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, October 2011.

I certify that all material in this thesis which is not my own work has been identified and that no material has previously been submitted and approved for the award of a degree by this or any other University.

(Signature) ……

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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