

**THE HOUSE OF LORDS SELECT COMMITTEE ON SCIENCE AND
TECHNOLOGY**

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

This evidence is submitted on behalf of the ESRC Centre for Economic and Social Aspects of Genomics (Cesagen), a collaboration between Cardiff and Lancaster Universities. It is part of the ESRC-funded UK Genomics Network which was established to provide significant research capacity to examine developments in genetic technologies and their social implications. Cesagen was established in 2002 and has completed a substantial programme of research on biomedical genetics. Staff are drawn from a number of disciplines and work in interdisciplinary teams, which allows all aspects of social and scientific issues to be examined in a collaborative manner. The views represented here are based on our shared research expertise, but do not represent the views of the Economic and Social Research Council.

Our submission is addressed primarily to the Committee's interest in **The use of genomic information in a healthcare setting.**

2. Summary

We summarise two key, closely related issues concerning the development of genetic testing and its implications for primary and secondary health care. We identify the emergence of the 'genetic iceberg' of anxious patients who have moderate risk but limited access to counselling or other healthcare resources. We also note the spread of genetic medicine into clinical specialties that have not hitherto been concerned primarily with genetic testing and its management. We note that separately and in conjunction these two trends are likely to create a burden of need for genetically-based counselling, monitoring and support that will outstrip current provision. The submission is based on fundamental and strategic research carried out by ourselves and others in the UK research community.

3. Current trends

The following trends form the background to this evidence.

- i. The identification of risk and susceptibility genes for an ever widening range of conditions.
- ii. The transformation of disease categories and classifications, based on genetic evidence.
- iii. The likely expansion of genetic testing, including population screening, for major disorders: cancers, haemochromatosis, sickle-cell, thalassaemia.

- iv. The translation of genetic research into major clinical specialties – cardiology, neurology, haematology, psychiatry.
- v. The emergence of genetically-based methods of diagnosis and risk assessment beyond the conditions, and beyond the clinics, hitherto the preserve of specialists in genetic medicine, including genetic counsellors.

4. The research evidence

Our observations are based on the following robust research findings that have been replicated across a number of conditions:

4.1 Risk and anxiety

The identification of ‘risk’ – especially for a medical condition that is life-threatening or that may lead to significant impairment – can lead to expressed anxiety on the part of those at risk. Anxiety *per se* is not correlated with the level of risk in a simple fashion. Anxiety is most likely to be expressed by individuals identified as having a ‘moderate’ risk. Those with a low risk and those with high risk do not express the same anxieties (for different reasons). (Bharadwaj, 2006)

While those with a low risk may feel that that is sufficient cause for reassurance, those with high risk have greater access to genetic counselling, medical or surgical intervention (e.g. prophylactic surgery for breast cancer), regular screening (e.g. for colorectal cancer) and clinical treatment (e.g. haemochromatosis). In other words, anxiety is high when scarce resources of healthcare and counselling are not available to those with ‘moderate’ risk.

There is, therefore, the clear likelihood of creating a widening pool of anxious pre-patients who are at risk, but who do not qualify – on grounds of level of risk – for specialist intervention. Existing specialist services in genetic medicine and genetic counselling do not currently have the capacity to take up this potential burden of monitoring, advice and healthcare provision.

4.2 Translation of genetic medicine

The translation of genetically-based medicine into clinical specialisms beyond Genetic Medicine itself raises serious implications for the conduct of clinical medicine and intervention into family relations.

Within the specialty of medical genetics, there are highly developed professional procedures for the management and disclosure of genetic information. The translation of genetically-based diagnosis and prediction into other specialties, with practitioners not trained in genetic counselling or supported by specialised counsellors or genetic nurses, may lead to interventions that do not abide by such standards of practice. There is, for instance, as yet unpublished evidence of clinics contacting family members directly and/or engaging in much more directive interventions than has been the norm in medical genetics (where the practice has been based on non-directive counselling and the avoidance of direct family contact, except through and with the express consent of probands). (Marks, D, 2002)

4.3. Genetic information

All of the available social research indicates that the transmission of genetic information to probands, and from them to family members, is variable and contingent. (Gaff, C, 2007) Information imparted in the genetics clinic is often remembered and interpreted in ways that do not reflect the intentions of the professional practitioners providing such information. Genetic information (such as risk values and patterns of inheritance) is interpreted by family members in accordance with lay theories of inheritance, lay understandings of risk values, and the dynamics of family communication. (Featherstone, K, 2006, Arribas-Ayllon, M, 2008 a,b) The transfer of genetic information between family members is heavily dependent upon mutual perceptions of who is able to ‘cope’ with that information, and who needs that information. Such lay assessments and understandings cannot be assumed to be congruent with geneticists’ and other professionals’ assessments nor with biological/medical models of inheritance. (Atkinson, P, 2003) There is, therefore, considerable scope for misalignment between professionals and clients. There is evidence that such problems of misalignment can be exacerbated when diagnoses and risk assessments are being provided by specialists who are not familiar with genetic conditions: our own family interviews with worried parents of children with conditions such as haemophilia. (Gregory, M, 2007, Boddington, P, 2008) If genetic medicine is to be used to inform individualised medical advice, it is clear that detailed consideration needs to be given to the management of genetic information, not only in specialist genetics clinics, but – even more pressingly – in primary and secondary healthcare settings where trained genetic specialists are not available.

5. Training and guidance

As we have indicated, the expansion and translation of genetically based medicine is predicted to create a considerable burden on healthcare professionals and services. Unless substantial resources are made available for the continuing professional education of existing practitioners, and for the training of new genetic counsellors, then the visions for genetic medicine (for instance as outlined in the 2004 white Paper) (Department of Health, 2003) cannot be realised. There is the danger that genetic information – poorly understood by lay clients – will not result in well-informed publics, and will not result in appropriate health behaviour. At present the only advanced training for genetic counsellors in the UK is provided through Masters courses at Manchester and Cardiff Universities. (for links, see references) These two courses currently produce c.25 graduates per annum (including overseas students who do not intend to practise in the UK). It is clear that such small numbers are insufficient to meet the needs of the public.

There is equally clear need for codes of professional conduct and protocols of practice to be agreed between specialists in medical genetics and other specialists, given that the latter will increasingly find themselves transmitting genetic information in clinical settings. Translation of genetic medicine needs to be accompanied by the translation of best practice into new fields of specialist practice.

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