist care, and I think that sort of model would be very suitable (Surgeon1)*

However, whilst providing better access to care for patients, outreach services in practice could be a very challenging project to realise. It would increase for example specialists' travelling time and the time spent away from their hospital base. Moreover, it may be logistically difficult to organise the specialists' team travels. Thus in terms of costs, processes, and time management it may be easier to concentrate all the specialists in one place rather than organising outreach clinics.

### **Family Advisors**

Another idea for making support more accessible to patients was suggested by the NFA staff member and the genetic counsellor who were interviewed. They both stated it would be beneficial for patients to have more NF1 family advisors who could visit them in their own home at their own time. Family advisors are NF specialist advisors who work regionally within hospitals and give support to anyone affected by NF1. They normally come from nursing or other medical backgrounds and are given further training by the NF foundation in order to be able to provide support to NF1 patients and their families following a diagnosis or other stages. The main functions normally covered by the NF1 family specialist advisors are the provision to patients and families of updated information about the condition at their level of understanding, emotional support, guidance on educational problems including liaising with teachers, educating healthcare professionals as well as acting as a link between families and specialists.

Family advisors are also very family-focused as they normally see the whole family in

the community. Currently, there are few NF1 advisors in the UK. As the NFA staff member pointed out, in the year 2004 there were twelve specialist nurses in the whole country, at the time of her interview (year 2009) only five. This is mainly due to lack of resources.

Family advisors can act as important gatekeepers and coordinators for medical and educational support. Furthermore, by visiting families in their homes and listening to their problems, family advisors can also offer emotional support, which is equally important.

### **Dealing with Family Dynamics**

Paediatricians and genetic service providers, who normally deal with patients and families, observed that familial dynamics influenced the way NF1 patients experience their condition and the clinical management itself of the patients. This is in line with other research discussed in Chapter 3 about the important role of familial and biographical elements on patients understanding of biomedical information and construction of disease (Parsons and Atkinson 1992; Cox and McKellin 1999; Skirton 2001; Featherstone, Atkinson et al. 2006).

The genetic counsellor observed, for example, that when there is a family history of NF1, the way individuals construct the syndrome often depends on the specific symptomatology and degree of seriousness of the already affected family members:

*If they (the NF1 patients) have a member of the family affected by multiple tumours...disfiguring facial neuromas, their experience is different from somebody who just had some café-au-lait spots...or learning difficulties*

(Genetic counsellor)

She broadly named these syndrome-related familial dynamics “*family myths*” and “*legends*”. Throughout her experience as a counsellor, she observed that familial beliefs about the syndrome can be, like legends, very pervasive and stay with patients despite advice in counselling sessions (Emslie, Hunt et al. 2003; Featherstone and Atkinson 2012).

Acknowledging similar familial dynamics and their relevance in clinical practice, one of the paediatricians interviewed claimed that when there is an already known case of severe NF1 in the family it is not uncommon for a physician to avoid disclosing that a new born (who is mildly affected) has the condition to other family members in order not to worry them too much:

*If somebody else in the family system has got serious, severe, perhaps life threatening problems with NF1, the professional is afraid that by... placing attention on the relatively well person, then... everybody would be misled*  
(Paediatrician 3)

As already discussed NF1 is highly variable even within the same family. Hence, the fact that a family member has a serious case is not predictive for the degree of severity of new affected generations. However, serious cases in the family normally become the familial archetype for the condition (see chapter 5 i.e. family experience with NF1 dominated by the affected father). Therefore, the physician’s concern is that parents and family members would associate the NF1 diagnosis with their familial archetype of the syndrome and think that the new born would end up being as seriously affected (Skirton and Patch 2002; Featherstone, Atkinson et al. 2006).

One of the paediatrician interviewed (Paediatrician 1) stated that the management of NF1 (and many other genetic problems being discovered in children) is “*tricky*” as it



involves the parents more than the child. The management of these conditions has to be negotiated with the parents.

The NFA staff member observed that during the teenage years NF1 patients could be difficult to manage because, in contrast to young children, they tend to display more independence and resistance to parental and other forms of authority. According to her, teenagers' tendency to resist authority coupled with the reduction of services and follow up for adult patients make the transition between paediatrician and GP a potentially delicate period (Skirton and Patch 2002; Davidson 2010).

*Children with NF got a very good service, particularly if they have motivated parents, the problem comes at the stage of transition between 16 and 19 when they sort of don't want to go along with any suggestions whatsoever. They are stopped been seen by the paediatric services [...] and then the adult services don't pick them up, or they've got a lot of help at school [...] and then they just get thrown out and that's one of the problems*  
(NFA Staff Member)

One of the genetic consultants interviewed (genetic consultant 1) explained that when a patient is affected and the family history is unknown to the clinic, the geneticists discuss with the patient the importance of assessing other individuals in the family and encourages the patient to speak to their relatives. It is, then, the patient who normally has to contact other relatives. Therefore, the process of disclosure is performed by and filtered through the family and its complex dynamics. The genetic consultant commented for example that disclosure can be problematic when patients "*have fallen out with their relatives and they don't know where they are*". In this case, she commented, "*you come up against a dead end*". This clearly resonates with what was observed in relation to family communication in Chapter 7, that is communication

within families and disease is generally burdensome and frequently hindered by complex family dynamics, moral judgement and beliefs about inheritance (Clarke, Parsons et al. 1996; Elwyn, Gray et al. 2000; Clarke, Richards et al. 2005; Edwards, Gray et al. 2008; Seymour, Addington-Hall et al. 2010).

Family clinical genetics appointments would seem to be an advisable strategy to explore some of the familial dynamics and issues that seriously threaten an adjustment to a genetic diagnosis or predisposition. However, the consultant pointed out that family appointments could also be unproductive, since they may be skewed by dominant family figures and consequently they may not allow all the attendants to speak, ask questions and express concerns:

*We often see several people from the same family in one clinical appointment, but that can be problematic because different people will have their own questions and different agendas and you can't really focus on several people at the same time, it's always the one that has the most to say that gets the most out of it. We would try to avoid that while possible, or at least followed it up with one-to-one, or maybe a couple, rather than the whole family (Genetic consultant1)*

### **8.3 Patients and Families' Experience with the Healthcare System**

#### **Benefits of Diagnosis**

Despite the unpredictable and uncertain symptomatology associated with NF1, some participants regarded receiving a diagnosis as having a positive effect on their lives. Claire observed, for example, that the diagnosis of her son Leonard was crucial to obtaining educational assistance in his primary school:

*Claire: Yeah, the teachers didn't understand it or – until we got the medical side helping us, you know, they didn't realise there was a problem so much, and then gradually he did get a – we had a great teacher – primary school teacher; she really stuck by us, with us and helped us to get him statemented in the end. In the end we had a school educational.....oh, I can't think of what it's called*

This findings are in line with research which explored parental experiences of raising a child with undiagnosed genetic conditions (Rosenthal, Biesecker et al. 2001; Lewis, Skirton et al. 2010). Parents in these studies reported that the main reasons for wanting a diagnosis were related to the improvement the treatment, care and management of their children condition, including help with schooling and access to other forms of support. The medical label and the involvement of healthcare professionals prompted the school to recognise the 'reality' of Leonard's learning difficulties and plan some educational intervention. Drawing a parallel between Leonard and his father Tim, the case also highlights the potential benefit of a NF1 diagnosis. Despite his serious symptomatology, Tim was discovered to have NF1 only as a consequence of Leonard's diagnosis. Hence, Tim spent a considerable part of his life facing difficulties without receiving any recognition and support. Lamenting the incapacity of the school and healthcare system to recognise his difficulties, Tim described his school years by adopting particularly negative tones:

*Tim: It wasn't good at school at all – no. really just left to one side – basically because in those days they didn't seem to pick up on anything and so I was always told to try harder and basically left to it, and then come by the end of school with exams starting, I wasn't no good at anything, and I*

*just got told, well you're not being put through anything, so I had no exams or anything when I left school*

Gabrielle explained that the diagnosis of her son Dylan ended a frustrating and difficult period of tests and queries. As exemplified in her passage below, knowing eventually the name and typology of Dylan's disorder served as "a base to work from", helping Gabrielle and her husband Ethan to deal with it:

*Gabrielle: I think my husband found it quite frustrating at the beginning when they couldn't find a diagnosis because I think with everything, when you are going through tests - that's one of the most difficult times. When you have a diagnosis it's almost a base to work from. You can start looking at things [...] we deal with things much better if we know exactly what it is and what we're dealing with*

### **Lack of Straightforward Diagnostic Pathways**

When asked about the pathway of diagnostic experience, the majority of the participants described the process of reaching a diagnosis as not easy. Ethan, for example, stressed that Dylan was eventually diagnosed by clinicians in a hospital in London only after a long period of unsuccessful appointments with different consultants from other hospitals. He complained in hindsight about the delay in his son's diagnosis, pointing out that Dylan presented relatively typical characteristics of NF1.

*Ethan: We got off to a bad start to be fair, because no-one was able to make a diagnosis. We went to three different hospitals to see three different consultants before we went to X (name of a London hospital) who made the*

*diagnosis immediately you know, and when you think about it, it's not a difficult condition to diagnose compared to some neurological disorders. He [Dylan] had all the fairly classic characteristics of NF, so that was – we had – a pretty bad experience to start with, but then obviously X (name of a London hospital) changed all that*

Referring to the diagnostic pathway of her brother Robert, Alice described a similar experience of delay:

*Alice: it wasn't a straightforward diagnosis, I don't think people really knew what it was, so he had several appointments and several different consultants were involved, and finally...yeah they realized what it was...I think they had to, I am not completely 100% sure, but I think they had to take a small biopsy as well*

Thus, whilst diagnoses were often seen as being useful, it was also common amongst families and patients to have experienced difficulties in obtaining them. This turned out to be mainly due to a lack of expertise among primary and secondary healthcare providers who tended to be the first point of contact that families had with the healthcare system.

### **Healthcare Professionals not Knowledgeable about NF1 and not Enough Supportive**

When asked about their experiences with the healthcare system, patients and families reported that a general the lack of knowledge had prevented healthcare professionals from offering support after diagnosis.

*Alice: I think people (healthcare professionals) were very frightened because they didn't know enough themselves to be honest...there was a feeling that we, well my mum, couldn't have all of her questions answered, and definitely a feeling that my mum and my father had to do a lot of research themselves*

In Alice's recollection, after the delayed diagnosis, healthcare professionals appeared to be 'frightened' by Robert's NF1 and incapable of adequately informing and reassuring his parents. Alice's family appeared to be partially abandoned by the healthcare system. One of Martha's GPs was involved in diagnosing her NF1: "*he was the one who named it*". However, he did not appear to be able to answer Martha's questions about the syndrome. Moreover, this GP delegated the responsibility to obtain information about the syndrome to Martha:

*Martha: the doctor I've got now, at the time he was the one who named it [NF1], but he didn't really know anything about it, he said "you have to look for support groups or find out about support groups" he said "if you go on the internet you can find more about it"*

Paige became aware of having NF1 after being referred to clinical geneticists following her grandchildren's diagnosis. During her interview, she mentioned several times that her GP '*does not believe in NF*'. Recounting the GP's reaction on comprehending her NF1 diagnosis Paige, for example, stated:

*Paige: I don't think that ordinary GPs seem to really know very much about NF. [...] My GP, when I went, she said "well everybody's got NF! If you had a blood test everybody would have NF!"*

Thus, certain healthcare professionals were not only unable to provide information about NF1, but they could also deny the very existence of the disorder, possibly undermining patients' illness identity.

Relying upon her long experience both as a NF1 patient and as part of the NFA staff, Nicole pointed out that many of the NF1 patients she dealt with knew much more about their condition than healthcare professionals. Moreover, she claimed that not only doctors, but also health visitors should be better educated about the syndrome. Health visitors are the nurses and midwives principally involved in supporting families and young children around the ante-natal period. During the child's early months and years, they can represent an important bridge between families and the healthcare system. Therefore, Nicole argued, these nurses should be trained to individuate early signs of NF1 and, if needed, make sure the child is referred to the right specialists. In the following passage Nicole recalled an experience she had when she was helping a family of a child who presented the signs of NF1 to get a diagnosis. This example highlights how the health visitors' lack of familiarity with NF1 can pose an obstacle for families attempting to reach a diagnosis:

*Nicole: I showed cafe au lait patch to a nurse and she, and to an health visitor, and she said 'oh they're nothing, they're just birth marks', and they were more than that, the mother knew they were more than that and yet the health visitor just poo pood it...I have had that.*

Edward underwent considerable difficulties in getting his GP to validate the symptoms and concerns he had about his health.

*Edward: I get a lot of light headiness and dizziness, and things like that and I was told 'oh this is one of those things that you have to put up with' and I thought NF maybe I have some knot which is growing in the middle, my wife she's got 2 lesions in her head...for a couple of years I had it and I kept going to my doctor and "there is nothing wrong with you, you are imagining it all" [...]. One day 2 or 3 years ago everything span around, I fell down...I was diagnosed with benign positional vertigo*

Reflecting on her personal experience with different healthcare professionals and that of her partner John and other affected family, Nora raised similar important issues. She claimed that healthcare professionals have tended to display a rather impersonal and superficial approach towards NF1 patients. Doctors do not seem sufficiently to acknowledge NF1; consequently they do not scrupulously investigate and inform NF1 patients and, finally, do not provide adequate follow up and support.

Nora believed that possible causes for the service deficiency could be healthcare professionals' general lack of familiarity with NF1 and the high variability of the syndrome which can make its clinical management very challenging.

*Nora: the doctors just say, "Yes, you've got neurofibromatosis", that's it, end of story. Then John will go up and say, "Can we remove this lump, remove this lump"- "Yes, OK". And that's it. They've not - because they don't know enough about it, they don't go into great detail about it either. They don't give you help and support like you would imagine that they*



*would. They don't, because they don't know how to deal with it and probably because there's so many different variations of it*

Moreover, alongside the medical aspects of the syndrome, Nora emphasised the importance of psychological implications, which, once again, do not appear to be acknowledged and dealt with by the medical staff. Nora and her family members were able to eventually find some psychological support for NF1, but only after managing to get referred to a clinical genetics specialist. However, obtaining this referral was not easy; her affected family members had to insist several times in order to convince their GP. Thus, according to her, strengthening the links between GPs and clinical genetics departments could potentially help patients and families to obtain a level of support.

*Nora: I honestly feel that, the GPs, it's easier for them to turn round and say, "Yes, you've got NF", and be done with it. I don't think they realise the mental effects it has on people. I know from living with John that it's had some devastating effects at times on him, and I also feel that they should either look into it more or be more prepared to help in maybe counselling or bring more people like Dr X (genetic consultant) in, or, you know, I just think that they should try and help more. If I had scabies then they'd say, "Here you are, we can do this, this and this". They can't do that with NF. But they could say, "You have got NF and it can get really bad, I'll put you in touch with Dr X- or whoever might be available at the time- and he can go through it with you". But it was (name of her daughter in law) that brought Dr X in, in the first place. It was (name of her daughter in law)'s persistence at her doctor that brought Dr X in*

Like the vast majority of the participants interviewed, Nora called for a significant improvement in the healthcare service for NF1 and, essentially, for more awareness and recognition of the disorder among doctors. The parallel she traced with scabies is particularly revealing. Whilst GPs and other doctors are able to manage patients with scabies and offer them standardised treatment pathways, they do not seem to be able to respond in a similar way with NF1. As reported by many respondents, GPs and physicians do not appear to be familiar with the syndrome, consequently they are not always able to offer support and they also appear to be prone to disregard or play down NF1 patients' concerns. Clinical geneticists are instead regarded as the professionals who possess expertise in the condition. However, as illustrated in the section of this chapter 'problems of coordination and management of care', clinical genetics departments mainly provide diagnostic and information giving services and do not tend to see patients regularly and to provide an on-going service. In this health system it is the GP and some cases other physicians, who normally have to deal with the day to day symptoms and the bulk of the concerns that NF1 patients experience. Nevertheless, GPs and physicians do not seem to always be able to recognise and tackle these symptoms and concerns. Therefore, patients and families may be trapped in the loop described above and may experience their NF1 as a 'medically orphaned' condition.

#### **8.4 Concluding Remarks**

This chapter has argued that the healthcare system around NF1 is partial and fragmented. The minimal clinical recommendations for NF1 patients, independent from the severity of their condition, comprise an annual blood pressure check and review of any unusual symptoms (Ferner, Huson et al. 2007; Schaefer, Bull et al. 2008). Early detection of symptoms facilitates early intervention which, in turn, can dramatically change the life of patients, both paediatric and adult. Yet, both the healthcare

professionals and the patients interviewed lamented a lack of coordinated and effective care. NF1 patients and families often experienced difficulties in obtaining a diagnosis and, once diagnosed, they could find themselves in a referral loop, falling between genetic service providers and other primary and secondary healthcare providers.

As already pointed out, these findings are in line with what observed by Ponder, Murton et al.'s (1998) research on NF1 conducted a few decades ago in the UK

Furthermore, the problems of partial and fragmented care are not only related to the management of multisystem syndromes, but they represent part of a broader need that many European and Western countries are increasingly facing to restructure their healthcare services.

With the rapid advancements in genetic medicine, genetic knowledge and technologies are increasingly moving into clinical practice (HGSG 2012). These advancements and the consequent demands posed on clinical genetics units have led to discussions about the future provision of genetic services within the healthcare system (Emery, Watson et al. 1999). Genomic technologies are not only enhancing the capabilities within clinical genetics, but can be 'mainstreamed' in every medical specialty. Genomic sequence data is already used in the care of oncology, cardiology and paediatric patients (Charron and Elliott 2011).

However, research and systematic reviews conducted across different European and Western countries converge in reporting that the number of healthcare professionals who are 'genetically literate' (Petersen and Bunton 2002; Bunton and Petersen 2005) does not meet the demand for genetic services and counselling.

For examples, studies have shown that whilst primary carers (e.g. GPs, paediatricians, obstetricians) accept that they have an increasing role to play in genetics, they also

appear to lack confidence in their ability to do so because of limited knowledge of genetics (Emery, Watson et al. 1999; Skirton, Lewis et al. 2010).

Primary care providers are often unable to properly interpret genetic test results ordered by them, to communicate these results to patients and are also unaware of the ethical, legal and psychosocial issues that arise around genetic testing (Hunter et al 1998; Emery Watson et al 1999; Geller 1999).

In particular, as concerns diagnosis, preventative and treatment options, in the UK the Confidential Enquiry into Counselling for Genetic Disorders by non-geneticists (CEGEN) (Modell, Harris et al. 2000) reported that non-genetics healthcare providers tend to be much more focused on the medical treatment of specific immediate symptoms rather than on genetic counselling, screening and monitoring likely to prevent the emergence of future problems in the patient and their relatives. This again clearly resonates with the fragmented and partial care around NF1 highlighted in this chapter.

It is important to point out that the increasing process of mainstreaming of genetic medicine in medical practice urgently requires medical specialties to be more educated in genetics (Skirton, Lewis et al. 2010; HGSG 2012).

Regular updates, educational programmes and referral guidelines, may provide useful methods of supporting the new field of mainstream genetic care (Emery, Watson et al. 1999; Skirton, Lewis et al. 2010). A common minimum standard of competence in genetics for healthcare providers working in primary, secondary and tertiary care has been developed in Europe (Skirton, Lewis et al. 2010).

The findings in this chapter have significant implications for the discussion of genetic subjecthood and citizenship (Rabinow 1996; Petersen and Bunton 2002; Taussing, Rapp

et al. 2003; Rose and Novas 2004). Chapter 6 reported a widespread tendency among patients and families interviewed to downplay NF1.

In the light of the findings of this chapter, it is possible to hypothesise a mirroring of the healthcare service structure around NF1 and patients' attitudes towards their condition.

In fact, the tendency to minimize, disregard and not think about NF1 as much as possible demonstrated by patients and families interviewed, could also represent a mode of 'adjustment' to the lack of medical service offered around the syndrome.

Concerning the structure of the healthcare system, the existence of NF1 does not appear to be reflected in NF1 dedicated services. As it has already been observed, specialised NF1 clinics are very rare. Furthermore, the medical support for this syndrome is often partial and fragmented. If NF1 patients manage to receive any treatment and monitoring, this is normally uncoordinated and related to the occurrence of specific symptoms. Patients for example may see orthopaedics if they have osseous lesions, surgeons if they need neurofibromas removed, or a neurologist if they develop neurological problems. This hinders the possibility of early detection of symptoms and complications as well as the availability of treatments which take into account the complex nature of the whole syndrome, rather than the specific symptom.

This fragmented, symptom-related medical approach seems to be reflected in the fact that patients and families, whilst showing the tendency to downplay their condition as a whole, tend to give importance to NF1-related specific symptoms. In chapter 6, it has been observed that the illness identities of individuals with the condition are frequently fragmented according to their specific symptoms (e.g. malignancy, cognitive difficulties, and cosmetic problems). It could be argued that patients assign importance to symptoms also because the healthcare system is able to address them and, ultimately, to hospitalise or 'institutionalise' them, unless very severe (e.g. oncology, neurology,

surgery units and their treatment and management pathways). So the patient may be more inclined to see themselves as having cancer, or deafness, but not NF1.

In terms of clinical practice, this chapter has also shown that GPs and other physicians are often perceived by patients as not being able to provide information and support around NF1, failing to validate their symptoms and concerns. This 'style' of medical practice, can diminish the 'meaningfulness' of the syndrome. The fact that healthcare professionals do not often acknowledge through their everyday discourses and practices the reality of NF1 could represent one of the influences that lead patients and families to disregard and minimise their disorders. Patients may feel abandoned by healthcare providers who undermine in different ways their symptoms and concerns and they may adjust to this, by dismissing their syndrome in a similar manner to the doctors they deal with directly or indirectly. On the other hand, this relation between patients' attitudes towards NF1 and healthcare professionals' way of dealing with it could be seen as being bidirectional. Thus, it could be argued that healthcare professionals' tendency to minimize the impact of NF1 may be influenced by patients downplaying attitudes.

Furthermore, through the interpretative lens of the ideas of genetic responsibility and citizenship (Petersen and Lupton 1997; Hallowell 1999; Petersen and Bunton 2002; Rose and Novas 2004), patients' downplaying strategies can be interpreted as a form of response to the social expectation to be informed about and engage with risk. The fact that the healthcare system lacks NF1-centred services, may lead patients to minimize their disorder. As patients do not have access to specific NF1 related structures that allow them to perform their duty of risk monitoring and minimization, they ignore their syndrome. When, instead, there is a healthcare service offer available i.e. for specific

NF1-related symptoms, patients tend to use the medical system and to give greater importance to their symptoms (rather than their syndrome as a whole).

Patients' tendency to downplay NF1 could also be influenced by important aspects that characterise the practice of genetic service providers in particular. Part of the work of the clinic consists in attempting to remove from patients any sense of responsibility or guilt for having, or having transmitted, a genetic condition. Besides, genetic service providers work as well to de-stigmatise disorders when there are other psychological and social implications attached (Bosk 1992; Featherstone, Gregory et al. 2007; Sanders, Campbell et al. 2007).

Moreover, as corroborated by the genetic counsellor, clinical geneticists- in order to help shocked patients and families adjust to a diagnosis, stress the universality and randomness of having "*faulty genes*". By deploying the argument that we all have faults in our genes they help patients to realise that "*nobody's perfect*" and put their disorder in perspective.

The multidirectional lack of recognition of the syndrome reported by physicians, coupled with these specific discourses and practices of genetic counselors may be mirrored in the NF1 illness experience and create a reflexive arch of understanding that NF1 is "*no big deal*". These aspects of medical and clinical practice are juxtaposed with patients' tendency, described in Chapter 6, to put their syndrome in perspective, considering it (as many participants reported) "*just an illness*", "*nothing to worry about*", "*just one of those things*" and extending at the same time the state of 'defectiveness' and 'imperfection' to society as a whole.

These links traced between the structure of the healthcare system and patients' experiences with their illness represent an additional justification for the lack of biosociality or biocitizenship around NF1. It could be argued that if NF1 had a more visible medical status, that is, if there was more recognition and support provided by the healthcare system for this disorder, patients' experience with the condition (e.g. the lack of biosociality) may be different. NF1, as such, and not for example specific pressing symptoms, or singular aspects related to the disorder, may become more visible and important to patients. This may contribute in strengthening patients' sense of NF1 illness identity and consequently lead them to engage more with support groups around the syndrome.

The ideas of patient autonomy and empowerment which pervade public health discourses and which equally inspire theoretical constructs like biocitizenship (Rose and Novas 2004) and biosociality (Rabinow 1996) presuppose a certain type of citizen. This paradigm favours articulate, educated middle-class individuals who feel equal to healthcare providers in terms of power and knowledge. On the other hand, this paradigm can be disenfranchising to less powerful groups who may be less educated and may also have cognitive and physical difficulties which can prevent them from understanding information on their health, coordinate their care and initiate treatment. A lot of individuals with NF1 may belong to this second group as the syndrome can cause cognitive difficulties and because of the lack of disease identity.



## 9. Conclusions

This thesis aimed to explore the role of genetic knowledge in patients and families experiences with NF1 and in the healthcare management of this condition.

Theoretically, the study was contextualised within work on genetic medicine and subjecthood (Petersen and Bunton 2002), family dynamics (Cox and McKellin 1999; Finkler 2000; Featherstone, Atkinson et al. 2006; Hamilton, Williams et al. 2009), genetic responsibility (Hallowell 1999; Novas and Rose 2000), biocitizenship (Rose and Novas 2004) and biosociality (Rabinow 1996). This thesis focused on NF1: a complex, variable and unpredictable condition - a *condition without parameters* in Ablon's vivid words (Ablon 1999).

In Chapter 3, I discussed the idea advocated by several social theorists that genetic practices are entering into the medical domain and transforming individual subjectivity. Individuals have been described as increasingly incorporating genetic information into their everyday lives and understanding their personhood in genetic terms. This phenomenon has been interpreted as being both empowering and/or as a form of subjectification (Lippman 1998; Kerr and Shakespeare 2002; Petersen and Bunton 2002; Rose 2006). I hypothesised that genetic subjecthood may be less evident in the case of multisystem, highly variable and unpredictable genetic disorders like NF1 which may be managed, alongside clinical genetics, by other medical specialties (Cox and Starzomski 2004; Weiner 2006; Weiner 2010). The uncertainty and variability of NF1, that is, the lack of a model, or constant prognostic *parameters*, could prevent patients and families from understanding their syndrome and thereby endorsing a clear NF1 disease identity. The neurologic and cognitive symptomatology could represent another important mitigating factor, as it may hinder patients' capability to understand the complexity of their syndrome.

In Chapters 2 and 3 I highlighted that within the existing psychosocial qualitative research on NF1 there is a lack of studies which directly explore how NF1 patients, their family networks and healthcare providers make sense of genetic knowledge and uncertainty and manage this complex syndrome. Thus, alongside the sociology of genetic medicine, this study has also contributed to the psychosocial qualitative research on NF1.

In summary, this dissertation project has contributed to the academic literature on individual and familial construction of genetic disorders, lay understandings of risk, genetics and heritability and to the psychosocial qualitative research on NF1. It may inform clinical services for NF1 patients and their families and add to the policy debate over the treatment of this and other similar genetic conditions, within the NHS. This final chapter will summarise the main findings of the thesis and discuss its theoretical implications. It will, then, consider the significance of the findings in relation to policy and clinical practice.

## 9.1 Summary of Main Findings and Interpretations

This research investigated:

- (1) The relevance of the notions of genetic responsibility, medicalization of the family, biocitizenship and biosociality to NF1, that is, the ways in which knowledge about the genetic basis of NF1 influences individual and familial experiences with the syndrome, the definition of the self, the management of the condition and the use of structural support such as support groups.
- (2) The interrelation of individual and familial experiences of disease, particularly individual and familial meaning making practices around uncertainty.
- (3) How patients and families are managed by the healthcare system, how they experience it and how genetic classifications play (or do not play) into this.

### Subjecthood and Identity

In Chapter 6, I discussed the results principally related to the first research aim concerning investigating the relationship between genetic medicine and subjecthood in the context of NF1. Data analysis suggested that NF1 does not appear to be associated with a strong disease identity or community. The identities of individuals with the condition were fragmented according to their specific symptoms (e.g. malignancy, cognitive difficulties, cosmetic problems etc.). Mildly affected individuals often refused to identify themselves with the more seriously affected ones and vice versa. The majority of the respondents - irrespective of the severity of the condition - tended to reject NF1 identities by employing diverse downplaying strategies such as normalising the disorder, avoiding thinking about it or focusing on the positive or more urgent aspects of daily lives. The downplaying attitudes and fragmented disease identities can be also interpreted as a modality of dealing with NF1's uncertainty and lack of *parameters* (Ablon 1999). At the level of patients' experiences, the variability and

uncertainty of the syndrome was not made more coherent by the fact that it has a genetic basis.

NF1 was generally minimised and downplayed, but it could become salient at certain junctures in the lifecourse of individuals, especially in relation to reproductive choices. In fact, the majority of respondents - once again irrespective of the severity of the condition - engaged with aspects of 'genetic responsibility' (Hallowell 1999; Novas and Rose 2000; Petersen 2006) in relation to NF1 in terms of making certain reproductive choices, levels of disclosure and management of genetic risk and information. Making reproductive decisions was also a strategy to exert some control over the uncertainty of the syndrome (Petersen 2006; Kelly 2009; Raspberry and Skinner 2011).

The salience of the genetic aspect of the syndrome was not a stable phenomenon in individuals' lives. The discourses deployed by the participants of my study were more in line with other empirical research on individuals' constructions of genetic disorders (reviewed in Chapter 3) which highlighted the heterogeneity and situatedness of disease experience (see for example (Parsons and Atkinson 1992; Cox and McKellin 1999; Hallowell, Arden-Jones et al. 2005; Petersen 2006; Sanders, Campbell et al. 2007; Klitzman 2009)).

The majority of the respondents did not engage with NF1 support groups. Similar to what was described by Ablon in the USA context, the variability of NF1 was an important factor (Ablon 1996; Ablon 1999). However, I also found that participants' reluctance to join NF1 support groups was justified by contextual-historical characteristics of the UK NF1 association (for example, perceptions of the lack of structural support or the negative way in which the association portrayed NF1). Furthermore, some respondents to my study sought support from organizations related to specific symptoms (e.g. malignancy, blindness, deafness etc.)

These results mitigate Rose and Rabinow's idea of biocitizenship and biosociality, the idea that individuals are increasingly seeking social support based on shared biological identities and activism (Rabinow 1996; Rose and Novas 2004). These notions may be less evident or not applicable to NF1 and other genetic conditions which are very variable, uncertain and with unclear prognostic parameters.

I finally observed that the findings about the lack of biosociality and biocitizenship for NF1 are not only theoretically salient, but can also have important clinical implications. As the NHS does not always possess the structural capacity to manage certain complex or rare disorders, healthcare providers may refer patients to specialized disease specific support groups, assuming that they will join and find the help and information they need. By knowing that patients with NF1 (or other multisystem variable disorders) may not join disease-specific support groups as they are not consonant with their own self-identities in relation to NF1, healthcare providers can seek for alternative solutions to provide support to patients.

### **The Impact of Family Dynamics on Individual Experiences**

In Chapter 5, I delved into one of the key interest of this thesis i.e., the link between familial dynamics and individual responses to genetic knowledge and diseases (the first two research aims). In this chapter I concentrated on the interviews of one particular family from the sample: Grace and Benjamin's family. By analysing the entanglement of familial meanings in the fabric of individual illness narratives and health management behaviours, I showed different modalities in which this syndrome was co-understood and co-negotiated by patients and their family members. The chapter illustrated how family dynamics and models can strongly influence individuals' experiences with this disorder (Bylund, Galvin et al. 2010).

Grace's arc of illness narrative was characterised by crucial phases strongly entangled with familial elements e.g. the diagnosis performed by her mother Rosemary when she was a young teenager, her father Benjamin's experience with NF1, and her pregnancy which finally made her accept and deal with her diagnosis through the 'responsibility' of having an affected child (i.e. acceptance by proxy).

Benjamin was an orienting figure against whom all other instances of NF1 (in the family) were compared or even negated: other affected family members were always in the background and almost considered as not having the condition.

In this chapter, I observed that the family can be seen as a primary source of help, but family relations may also affect disclosure and the way families cope with a genetic condition. This can depend on the fact that family members are linked by complex biological and affective bonds which may hinder their ability to hear and deal with familial illness narratives (as they are part of their own personal stories too).

Communicating and living with Benjamin became incredibly arduous for his family members during the period in which he was "*very angry*" about his NF1 and quadriplegia. During this 'chaotic' period family relations and bonding were seriously compromised; the family managed to surpass this critical phase (which lasted for about five years) through external help, particularly that of a specialist home carer.

The chapter ultimately highlighted the theoretical and clinical importance of contextualizing individual constructions of genetic disorders within the familial environment and of increasing our awareness on the complexity of family relations (Featherstone, Atkinson et al. 2006; Williams, Skirton et al. 2009). In the light of these results, I also argued that it is important to offer the possibility to patients and families of expressing, if they have a need and wish to, their anxiety and suffering to an external advisor. The narrative analysis approach allowed to uncover this dimension.

In Chapter 7 I presented results principally related to the second research aim on the interrelation of individual and familial dynamics around the genetic aspects of NF1. The analysis suggested that although NF1 displays high variability and unpredictability, both within and between individuals, surveillance of other family members with the condition was an important tool used by respondents in managing their concerns over the possible developments of the condition (Hallowell, Jacobs et al. 2001; Kenen, Ardern-Jones et al. 2003). As already found in Chapter 5, the family provided *parameters* to the uncertainty of NF1; the absence of a clear prognostic model tended to be replaced, when possible (when there was a family history of the condition), by family models. In spite of there being an identified genetic basis, it was the family context, and not the genetic, that provided parameters. The results in Chapter 5 and 7 triangulate to strengthen the robustness of this particular finding and align to support my interpretation.

The space and management of uncertainty within families was not univocal, i.e. it could be experienced differently by different members. Similarly, the space of communication within families was multifarious; it could conflate positive and negative instances and could also be experienced differently by different family members (Clarke, Richards et al. 2005; d'Agincourt-Canning 2005).

Overall, these findings support the idea of practical kinship (Featherstone, Atkinson et al. 2006). Family dynamics are complex and can strongly influence the way individuals relate to genetic information and manage their diseases. I also showed that the experience and management of genetic diagnoses and their uncertainty is strongly influenced by family dynamics and bonds.

This chapter corroborated one of the key tenets of this thesis already discussed in Chapter 5: the value of exploring families. Analysis of the interviews of different family

members offers a rich point of access into familial patterns of communication and relation, providing, at the same time, useful information to contextualise and interpret respondents' accounts. The family approach, for example, allowed to give more meaning to participants' downplaying discourses. Individual discourses about the lack of discussion or minimization of NF1, rather than simply being a sign of a family/individual well-adjusted to the disorder, could also hide familial tensions and concerns.

### **The Role of the Healthcare System**

In Chapter 8, I examined the results principally related to the third research aim on the role played by the healthcare system in familial and individual experiences with NF1. The chapter was based on interviews with healthcare providers (e.g. medical staff, genetic consultants, and NF1 specialist advisors). The study revealed problems of coordination and management of healthcare. The most common problem raised revolved around the difficulty in offering a coordinated and effective service for patients with NF1 and their families. Healthcare providers observed - similarly to what they had noticed with other lifelong complex multisystem conditions (e.g. Marfan syndrome, cerebral palsy, spina bifida etc.) - that the symptoms of NF1 tend to be addressed separately by different medical specialties with no one specialty taking a comprehensive view of the disorder and coordinating the service. Following the line of reasoning of the healthcare providers interviewed, I argued that this problem may be common with syndromes where the specific symptoms rather than the whole disorder are treated (Cox and Starzomski 2004; Weiner 2006).

I identified a trend for the patient's illness identity to mirror the structure and practices of the healthcare system. I interpreted the patients and families' tendency to minimize



and not think about NF1 as being a possible modality of ‘adjustment’ to the lack of medical service offered around the syndrome. Furthermore, I suggested that the prevailing medical approach characterised by the treatment of specific symptoms, without an overall understanding of the disorder, was reflected in the patients and families’ tendency to downplay their condition as a whole, and only to consider specific symptoms when they become pressing.

I also observed that in the light of the widespread public health discourses about active and responsible citizens-patients (Petersen and Lupton 1996; Rose 2001; Petersen and Bunton 2002; DoH 2010), the respondents’ tendency to downplay NF1 could also be interpreted as a modality of adjustment to the social expectation of being informed about and act to minimise genetic risk and diseases. Patients’ discourses of disengagement with the syndrome could serve as a strategy to protect their image of responsible patients. As the healthcare system often does not treat NF1 as a coherent disorder (but often specific related symptoms), patients downplay their disease; since patients cannot access/are not offered the healthcare structures that allow them to perform a duty of responsible patients, they deflate the cause, i.e. the disease itself.

Finally, I pointed out that these links traced between the structure of the healthcare provision and patients’ experiences with their illness could also represent a further justification for the lack of a common identity or biosociality around NF1. I argued that if NF1 had a more visible medical status, i.e. if there was more education amongst healthcare providers and services for this disorder- or if it were treated as a ‘whole’ rather than a disparate set of symptoms with uncertain meaning- patients’ individual and collective experience with the condition may be different.

## **9.2 Genetic Subjecthood, Uncertainty and Family Dynamics of a**

### ***Condition without Parameters***

The case of NF1 does not seem to fit neatly with the position held by many scholars that genetic knowledge is profoundly transforming individual subjectivity - either in positive or negative terms (Lippman 1992; Lippman 1998; Kerr and Shakespeare 2002; Rose and Novas 2004). The main results summarised in the previous section suggest that individual and familial responses to genetic medicine are heterogeneous and situated. This thesis is closer to research that acknowledges individual agency and the complexity of responses to genetic knowledge (Parsons and Atkinson 1992; Cox and McKellin 1999; Hallowell 1999; Cox and Starzomski 2004; Petersen 2006; Soltysiak, Gardiner et al. 2007).

The data derived from this study problematizes theoretical constructs like biosociality and biocitizenship (Rabinow 1996; Rose and Novas 2004) which advocate for the idea that genetic illness identities are increasingly utilised in the creation of communities of shared recognition, patients' advocacy, expertise, support and activism. In this study, the vast majority of the respondents, notwithstanding the multiple NF1-related difficulties experienced, preferred not to engage with NF1 associations, precluding themselves from the assistance offered by these groups. Moreover, they also tended to normalise and minimize their syndrome.

Being a complex genetic syndrome, NF1 is a collection of many different symptoms and features subsumed under one genetic-syndrome label. Due to the compound and uncertain nature of this condition - which is also reflected in a fragmented healthcare service - patients may not endorse an NF1 identity, but, if any, a more symptoms-related identity. Therefore, the genetic basis of the syndrome notwithstanding, patients and families may struggle to 'encompass' and 'construe' NF1 as such and engage with it.

The lack of a 'clear illness' is reflected in the lack of a 'clear illness identity'. Patients and families' tendency to downplay the condition may be also seen as a way of coping with this fragmentation and uncertainty; a way of dealing with the anxiety related to the possible progressions of the syndrome. The cognitive and neurological symptomatology represents another important obstacle that can prevent patients and family from understanding information about their disease and acting to monitor and minimise their risk. Arguably, the high spontaneous mutation rate of NF1 (50%) could also be destabilising in terms of family and individual genetic identity as it subverts the link between genetics and heritability.

Together, all these characteristics of the condition hinder considerably the performance of the responsible patient/citizen duty. Therefore, notions of biocitizenship and biosociality, which are linked to the idea of genetic identity and are tied with public health discourses about responsible patients-citizens, may be less evident for this complex and variable genetic condition.

Nevertheless, it has to be acknowledged that the genetic categorization of NF1 has produced some important results. As discussed in Chapter 2, it attracted more scientific and clinical research on the syndrome, which has led, for example, to identify three genotype-phenotype correlations. Some patients who are found with this typology of mutations may face less uncertainty (e.g. they will not develop neurofibromas) and may deploy a clearer understanding of their disorder. However, as I highlighted in Chapter 2, genetic testing is rarely employed and these mutations are rarely found. Furthermore, notwithstanding the genotype phenotype correlations, it can be argued that the genetic categorization of the syndrome has still not significantly 'solved' the uncertainty of the

syndrome.<sup>25</sup> This is probably also due to the significant number of symptoms which are associated with this syndrome.

Therefore, alongside recognising the positive aspects of the identification of the genetic basis of NF1, it is equally important to highlight that the genetic categorization has not significantly changed the way NF1 is experienced at the individual and familial level, and the way it is managed by the healthcare system (Cox and Starzomski 2004).

The syndrome - despite being genetic - is still experienced by patients and treated by the healthcare system, in Ablon's terms as a *condition without parameters*, that is, as a disparate set of symptoms with uncertain meaning rather than as a 'whole'. The example of NF1 shows that the identification of the genetic basis of a condition does not necessarily provide patients and healthcare professionals with more parameters to manage it; genetic information does not always deliver more certainties and does not necessarily increase or open the possibilities of intervention apt to minimise disease or risks. In the postgenomic era, it has become undeniable that understanding and manipulating the molecular mechanisms of disease is an enormous task which often goes beyond the identification of genetic links (Lock and Nguyen 2010). The results of my research apply to NF1, but can also be relevant for other conditions with no constant and clear parameters (e.g. spina bifida, cerebral palsy etc.).

Following Petersen and Bunton's critical stance towards ideas of a direct correlation between developments in genetic medicine and increases in patients' empowerment (Petersen and Bunton 2002), I am inclined to argue that the notions of biocitizenship and biosociality may lead to overlooking situations where patients do not or cannot engage with their genetic disorders and bring forward their health-related needs.

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<sup>25</sup> With reference to '*overall prognostic parameters*', guidelines converge in estimating that generally two thirds of people with NF1 are mildly affected and can live a healthy life (Ferner 2010).

Broadly speaking, the notions of biosociality and biocitizenship are more likely to suit (in practical and political terms) articulate, educated middle class individuals who feel equal to healthcare providers in terms of power and knowledge. This paradigm can be disenfranchising to less powerful groups who may be less educated and may also have cognitive and physical difficulties which can prevent them from understanding information on their health, coordinate their care and initiate treatment, particularly for fragmented disorders such as NF1 where multiple specialities need to be involved

Concerning the implications of genetic knowledge on family and kinship relations, contrary to the notion of medicalization of the family (Finkler 2000), the findings suggest that genetic medicine did not necessarily strengthen ties of biological relatedness. Although family dynamics were often intertwined with respondents' accounts of their experiences with genetic conditions, the relations of family or kinship, health behaviours, familial surveillance and disclosure did not necessarily follow the lines of biomedical knowledge and genetic inheritance. Genetic information was filtered by a pre-existing gamut of familial relations. Within these relations and their personal circumstances, respondents engaged in complex practices of acceptance, rejection and interpretation of biomedical knowledge. Therefore, the data derived from this study confirms research on family experience with other genetic conditions which emphasises the significance of contextualising 'new' genetic knowledge within existing frameworks (Cox and McKellin 1999; Featherstone, Atkinson et al. 2006; Hamilton, Williams et al. 2009).

Theoretical constructs like medicalization of the family, biocitizenship and biosociality have an ambiguous status; they are not simply descriptive, but they also seem to be charged with normativity. Family bonds may be medicalized as a result of the presence

of a genetic risk or disorders in a family. Moreover, from a certain perspective, it may look advisable (for scholars, healthcare providers, policy makers) to think that as a result of innovation in genetic medicine families are increasingly undergoing a process of reconfiguration (or even historical reversion to a 'traditional family' model (Finkler, Skrzynia et al. 2003; Finkler 2005)) whereby biological ties are strengthened, and to assume that individuals are consequently able to disclose genetic information to their kin.

However, in line with existing research, my study has shown that families are not necessarily unified by shared genetic ailments; disclosure and family relations are intricate and may resist this (ideal or advisable) process of medicalization. For a number of contextual reasons (for example, instances of practical kinship (Featherstone, Atkinson et al. 2006)), genetic information about risk or disease may not flow easily, and families with genetic risk or disorders may experience many challenges.

Similarly, I would argue that biocitizenship and biosociality have received considerable interest and attention in the last decade not just for their heuristic potential, but also because they evoke a convincing scenario where - as a result of technological innovations - 'lay' expertise is celebrated. These notions and the scenarios they evoke, reverberate with the public health policies of the UK and many other countries characterised by high hopes towards biomedical technological innovation and a set of expectations towards responsible citizens, underpinned by assumptions that patients and families will engage with genetic risk and seek preventive and management strategies. Therefore, the tenets that underlie these theoretical constructs may please many ears, reinforcing in turn the uptake and success of these theories.

However, as I explained above, the scenario depicted by biosociality and biocitizenship (notwithstanding Rabinow's explicit mention to NF1 as a possible embodiment of biosociality) does not seem to fit the experiences of individuals with NF1 and families. It could be argued that this was a consequence of my sample being too limited. Yet, it is important to point out that statistically in the UK there should be at least 20000 people with NF1, but - up to two years ago - the Neuro Foundation had a membership of less than 2000 (inclusive of members with NF2). This means that it was missing the vast majority of the NF1 population. Moreover, the problem of a lack of a common disease identity among NF1 patients is not only a result of my research, but was also found by Ablon in the US a few decades ago (see Chapters 2 and 3).

It is important to highlight that generalizations about profound positive or negative changes in subjectivities, family and society due to innovations in biomedicine may contribute to healthcare providers and policy makers being given potentially misleading accounts of the role played by genetic innovation in improving healthcare services. This may hinder a rigorous understanding of patients' views and the possibility of developing evidenced, effective policies and interventions apt to improve the quality of care, health and wellbeing.

### **9.3 Clinical and Policy Suggestions**

As I stated in the introduction of this chapter, another objective of my thesis was to collect data on individual and familial experiences with NF1 with the hope of providing healthcare professionals with valuable information about the needs of service users and to make recommendations for clinical practice within the NHS in relation to this and possibly other similar genetic conditions. I will discuss my contribution in this area into two main points.

First, there certainly is a benefit in treating the family as a unit. It allows for a more nuanced understanding of the psychosocial impact of medical genetics and thereby to gain a greater insight into the dynamics and processes by which families face genetic conditions, genetic uncertainty, and healthcare.

In line with other studies (Featherstone, Atkinson et al. 2006; Hamilton, Williams et al. 2009; Williams, Skirton et al. 2009) my research has highlighted that individuals and families can face many challenges (e.g. disclosure, communication, relations, reproductive decisions etc.) in living with genetic disorders or risk.

The improvement of genetic services' strategies to facilitate family communication about genetic disorders could have a positive impact on the social wellbeing and health outcomes of patients. There is a pressing need for more investigation on how families interpret and manage genetic risk and diagnoses. In fact, although families can be complex entities to explore, focusing on how at risk individuals understand and manage hereditary risk in the everyday family life context may enrich research in clinical settings.

Second, increasing the number of NF advisors could considerably improve the healthcare service for NF1 patients (Huson 1999). NF specialist advisors work regionally within hospitals and give support to anyone affected by NF1. They normally come from nursing or other medical backgrounds and are given further training by the Neuro Foundation in order to be able to provide support to NF1 patients and their families following a diagnosis or other stages. NF advisors usually work with the whole family from the outset. Advisors effectively connect patients with specialists and can represent a valuable contact point to help patients address a wide spectrum of problems (educational, physical, psychological). Having a contact point could also be valuable in view of the variability of NF1, and its lack of biosociality. In fact, the contact point



would be a resource that both seriously and mildly affected patients and families can refer to when needed, without too much 'disease identity commitment'. From a social point of view, receiving home visits and help from NF advisors may be less distressing than joining a NF support group and attend 'public' meetings with other affected individuals. Moreover, increasing the number of NF1 advisors may be easier and more sustainable than other solutions, like multidisciplinary clinics. Finally, NF specialist advisors are also very family-focused as they normally see the whole family in the community. This factor is particularly significant in relation to one of the key points of my thesis i.e., the importance of taking into account the context of family relations and bonds while studying patients' experiences with genetic disorders or managing medical genetics patients. The importance of the role of professionals such as family advisors (and health visitors) has been highlighted also in relation to other disorders (Lewis, Skirton et al. 2010)

Given the recession, the UK Governments policy on financial cuts, and the 'Big Society' climate, I believe that the financial resources to increase the number of advisors would need to come from the private voluntary sector, for example NF1 support groups. The Neuro Foundation is already moving towards this direction as it is currently funding a team of five NF1 and NF2 advisors that is supposed to cover the whole UK (see for example spring newsletter 2011

[http://www.nfauk.org/uploads/NF\\_News\\_Spring\\_2011.pdf](http://www.nfauk.org/uploads/NF_News_Spring_2011.pdf)).

Nevertheless, the number of advisors needs to increase in order to reach more families with NF1. A possible solution would be to augment the visibility and income of the Neuro Foundation, in order to attract more members and resources. However, this thesis has highlighted that NF1 patients and their families may prefer not to join an NF1 support group. The variability of the condition was found to prevent the formation of a univocal NF1 identity. This was related to tendency to join, if any, support groups

related to specific pressing symptoms. To address this problem, the Neuro Foundation could probably try to establish more partnerships with other support groups visited by NF1 patients (for example cancer, cosmetic disfigurement, blindness groups).

## **9.4 Self-Reflexivity**

### **Language**

The fact I am not a native English speaker represented at the same time a problem and an interesting vantage point. First, I occasionally experienced some difficulties - both during the interview and in transcribing them – in understanding some idioms and words, but this never jeopardised the flow of the interviews.

However, the awareness of the possibility of a linguistic barrier strengthened my concentration and listening skills during the interviews and made the transcribing process slower, but more rigorous. It could be argued that this extra effort put into listening to participants' accounts has probably contributed in making them feel valued and therefore more comfortable and willing to share their experiences with me.

Moreover, my being foreign (i.e. my 'otherness') may have also helped in making participants more willing to share intimate and delicate aspects of their experiences.

Some may have found it easier to talk to a researcher who was a 'stranger' in many respects i.e. not only somebody who was not linked to their networks, but also somebody who comes from a different cultural and linguistic background.

### **Fieldwork (adapted from one of my memos 22/11/2008)**

In the first interview, I was probably biased and unready to respond properly to the interviewee (Seth). I was seeking for a long narrative of an experience with NF1, whereas in this case the narrative was of a very sudden fatal event, and the life before

that was presented as a normal (un-eventful as concerns NF1) life (just with some “minor inconveniences”: dyslexia, neurofibromas, blood pressure). By asking many questions about the experiences with NF1, I was also a bit afraid to give the impression to the interviewee that I was pushing a ‘normal’ case to be ‘abnormal’, or ‘special’. With more experience I realised (after the third interview) that the lack of an eventful NF1 story could represent in itself very rich data.

On a personal level, these initial experiences made me understand how to better prepare before the interviews. It made me also modify, slightly, the interview schedule to reflect my research aims more faithfully (see also Chapter 4).

The interview schedule after the first three interviews was changed with the intent of eliciting, first of all, more narratives. Some questions have been reformulated in order to ask the interviewee to provide salient episodes or illustrative events (i.e. they included more probes).

Secondly, I noted that the family members’ interview were not enough centred on them, but rather on the relatives with NF1; thus some questions have been reformulated to acquire more personal experience from the interviewee (the interview schedule in the Appendix 3 is the final version).

### **Theory and Analysis**

My training prior to the start of the PhD was mainly in Philosophy. Therefore, Sociology and the subfield of Sociology of Health and Illness (and genetic medicine) were almost novel disciplines to me. Again this aspect probably represented both a weakness and strength. On one hand, I started from a disadvantageous position (with respect to somebody with training in Sociology). Whilst undertaking my PhD, I had to get acquainted with a fascinating but novel subject, acquire a different sensitivity, language, style of thinking and process a considerable amount of new information. My

lack of experience and expertise in this discipline (and in conducting fieldwork) may have lessened the depth of my work. However, I believe that my situation could have some points of contact to the GT ideal of the researcher being a tabula rasa without preconceived knowledge developed from experience and literatures already read (Strauss and Corbin 1990). Although, as I argued in Chapter 4, I find this idea of tabula rasa to be problematic, I can claim that my lack of previous training in Sociology has also given me a fresh perspective on the theory and the data collected. In particular, it has allowed me to dwell on participants' experiences and words for a longer time without being too hasty in categorizing their discourses under theoretical notions.

## **9.5 Limitations of this Thesis and Future Research**

This research project has focused on one specific syndrome (NF1) and geographical location (UK). Moreover, although the patients and families interviewed came from different education and working backgrounds, they were all white and British. It would have been more advantageous to collect illness experiences and understanding of genetic knowledge from a more culturally varied sample. As different cultures can hold, among other things, different identity and familial values, a more varied sample could have enriched my exploration of individual and familial dynamics around genetic knowledge. This could be an object of future research.

Moreover, I have not covered certain NF1 symptoms like blindness (although I interviewed individuals with visual impairment), deafness and cognitive impairment (although I interviewed individuals with mild learning difficulties like dyslexia).

Individuals affected by these symptoms may have different needs and bring new perspectives and experiences on the psychosocial impact of the syndrome (Shakespeare 1994). Likewise, I have also not interviewed many adolescents, although this category of people could be particularly affected by the cosmetic and other potentially social

distressing features of NF1 (Counterman, Saylor et al. 1995; Sebold, Lovell et al. 2004).

I have highlighted throughout this thesis that only a few studies of experiences with genetic disorders have looked at multiple family members, and I have emphasised the importance of the family network approach I adopted. However, in only 4/13 families I managed to interview three or more family members. This was mainly due to the time constraint of the PhD, but probably also to more general difficulties in accessing family networks.

As I observed in Chapter 4, families can be fragmented complex entities and therefore difficult to explore. In particular, recruiting many individuals within the same family network can also be problematic. For example, participants from the same family network may feel the condition of confidentiality to be undermined.

A longitudinal study based on following families throughout the course of their illness would also better illuminate how experiences and meanings related to this uncertain condition can change over time. My research has highlighted how individual and familial perceptions of risk varied throughout time the lifecourse. Interviewing families over a period of a few years would probably allow researchers to get a richer insight of these experiences and better test theories and hypothesis. For example it could to test whether introducing more family advisors and/or a more social community based model of care would actually help families.

For this PhD, due to time constraints, I interviewed healthcare professionals working mainly in the south of England.

Sampling more healthcare professionals from a wider amount of clinics in the UK would allow for richer data about the healthcare management of NF1 and for a more nuanced understanding of the differences in the healthcare provision for this and similar syndromes across the country. In particular, I would be interested in investigating whether and how NF1 patients and families' experiences with NF1 are different in Manchester and London where the two national NF1 specialist clinics are based.

Despite these limitations, it is also important to highlight that the collection of data from families and healthcare professionals has enabled some degree of triangulation of results (see Chapter 8).

It is also important that more empirical detailed studies are conducted with a greater variety of conditions in order to delineate a nuanced and encompassing picture about the social impact of genetic medicine. In particular, it would be interesting to conduct research on other disorders where symptoms rather than the whole condition are treated and see if there are similarities with NF1. It may be worth exploring whether the genetic basis of these syndromes has made their variability and uncertainty more coherent.

Similarly to the research aim of my PhD I would focus on patients and families experiences - in particular the interrelation of individual and familial sense-making practices around uncertainty - and the management of the syndromes in the healthcare system.

Finally, NF1 could be a very interesting case study in the context of the rapid development of quicker, more accurate and less invasive forms of prenatal testing techniques. Because of its high unpredictability and variability, which encompasses both physical and cognitive symptomatology, NF1 would raise complex questions in relation to possible decision processes among prospective parents, for example, what

parents value as an healthy child, whether there are any differences between cognitive and physical symptoms and how parents deal with the potential of uncertainty.

# Appendices

## Appendix 1



SCHOOL OF HUMANITIES  
AND SOCIAL SCIENCES  
incorporating the Institute of Arab and Islamic Studies

SCHOOL OFFICE  
Amory Building  
Rennes Drive  
Exeter  
UK EX4 4RJ

Telephone +44 (0)1392 263301  
Fax +44 (0)1392 263305  
Email [huss-schooloffice@exeter.ac.uk](mailto:huss-schooloffice@exeter.ac.uk)  
Web [www.exeter.ac.uk/huss](http://www.exeter.ac.uk/huss)

### CERTIFICATE OF ETHICAL APPROVAL

School/Academic Unit:

Egenis, School of Humanities and Social Sciences, University of Exeter

Title of Project:

The psycho-social construction of neurofibromatosis

Name(s)/Title of Project Research Team Member(s):

Daniele Carreri

Project Contact Point (incl. telephone no.):

Daniele Carreri

Tel: 01392 269142

Email: [dc233@exeter.ac.uk](mailto:dc233@exeter.ac.uk)

Brief Description of Project:

The aim of the project is to study the psycho-social and structural understandings and construction of neurofibromatosis (NF). It will investigate how the condition is understood by family and patient organisations, counsellors, parents and individuals with NF.

This project has been approved for the period

From: November 2007

To: September 2009

School Ethics Committee approval reference: 12.11.07/v

Signature.....*Mary Carter*..... Date.....*22-11-07*.....  
(Mary Carter – Chair HUSS School Ethics Committee)



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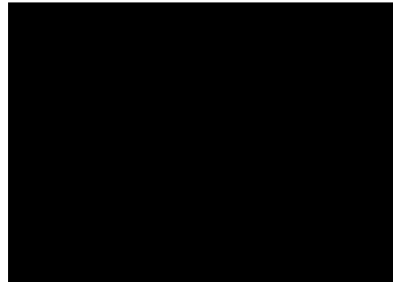


## National Research Ethics Service

Our ref LCH

07 November 2008

Mr Daniele Carrieri  
1st year PhD Student, Egenis  
Byrne House  
St Germans Road  
Exeter  
EX4 4PJ



Dear Mr Carrieri

**Full title of study:** Perceptions and Experiences with Neurofibromatosis Type 1

**REC reference number:** [Redacted]

Thank you for your letter of 05 August 2008, responding to the Committee's request for further information on the above research and submitting revised documentation.

The further information has been considered on behalf of the Committee by the Chair.

### Confirmation of ethical opinion

On behalf of the Committee, I am pleased to confirm a favourable ethical opinion for the above research on the basis described in the application form, protocol and supporting documentation as revised, subject to the conditions specified below.

### Ethical review of research sites

The Committee has designated this study as exempt from site-specific assessment (SSA). The favourable opinion for the study applies to all sites involved in the research. There is no requirement for other Local Research Ethics Committees to be informed or SSA to be carried out at each site.

### Conditions of the favourable opinion

The favourable opinion is subject to the following conditions being met prior to the start of the study.

Management permission or approval must be obtained from each host organisation prior to the start of the study at the site concerned.

Management permission at NHS sites ("R&D approval") should be obtained from the relevant care organisation(s) in accordance with NHS research governance arrangements. Guidance on applying for NHS permission is available in the Integrated Research Application System or at <http://www.rdforum.nhs.uk>.

### Approved documents

The final list of documents reviewed and approved by the Committee is as follows:

This Research Ethics Committee is an advisory committee to South West Strategic Health Authority  
*The National Research Ethics Service (NRES) represents the NRES Directorate within the National Patient Safety Agency and Research Ethics Committees in England*

Document	Version	Date
Interview Non NHS Invitation letter including reply slip - Family/Patients	2	28 October 2008
Interview Follow up NHS Invitation letter including reply slip - Family/Patients	2	28 October 2008
Interview NHS Invitation letter including reply slip - Family/Patients	2	28 October 2008
Interview Follow up letter including reply slip - Doctors	2	28 October 2008
Interview Invitation letter including reply slip- Doctors	2	28 October 2008
Response to Request for Further Information		05 August 2008
Participant Information Sheet: Interview	2	28 October 2008
Letter of invitation to participant	2	28 October 2008
Interview Schedules/Topic Guides	2	28 October 2008
Follow up NHS Q Methodology including reply slip	2	28 October 2008
Invitation letter including reply slip - NHS Q Methodology	2	28 October 2008
Information Sheet - Q Methodology	2	28 October 2008
Interview follow up Non NHS invitation letter including reply slip - Family/Patients	2	28 October 2008
Follow up Non NHS Q Methodology Invitation Letter including reply slip	2	28 October 2008
Non NHS Q Methodology Invitation Letter including reply slip	2	28 October 2008
CV	Dr Hannah Farrimond	
Flowchart	1	16 June 2008
Participant Consent Form: Interview	1	16 June 2008
Participant Consent Form: Q Methodology	1	16 June 2008
Letter from Sponsor		27 April 2008
Covering Letter		16 June 2008
Protocol	1	16 June 2008
Investigator CV		
Application	1	
Letter of Invitation - Q Methodology	1	16 June 2008
University Certificate of Ethical Approval		22 November 2007
CV	Dr Susan Kelly	16 June 2008
Questionnaire - Q Sort	1	16 June 2008
Letter Of Invitation - Non-NHS Q Methodology	1	16 June 2008

#### Statement of compliance

The Committee is constituted in accordance with the Governance Arrangements for Research Ethics Committees (July 2001) and complies fully with the Standard Operating Procedures for Research Ethics Committees in the UK.

#### After ethical review

Now that you have completed the application process please visit the National Research Ethics Website > After Review

You are invited to give your view of the service that you have received from the National Research Ethics Service and the application procedure. If you wish to make your views known please use the feedback form available on the website.

The attached document "After ethical review – guidance for researchers" gives detailed guidance on reporting requirements for studies with a favourable opinion, including:

- Notifying substantial amendments
- Progress and safety reports
- Notifying the end of the study

The NRES website also provides guidance on these topics, which is updated in the light of changes in reporting requirements or procedures.

We would also like to inform you that we consult regularly with stakeholders to improve our service. If you would like to join our Reference Group please email [referencegroup@nres.npsa.nhs.uk](mailto:referencegroup@nres.npsa.nhs.uk).

**Please quote this number on all correspondence**

With the Committee's best wishes for the success of this project

Yours sincerely

Enclosures: "After ethical review – guidance for researchers" SL- AR2

Copy to:

## Appendix 2

### Patients' Invitation Letter



Dear

My name is Daniele Carrieri, and I am a PhD student at the University of Exeter currently working with Dr [REDACTED] Consultant Clinical Geneticist at the [REDACTED] Hospital and Hon Senior Clinical Lecturer, [REDACTED] with whom you may be familiar.

Your name was suggested to me by your clinician, Dr [REDACTED]. The purpose of my writing is to ask you whether you would be kind enough to participate in some research about Neurofibromatosis Type 1 (NF1). The main purpose is to collect relevant information on the experiences of those directly or indirectly involved with the syndrome. It is hoped that the results will inform patient care for the disorder in the future.

If you decide to take part, we may also ask you if you know someone else who would like to participate.

Your participation is entirely voluntary. It will involve an informal face to face interview with me which will take about an hour of your time. It can take place where you are most comfortable, eg at home, in the University of Exeter or [REDACTED] Hospital. Travelling expenses will be refunded by the University.

Further information is included within the information sheet. If you would like to take part, please fill in the reply slip on the next sheet, and return it using the stamped addressed envelope.

I very much hope you will consider participating, as your contribution will be very valuable.

Thank you very much for your time, and I hope to hear from you soon.

Kind Regards,

Daniele Carrieri  
ESRC Centre for Genomics in Society  
Byrne House,  
St German's Road,  
Exeter,  
Devon, EX4 4PJ  
+44 (0) 7874051649  
[dc233@exeter.ac.uk](mailto:dc233@exeter.ac.uk)

Perceptions of and Experiences with NF1  
Reply Slip

Name \_\_\_\_\_

I am willing to participate in the study

I would prefer not to participate in the study

Address \_\_\_\_\_

Telephone-contact number \_\_\_\_\_

Mobile \_\_\_\_\_

Preferred time to be called \_\_\_\_\_

Email \_\_\_\_\_

Date \_\_\_\_\_

Signature \_\_\_\_\_

Please fill and return this slip in the free stamped addressed envelope. Thank you for your time! If you have any queries, call Daniele Carrieri on +44 (0) 7874051649 or email [dc233@exeter.ac.uk](mailto:dc233@exeter.ac.uk)

## Healthcare Providers Invitation Letter



Dear

My name is Daniele Carrieri, and I am a PhD student at the University of Exeter currently working with Dr [REDACTED] Consultant Clinical Geneticist at the [REDACTED] [REDACTED] Hospital and Hon Senior Clinical Lecturer, [REDACTED].

The aim of my writing is to inform you about the research project I am currently undertaking for my PhD, on Neurofibromatosis Type 1. The main purpose of my thesis is to collect and analyze the experiences of those directly or indirectly involved with the syndrome. It is hoped that the results will inform clinical services for NF1 in the future.

You have received this letter, upon recommendation from Dr [REDACTED] as you are involved with NF1 and able to provide a significant contribution to this project.

Participation is entirely voluntary. It will involve a face to face semi-structured interview, which will take 30 minutes to an hour of your time. The interview can take place where and when you are most comfortable, in your place of work, the University of Exeter, or another location of your preference.

Further information is included within the information sheet. If you would like to participate, please fill out the reply slip on the next sheet and return it in the stamped envelope provided.

I very much hope you will consider participating, as your contribution will be very valuable. Thank you very much for your time, and I hope to hear from you soon.

Kind Regards,

Daniele Carrieri  
ESRC Centre for Genomics in Society  
Byrne House,  
St German's Road,  
Exeter,  
Devon, EX4 4PJ  
+44 (0) 7874051649  
[dc233@exeter.ac.uk](mailto:dc233@exeter.ac.uk)

Perceptions of and Experiences with NF1  
Reply Slip

Name \_\_\_\_\_

I am willing to participate in the study

I would prefer not to participate in the study

Address \_\_\_\_\_

Telephone-contact number \_\_\_\_\_

Mobile \_\_\_\_\_

Preferred time to be called \_\_\_\_\_

Email \_\_\_\_\_

Date \_\_\_\_\_

Signature \_\_\_\_\_

Please fill and return this slip in the free stamped addressed envelope. Thank you for your time! If you have any queries, call Daniele Carrieri on +44 (0) 7874051649 or email [dc233@exeter.ac.uk](mailto:dc233@exeter.ac.uk)

## **Participant Interview Information Sheet**

### **Perceptions and Experiences with Neurofibromatosis Type 1**

Thank you for showing interest in this project. You are invited to take part in a face-to-face interview for the above research study. Please read this information sheet carefully before deciding whether or not to participate. If there is anything you do not understand or if you would like any more information, please call the research team or call the numbers given at the bottom of the last page. If you decide to participate, I thank you. If you decide not to take part there will be no disadvantage to you of any kind and I thank you for considering our request.

#### **What is the purpose of the project?**

The project aims to study the perceptions and understandings of Neurofibromatosis Type 1 (NF1).

#### **What is the purpose of the face-to-face interviews?**

The purpose of the interviews is to obtain information about the views and concerns on NF1 from individuals who have experience/contact with this disorder.

#### **Why have I been chosen?**

You are being invited to participate in the above study because you are affected by NF1 or because you have had a significant contact with individuals with NF1. If you know anyone else who you think might like to take part (e.g. other family member/friend/person with NF1), please let me know.

#### **What will happen to me if I take part in the interviews?**

If you agree to participate, we would be grateful if you could:

1. Complete the reply slip at the bottom of your letter and return it in the stamped addressed envelope.
2. You will be contacted by the research team who will arrange the interview with you either via telephone or face-to-face at a time and place that is convenient for you (this could be at your home, if you prefer).
3. The research team will meet with you. The interview will last approximately 40 – 60 minutes.
4. The interviews will be audio-taped. All data and results arising from the project will be anonymised and you can be in no way identified.
5. You may withdraw from the study at any time, without giving a reason. Your withdrawal will not in any way affect the care you receive.



### **What are the possible benefits of taking part?**

You will be giving the medical staff a greater understanding of everyday life of those affected by NF1, placing them in a better and more knowledgeable position to deal with NF1 and NF1 individuals. This research aims to add to our understanding of the experiences of those affected by NF1. It is hoped the result will inform care of those with NF1 or their families.

### **What are the possible risks of taking part?**

Some people, especially parents of NF1 individuals, may find the discussion of this genetic condition distressing. However it might be also de-stressing to be able to share experiences about the disease. You may withdraw from the study at any time, without giving a reason. If you have any concerns or would like to talk things through you can contact Dr [REDACTED] the hospital consultant involved in this study, through your GP or we can contact him on your behalf.

### **Confidentiality**

All data, including audiotapes, from the interviews will be anonymised and you can be in no way identified. All the information we collect will be treated as confidential and will only be given to the research team. We will also make sure that the information is stored securely. The audiotapes used will be destroyed five years after the study.

### **What will happen to the results of this study?**

Short quotes from your interview may be quoted in this research papers in a way that will not disclose your identity. The final results of this research will be circulated to doctors and researchers in the UK and elsewhere to help improve patient care.

### **Who is organizing the study?**

ESRC Centre for Genomics in Society (Egenis), University of Exeter.

### **Contacting us**

If you have questions or want more information on the study, please contact the Research Student Daniele Carrieri by phone (07874051649) or by email ([dc233@exeter.ac.uk](mailto:dc233@exeter.ac.uk) ). Thank you for your help with this project. For further information you can contact my supervisors Dr. Hannah Farrimond +44 (0) 1392 269128 [H.R.Farrimond@exeter.ac.uk](mailto:H.R.Farrimond@exeter.ac.uk) Dr. Susan Kelly +44 (0) 1392 269139 [S.E.Kelly@exeter.ac.uk](mailto:S.E.Kelly@exeter.ac.uk) .

### **Support numbers**

**The Neurofibromatosis Association** [www.nfauk.org](http://www.nfauk.org)

Quayside House  
38 High Street  
Kingston upon Thames  
KT1 1HL  
Surrey  
United Kingdom  
Telephone: 0208439123

## Appendix 3

### OUTLINE FOR INTERVIEW OF FAMILY NETWORKS OF NF1 INDIVIDUALS

#### ILLNESS EXPERIENCE

1. How did you come to realize your relative/s had NF1? Can you tell me how it happened?  
(Pathway of diagnostic experience/first diagnosis/first heard/ how many affected individuals in the family)
2. What is your experience with NF1? What about your family? Can you recall some episodes to illustrate this?
3. Has your experience with NF1 changed over time? What made it change? Why? Do you remember any particular event?

#### CONTACT WITH HEALTH PROFESSIONALS

4. Could you tell me something about the experience of your affected relative/s with the healthcare system in relation to NF1? Is there anything salient that you still remember?  
(healthcare system knowledgeable/supportive)
5. Have you been personally involved with health service in relation to NF1? If so could you please tell me for which reasons?  
(GP, hospital, genetic service)

#### SOCIAL ENVIRONMENT

6. In what ways do you think NF1 influences (has influenced) the life of your affected relative/s?  
(Work/ social life/school/family)
7. In what ways do you think NF1 influences (has influenced) your life?  
(Idem)
8. What do you find are/have been the most difficult aspects of your life in relation to NF1? Do you remember any particular episode?
9. What about the positive aspects? Do you remember any particular episode?
10. Where and how did you get the information on NF1?
11. Have you ever had contact with other individuals with NF1 and/or their families?

12. Some people with NF join NF support groups. What is your take on this?

#### CAUSES OF NF1/ TECHNOLOGIES

13. If you had to explain NF1 to someone, how would you explain it?  
(Definition)

14. In what way do you consider NF1 to be similar to or different from other health problems?

15. Why do you think people get NF1?

(16. Some people refer to NF1 as a genetic disorder, what do you think about that?)

17. What do you think the future will be like for treating NF1?  
(Technologies)

18. New technology can give the possibility to know in advance if a baby has NF1, what do you think about it?

#### MEDIA

19. Have you ever seen, read or heard on the media about a person affected by NF1?

(20. If so, how much did you find the case close to the one of your relative/s?)

(21. How do you see the future of your affected relative/s shaping up?)

22. Do you know any other person affected by NF1 who might be interested in participating to this study?

23. I have finished all my questions now, but is there anything else that you think we haven't talked about, something relevant to your life that you would like to say now?

## OUTLINE FOR INTERVIEW OF INDIVIDUALS WITH NF1

### ILLNESS EXPERIENCE

1. How did you come to realize you had NF1? Can you tell me how it happened?  
(Pathway of diagnostic experience/first diagnosis/first heard/ how many affected individuals in the family)
2. What is your experience with NF1? What about your family? Can you recall some episodes to illustrate this?
3. Has your experience with NF1 changed over time? What made it change? Why? Do you remember any particular event?

### CONTACT WITH HEALTH PROFESSIONALS

4. Could you tell me something about your experience with the healthcare system in relation to NF1? Is there anything in particular that you remember?  
(healthcare system knowledgeable/supportive)
5. Have your relatives been involved with health service in relation to NF1? If so could you please tell me something about that?  
(GP, hospital, genetic service)

### SOCIAL ENVIRONMENT

6. In what ways do you think NF1 influences (has influenced) your life?  
(Idem)
7. In what ways do you think NF1 influences (has influenced) the life of your affected relative/s?  
(Work/ social life/school/family)
8. What do you find are/have been the most difficult aspects of your life in relation to NF1? Do you remember any particular episode?
9. What about the positive aspects? Do you remember any particular episode?
10. Where and how did you get the information on NF1?
11. Have you ever had contact with other individuals with NF1 and/or their families?
12. Some people with NF join NF support groups. What is your take on this?

## CAUSES OF NF1/ TECHNOLOGIES

13. If you had to explain NF1 to someone, how would you explain it?  
(Definition)
14. In what way do you consider NF1 to be similar to or different from other health problems?
15. Why do you think people get NF1?
- (16. Some people refer to NF1 as a genetic disorder, what do you think about that?)
17. What do you think the future will be like for treating NF1?  
(Technologies)
18. New technology can give the possibility to know in advance if a baby has NF1, what do you think about it?

## MEDIA

19. Have you ever seen, read or heard on the media about a person affected by NF1?
- (20. If so, how much did you find the case close to the one of your relative/s?)
- (21. How do you see the future of your affected relative/s shaping up?)
22. Do you know any other person affected by NF1 who might be interested in participating to this study?
23. I have finished all my questions now, but is there anything else that you think we haven't talked about, something relevant to your life that you would like to say now?

## OUTLINE FOR INTERVIEW OF PROFESSIONALS

### POSITIONING THE SYNDROME

1. Could you tell me something about your professional knowledge and experience with NF1?  
(How often the syndrome is encountered/from which routes the patients come through)
2. Has your attitude towards NF1 changed over time? If so what made it change?
3. In what ways do you find NF1 to be different from other illnesses?  
(Positioning NF1: Serious disease? Do they think of it primarily as tumour generating, cognitively impairing)

### PROFESSIONAL ENVIRONMENT

4. How does NF1 usually come to be diagnosed, and what kinds of treatment pathways do your patients usually take?  
(What specialties are involved, at what points, and with what kinds of outcomes? Do they think this works well for patients?)
5. How well do you think the NHS manages patients with NF1? Where does the system fall down when treating NF1 patients and their families?
6. Do you think the genetic service is important for the clinical management of NF1? If so, in what ways? If not, what is important?

### COMMUNICATION WITH PATIENTS AND FAMILY NETWORKS

7. How do you normally communicate the news of having NF1 to patients and their families? Which images or examples do you normally use?
8. How do you explain the heritable/genetic aspects of NF1? Do these aspects play a role in how you talk with patients about NF1?
9. Could you describe a most common, difficult and memorable case you have encountered?  
  
(How do you think the patients and their families normally cope with the counselling and treatment?)  
  
(Which are the problems that you encounter most often in dealing with NF1 patients and with their families?)

## GENETIC, GENETIC RISK, HERITABILITY

10. How knowledgeable do you feel your patients are about genetics and NF1? Can you recall any episode to illustrate this?

11. How according to you, do patients and their families understand the heritability of NF1? Can you recall any episode to illustrate this?

12. How do patients and their families make sense of the high variability and unpredictability of NF1? Can you recall any episode to illustrate this?

## CLOSING

13. Which do you think will be the next steps for the treatment of NF1? How do you see the future developing with regard to genetics and their practice?

14. Have you already been involved in researches on NF1?

(15. What are your views on this research?)

16. I have finished all my questions now, but is there anything else that you think we haven't talked about, something relevant that you would like to say now?

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