

The House of Lords Science and Technology sub-Committee II Inquiry into Genomic Medicine

Evidence submitted on behalf of the ESRC Centre for Social and Economic aspects of Genomics (CESAGEN) Cardiff and Lancaster Universities

1. Introduction

Cesagen's 'Biomedicine, identity and behaviour' theme has produced a large body of detailed, ethnographic work examining on biomedical knowledge, genetic medical conditions, kinship, personal identity and the communication of genetic risk information within families (cf. Featherstone et al, 2005,2006; Arribas-Ayllon et al., 2008a, 2008b). Its research agenda (2007-2012) is now turning to identification of 'susceptibility genes' for complex multifactorial conditions in the field of psychiatry. This prior and ongoing work informs this response.

Our submission is focused on the following questions presented in the call for evidence under the heading '**Use of genomic information in a healthcare setting**':

What impact will genomic information have on the classification of disease?

How will it affect disease aetiology and diagnostic labels?

How useful will genomic information be as part of individualised medical advice?

What provisions are there for ensuring that the individual will be able to understand and manage genomic information, uncertainty and risk?

2. What impact will genomic information have on the classification of disease? How will it affect disease aetiology and diagnostic labels?

Neuropsychiatric disorders represent a rapidly changing field where genetic medicine is emerging as an important diagnostic, treatment and risk assessment tool. The identification of 'susceptibility genes' for psychiatric conditions (including schizophrenia and bipolar disorder), has the potential to transform the classificatory basis of psychiatric diagnosis and treatment, and to challenge the epistemological basis of at least some psychiatric practice.

Within the field of psychiatry, there has been a long history of contested classifications, particularly for psychosis and bipolar affective disorders. At present, these disorders are treated as distinct entities with separate underlying disease processes and treatments. However, many individuals with severe psychiatric illness ("psychosis") have both prominent mood and psychotic symptoms 'raising the possibility, indeed the likelihood, that there is not a neat biological distinction between schizophrenia and bipolar affective disorder' (Craddock and Owen, 2005: 364). Recent evidence suggests that the current criteria used for the classification of these conditions does not

map neatly onto patient experiences and symptoms. Genetic epidemiology has already proved influential in shaping the nosography of psychiatry and recent developments in molecular genetics are beginning to provide evidence to challenge these traditional classifications. This makes it increasingly difficult for psychiatrists to unambiguously assign patients to distinct categories of major psychosis.

In addition, although the identification of susceptibility genes is at an early stage they are already having an impact on family understandings of risk and transmission (Meiser et al., 2005), and 'will have a major impact on our understanding of disease pathophysiology and will lead to changes in classification and the clinical practice of psychiatry' (Craddock, et al., 2005: 193). It is therefore timely and important to examine the personal and professional responses to these new challenges, the role of stigma and the personal consequences of the practices of medical classification and the assessment of risk.

3. How useful will genomic information be as part of individualised medical advice? What provisions are there for ensuring that the individual will be able to understand and manage genomic information, uncertainty and risk?

Genetic testing for psychiatric disorders is becoming commercially available with venture capitalists and scientists seeking to establish niche markets by selling 'direct-to-consumer' testing. Psynomics (www.psynomics.com) have developed a test that helps diagnose people with bipolar disorder (Couzin, 2008). Companies such as SureGene (www.suregene.net/home.aspx), are developing tests for other psychiatric disorders while NeuroMark (www.neuromark.com) are marketing a test for pharmacogenetic responses to anti-depressant treatment. The success of the business model in driving these developments is likely to encourage further biotech companies to circumvent existing psychiatric healthcare models in favour of personalised medicine. In the UK, recent media coverage (*Observer*, 03/02/08; *The Daily Telegraph*, 4/02/08; *The Express*, 5/02/08; *The Times*, 10/03/08) has raised public awareness and expectations of these scientific developments as well as fears and concern about the commercial availability of predictive and diagnostic testing. Psychiatrists and doctors have warned that the proliferation of such direct-to-consumer testing will mislead and confuse consumers (*Observer*, 03/02/08; *BBC News*, 11/03/08). Providing individuals with the likely risk of developing psychiatric disorders is not straightforward, and may not account for the complex interaction of genetic and environmental factors.

While susceptibility testing within psychiatric practice is only a theoretical possibility at present, the identification of susceptibility genes has significant implications for individual patients and their family-members.

New exploratory research is needed in order to assess the likely consequences of these developments for individuals and family members, for psychiatrists, genetic specialists and mental health professionals, and the potential demands on healthcare services. The ongoing collaboration between Cesagen, Psychological Medicine, Medical Genetics, Social Sciences, and the Wales Gene Park at Cardiff University will provide an evidence based assessment of the likely personal and professional consequences of new genetically-based diagnostic criteria and risk information. It will also inform the ethical debate concerning the consequences of risk evaluation for psychiatric conditions.

References

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