

Exploring the implications of genetic testing in mental health care

Submitted by

Christopher Elphick,

to the University of Exeter as a thesis for the degree of Doctor of Philosophy in Genomics in Society, January 2013

This thesis is available for Library use on the understanding that it is copyright material and that no quotation from this thesis may be published without proper acknowledgement.

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).....

Acknowledgments

I want to extend a warm thank you to my supervisory team who both went way beyond the call of duty to help me over the last four years. Professor Stephen Hughes met with me almost every single week over my time as PhD student and without his input, insights, questioning, and forensic eye for detail I would not be the person I am today. Doctor Susan Kelly is always encouraging and had time for me whenever I needed it. Her insights guided my ideas and her huge knowledge base really helped me to focus some of my thoughts over the PhD program. I hope that one day someone else can benefit from my help the same way I did from theirs – any form of written communication is a feeble attempt to express the gratitude I have for the pair of them. Professor Hughes is also going to be an excellent guitarist one day soon.

In addition to this I want to thank the staff and other students at Egenis, who provided an extremely stimulating learning environment and have always been friendly. Special thanks to Matt Hodges for his help with my first research proposal in the masters' year and Daniele Carrieri for his help with my NHS ethics proposal. In addition to this I want to thank all the people who participated in this research project – some of you were very welcoming to me and really helped me reach the projected number of interviews.

Any of this would have been impossible without the love and support of my family – Michael, Vicki, Daniel, Charlotte, Laura and Eliger. I would also like to thank Mei Kuen Wong for all her love and support. The same goes for David Leonard, Timian Brierley, Duncan Weatherhead, Jobis Linnelli, everyone at Kernow Karate and Michael Kirkpatrick – you all helped me through my darkest hours.

I would also like to extend my thanks to some people I have not had the pleasure of meeting in person, but who have been highly influential in shaping my approach to thinking and life. Several of these people do not have anything to do with any of what you are about to read in this thesis, but they all inspired me to keep going. Edward and Alex, Michael, Kim, Muhammad, Brian and Paul. One man that requires a special mention is Thomas Szasz, who passed away while I was writing this thesis, in September 2012. Had I not discovered his body of work I would not be the person I am today and this thesis would be *significantly different*. Although I do not claim to be an expert in his ideas, reading his legacy really changed the way I think about things and I owe him a great deal.

Abstract

Now is a time of dramatic change in mental health care as the world is witnessing a proliferation of research into the genetics of mental disorders. Despite several genetic test developments there is a paucity of qualitative research exploring the issues concerning its potential future introduction. This inspired my primary research question: What are the main implications regarding the developments being made in genetic testing for mental disorders in terms of their proposed introduction in a clinical setting? This was investigated through 33 semi-structured interviews with a range of psychiatric professionals from a single NHS trust location in the South West of England. As research has demonstrated that different medical professionals consider issues in mental health care in different ways (Colombo, *et al.* 2003 ; Fulford and Colombo, 2004) participants' personal constructs of mental disorder were examined to see if their accounts of the tests differed on the basis of their unique conceptualisations of mental distress. An additional component feature in this research relates to what these developments may ultimately *represent* or *provide* psychiatry and mental health care as a result of being able to consider mental disorders in terms of underlying biology. Historically there has been a persistent attempt to determine the underlying genetic components of mental distress, however, this always seems to fail or the next big development is always 'just around the corner' - this observation is considered when the major developments in psychiatric genetics are examined in light of the sociological field of the 'promissory nature of science' (Borup, *et al.* 2006) - I suggest that the developments in genetic testing for mental distress represent an iconic continuation of this process. Interview transcripts were subjected to thematic analysis and five themes were developed that cover aspects such as how the tests' introduction will alter perceptions in mental health care, issues concerning the tests' practical impact, their possible shortcomings, and how they may alter clinical practice. My findings indicate that, in the majority of themes, personal approaches to mental disorder do appear to influence participants' accounts of the tests. The overall trend is that if an interviewee personally endorsed a biological approach to understanding mental disorder they would be willing to see the tests used in clinical practice. There were two areas of thematic agreement between all psychiatric professionals regardless of their conceptualisations of mental disorder. These concerned the impacts genetic testing could have on different aspects of the legitimacy of mental disorders and the significance of using the tests to aid in treatment rather than diagnosis. Implications of my thematic findings for patient groups, mental health services, and policy makers are discussed.

Table of Contents

1.0 - Introduction	10
1.2 - Scale of mental disorders.....	12
1.3 - Official requests to conduct research into the underlying genetics of mental disorders.....	14
1.4 - Influence of advances and developments in personalised medicine	14
1.5 - Aims.....	16
1.6 - Thesis structure	16
2.0 – Literature Review	19
2.1 - The nature of psychiatry and mental disorders	19
2.1.1 - Mental disorders and psychiatric disorders.....	20
2.1.2 – A helpful take on Psychiatry	21
2.1.3 – A focus on psychiatric and psychological approaches	22
2.1.4 – Relationship to the characterisation of psychiatry.....	24
2.1.5 - The multidisciplinary team in psychiatry and mental health care.....	25
2.2 – Exploring the concept of mental disorder	26
2.2.1 - Psychiatry and psychology	27
2.2.2 – Medical/biological and psychological models of mental disorder.....	27
2.2.3 - Additional accounts of mental disorder	29
2.2.4 - Sociological perspectives regarding mental disorders.....	31
2.2.4 – Navigating the ambiguities of mental disorder	32
2.2.5 - Ontology and epistemology of social science research into mental disorders.....	33
2.3 - Models of mental disorder.....	34
2.3.1 - Brief background and how the methodology can be used when considering mental disorder	34
2.3.2 – A closer look at models	35
2.3.3 - Models of Madness - Sieglar and Osmond 1966	37
2.3.4 - A study of psychiatrists’ concepts of mental illness – Harland, <i>et al.</i> 2009.....	38

2.3.5 - Psychiatrists' models of mental illness and their personal backgrounds (Toone, <i>et al.</i> 1979 ; Kreitman, 1962).....	39
2.3.6 - Return to Fulford and Colombo research	39
2.3.7 - Quantitative and qualitative issues.....	39
2.3.8 - Addressing the implicit/explicit division	40
2.3.9 - Summary	40
2.4 - Previous research into genetic testing for mental disorders	41
2.4.1 - Summaries of relevant research	41
2.4.2 - Quantitative surveys of beliefs about genetic testing in psychiatry	42
2.4.3 - Qualitative studies of beliefs about genetic testing in psychiatry.....	43
2.4.4 – Summary	44
2.5 - Mental health genetic research initiatives and proposals in the United Kingdom	44
2.5.1 - Medical Research Council – Review of Mental Health Research Report 2010	45
2.5.2 - Direct-to-consumer genetic tests and the MRC suggestions.....	47
2.5.3 - Human Genomics Strategy Group Report January 2012 - ‘Building Our Inheritance’	48
2.5.4 - Translating genomic innovation to establish clinical validity and clinical utility	49
2.5.5 - Implications in this research	49
2.5.6 - Test criteria	50
2.5.7 - The CDC and the ACCE criteria	51
2.5.8 - EGAPP initiative and Testing for Cytochrome P450 Polymorphisms	52
2.5.9 - Reintroducing the ACCE criteria	53
2.5.10 - Summary	55
2.6 - The history of the field of psychiatric genetics.	55
2.6.1 - The field of psychiatric genetics around 1900 – early family and twin studies.	56
2.6.2 - Controversy in family and twin studies.	58
2.6.3 - Additional movement in a biological direction – 1930 to 1970.....	61
2.6.4 - Psychiatric medication and different hypotheses.	61
2.6.5 - The role and the influence of the DSM in psychiatry and mental health care.	63

2.6.6 - Linkage studies – 1970s, 1980s and 1990s.....	66
2.6.7 - Candidate gene association studies – post the 1990s.	69
2.6.8 - The Psynomics research.	70
2.6.9 - Genome Wide Association Studies and beyond – Post 2000.	72
2.6.10 - Summary.	77
2.7 - The sociology of expectation and the field of psychiatric genetics.	78
2.7.1 - The sociology of expectation	78
2.7.2 - The Sociology of Expectations in Science and Technology – central themes and findings.	80
2.7.3 - Expectation as Constitutive Force	80
2.7.4 - Expectations and Temporal Variability.....	82
2.7.5 - Expectations and Socio-spatial Variability.....	83
2.7.6 - Imagination, Materiality and Embodiment	86
2.8 - Summary.....	88
2.8.1 - Development of research questions and implications in this project.....	91
3.0 – Methodology.....	95
3.1 - Philosophical, Paradigmatic and Interpretive frameworks.....	95
3.1.1 - Interpretivist ontology, epistemology, methodology and method	96
3.1.2 - Revisiting the work of David Pilgrim	97
3.1.3 – Pilgrim’s ontology	98
3.1.4 – Pilgrim’s epistemology	98
3.1.5 – Additional sociological approaches	101
3.2 - Choosing methods	103
3.2.1 - Constructivist Thematic Analysis.	104
3.2.2 - ‘Inspired by’ Grounded Theory Thematic Analysis.....	105
3.2.3 - Overall analytical method	107
3.2.4 - Stages of Coding	109
3.3 - Determining participants models of mental disorder	112

3.4 - Participant recruitment.....	113
3.4.1 - Breakdown of interview participants (n = 33).....	114
3.4.2 - Execution of the interview	114
3.4.5 - Interview questions.	115
3.4.6 - Ethical considerations	116
3.4.7 - Credibility, transferability, dependability and conformability in this research.	116
3.4.8 - Credibility.	116
3.4.9 - Transferability.	117
3.4.10 - Dependability.	118
3.4.11 - Conformability.	118
4.0 - Findings.....	120
4.0.1 - Participant willing to use both tests in clinical practice	120
4.0.2 - Participant rejecting diagnostic test but endorsing pharmacogenomic test	120
4.0.3 – Participant rejecting both tests	121
4.0.4 - Summary of participants’ communicated models of mental disorder	121
4.1 - Theme 1 - Prioritisation of genetic explanations for the diagnosis and treatment of mental disorders	123
4.1.1 - Legitimacy of mental disorders	123
4.1.2 - Models and their influence on the legitimacy of mental disorders	124
4.1.3 - Legal issues regarding the legitimacy of mental disorders.....	127
4.1.4 - Streamlining different areas of practice	127
4.1.5 – Participants’ commentary relating to the DSM	134
4.1.6 - Resolution of diagnostic uncertainties and the prescription of appropriate medication	137
4.2 - Theme 2 – Receipt of a test result, practical considerations and their potential consequences	140
4.2.1 - Patient empowerment from receiving a test result	140
4.2.2 – Additional benefits of receiving a diagnosis	142
4.2.3 - Using the tests for early intervention	142

4.2.4 - When and where.....	144
4.2.5 - Prophylaxis	145
4.2.6 - Negative consequences of receiving a test result.....	146
4.2.7 - Education - management, applied use and consent.....	148
4.2.8 – Counselling services	152
4.2.9 - Practitioner education	153
4.3 – Theme three – Representations of mental disorder in light of the tests’ introduction	155
4.3.1 - Public perceptions of mental disorders and the genetic testing.....	155
4.3.2 – The media	155
4.3.3 - Public perceptions in light of the tests’ introduction	158
4.3.4 - Influence of participants’ models of mental disorder on thoughts concerning public perception	160
4.4 - Theme 4 - What the tests cannot do	161
4.4.1 – Participants’ criticisms of the Psynomics tests	162
4.4.2 - Psynome 1	162
4.4.3 - Psynome 2	164
4.4.4 - Additional accounts - Existing techniques.....	165
4.5 – Theme 5 - Using genetic testing for the treatment of mental disorders rather than the diagnosis of mental disorders.....	171
5.0 – Discussion	175
5.1 – Participants’ models of mental disorder	175
5.2 - Clinical Utility in the ACCE criteria.....	177
5.3 - Legitimacy of mental disorder	182
5.3.1 - Legitimacy defined	182
5.3.3 – Elliot Friedson and Professional dominance	185
5.3.4 - Further aspects of legitimacy as a sociological process.....	187
5.3.5 - Legitimacy is a problem in the construction of social reality	188
5.3.6 - Legitimacy is fundamentally a collective process	189

5.3.7 - Legitimacy depends on apparent consensus in the local situation that most people accept the object as legitimate	190
5.3.8 - Potential implications for different stakeholders.....	191
5.4 - Treatment rather than diagnosis.....	192
5.5 - Tests in practice.....	195
5.5.1 - Practice/professionals	196
5.5.2 – Patients/experience of mental health care.....	196
5.5.3 - Policy/delivery implications	197
5.6 - Limitations of the study	197
5.7 – Conclusion.....	199
6.0 - Appendix.....	206
6.0.1 – Contact Letter	206
6.0.2 – Participant information sheet.....	207
6.0.3 – Participant reply slip	210
6.0.4 – Participant consent form	211
6.0.5 – Psynomics Information presented to the interview participant	212
7.0 - Reference list.....	216