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MEMORANDUM FROM THE ARTS AND HUMANITIES RESEARCH COUNCIL TO THE HOUSE OF LORDS SCIENCE AND TECHNOLOGY COMMITTEE INQUIRY INTO GENOMIC MEDICINE

1. The Arts and Humanities Research Council (AHRC) welcomes this opportunity to respond to the committee's inquiry. This response does not include or necessarily reflect the views of the Science and Innovation Group in the Department for Innovation, Universities and Skills.
2. The AHRC supports research within a huge subject domain from traditional humanities subjects, such as history, modern languages and English literature, to the creative and performing arts. The AHRC funds research and postgraduate study within the UK's higher education institutions. In addition, on behalf of the Higher Education Funding Council for England, it provides funding for museums, galleries and collections that are based in, or attached to, higher education institutions in England.
3. This information in this response comes (except the *Research and Scientific Development* section, as indicated below) from Professor Graeme Laurie, Director of the AHRC Research Centre for Studies in Intellectual Property and Technology Law at the University of Edinburgh and Mr Shawn Harmon, Research Associate with the AHRC Centre and with Innogen (sponsored by the ESRC) also at the University of Edinburgh. The response focuses on issues from the legal, ethical and/or regulatory perspectives, being the focus of the work of the AHRC Law Centre. Further details can be seen at <http://www.law.ed.ac.uk/ahrc> and also <http://www.innogen.ac.uk>.

Policy Framework

- ***Does the existing regulatory and advisory framework provide for optimal development and translation of new technologies? Are there any regulatory gaps?***
4. We contend that the existing regulatory framework does not optimise the development and translation of new technologies. There are three hurdles of particular significance.
 5. Regulatory Joined-up-ness: There exists 'innovation drag' as a result of regulatory complexities (e.g.: regulatory overlap and co-regulation by a variety of bodies, from the Human Fertilisation and Embryology Authority (HFEA) and Human Tissue Authority (HTA), to UK Stem Cell Bank (UKSCB), to the General Medical Council, the patents regime and more. The inefficiencies created by multiple overlapping recommendations, directions, guidances, and laws from bodies/agencies (who do not necessarily coordinate or even communicate) with an interest in different but overlapping stages of innovation, complicates and elongates the journey from idea to socially-useful output. It also makes it difficult for public actors to steer innovation in socially-useful directions. In short, there needs to be regulatory 'joined-upness' so as to promote timely and valuable innovation. The Committee has the opportunity to map these regulatory pathways

and to recommend how more effective and efficient trajectories might be developed.

6. The role of ethics committees: A particular ‘pressure point’ in this arena – that is, activities directed at the translation of research into products/processes directed at humans – which deserves closer attention is the remit and transparency of local or multi-centre ethics committees; more particularly, how to articulate the scope of the former and ensure the latter.
7. Access to research data: There are barriers to researcher/innovator access to data and much work has been done with a view to examining the knowledge enclosure tendencies of existing intellectual property regimes. The Committee is, however, in a position to examine the UK intellectual property regime and its interactions with European and international institutions, and more particularly the application of these regimes to genomic medicine. One fruitful avenue to monitor in depth is the initiative from the UK Stem Cell Bank to rely more on Open Science. It is far from clear that this will serve the range of stakeholder interests.
- ***In what way is science and clinical policy decision-making informed by social, ethical and legal considerations?***
8. We would suggest that there are five core challenges facing the optimal governance of genomic medicine and which involve the above considerations.
9. Consent: The imperative to obtain informed consent has become the dominant paradigm in biomedical governance but we would point out that its origins lie in medical research focussed on the human body and that its application to research involving medical or genomic data should be explored in more depth. Such an imperative may stand in the way of valuable medical research and act as a hurdle to striking an optimal balance of interests. Consent is not a social value in itself, but merely a means to respect individuals and their interests; as we indicate below, it is neither necessary nor sufficient to protect the core individual interests involved in research involving medical/genomic data, viz privacy interests. We would urge the Committee to explore the debate about the value and limits of consent and to ask whether we ‘fetishise’ consent as a regulatory tool at the expense of other options.
10. Confidentiality: As a recent report from the Academy of Medical Sciences indicates¹, the current regulatory regime that operates within the United Kingdom with respect to protection of privacy does not take advantage of flexibilities within the law which provide for the adequate protection of privacy while also promoting medical research when informed consent is neither practicable nor possible. We would also refer the Committee to the recent report from the Article 29 Working Party on Data Protection which discusses e-health records and the possibility of regulation regimes that promote a public interest mandate.² We would urge the

¹ Academy of Medical Sciences on *Personal Data for Public Good: Using Health Information in Medical Research* (2006), report available here:

<http://www.acmedsci.ac.uk/p99puid62.html>

² Article 29 Data Protection Working Party, ‘Working Document on the processing of personal data relating to health in electronic health records (EHR)’ 15 February 2007.

Committee to consider, in particular, the role and approach of the Patient Information Advisory Group which takes a very consent-based stance to its work. This can be contrasted with the Privacy Advisory Committee for Scotland, of which Professor Graeme Laurie is Chair, and which seeks a balance of public and private interests.³

11. **Confidence:** Public trust and confidence is crucial to effective governance regimes and we would suggest that particular close attention should be paid to questions of access to genomic data. Good governance regimes should be transparent, robust, reasonable, involve clear due process for all parties, and be subject to effective oversight. We would point to the example of the UK Biobank Ethics and Governance Council, chaired by Professor Graeme Laurie, which is developing considerable experience in this realm; crucial to the success of the Council is a close working relationship with UK Biobank itself.
12. **Commercialisation:** Our research, and that of others, has revealed some evidence of public unease about the role of commercialisation of genetic/genomic research.⁴ While we accept commercialisation as a reality, we would nonetheless suggest that the Committee consider carefully whether and how such public attitudes might be taken into account.
13. **Collaboration:** International collaboration is essential if we are to realise the full promise of genomic medicine. The same is true in terms of governance, and in many ways the United Kingdom is leading the international field; once again, we offer the example of the UK Biobank Ethics and Governance Council. On international collaboration, we would also point to the example of the Public Population Project in Genomics (P3G) which is seeking harmonisation of efforts, both scientific and regulatory, among biobanks across the globe.⁵

Research and Scientific Development

- *What is the state of the science? What new developments are there? What is the rate of change?*

14. The AHRC has funded 'Philosophical Issues in Genomics'; a research grant award made to Professor John Dupré and Dr Maureen O'Malley at the ESRC Centre for Genomics in Society (Egenis); the information in this section comes from them. The award of £93,838 ran from 01/03/06 to 29/02/08 and addressed two topics that have enormous potential for relevance to future medical practice; systems biology and microbiology. Systems biology is an attempt to apply sophisticated computational methods to modelling complex biological systems. It is widely seen as a successor science to genomics, and is in part a response to the growing realisation that the once widespread assumption that causation ran in a linear fashion from DNA through RNA to proteins is entirely mistaken. Systems

³ Privacy Advisory Committee for Scotland, more information here:
<http://www.isdscotland.org/isd/2466.html>

⁴ Haddow, G., Laurie, G., Cunningham-Burley, S., & K. Hunter, 'Tackling Community Concerns About Commercialisation and Genetic Research: A Modest Interdisciplinary Proposal' (2007) *Social Science and Medicine*, 64: 272-282.

⁵ Population Project in Genomics: <http://www.p3gconsortium.org/>

biology explores the networks of typically cyclical and feedbacked causal interactions that constitute the functioning of biological systems. Although research in this area has yet to reach the point where it is directly applicable to interventions in human biological systems, this is likely to change. One area that is widely anticipated is the use, in part, of in silico models deriving from systems biology for the testing and discovery of new drugs.

15. Metagenomics can be seen as a subarea of systems biology. The systems it explores are microbial communities, reflecting the understanding that microbes generally function not as isolated individuals but rather as complex communities of cooperating microbes with very complex division of labour. This field has almost unlimited potential impact on medicine. About 90% of the cells that constitute the functioning human body are in fact microbial symbionts, and about 99% of the genes in a human body reside in these microbes. The human microbiome project is now beginning the project of cataloguing these genes. Microbial symbionts are now understood to be essential for human functioning, in areas including most notably digestion, immune response, and development. There is growing evidence that gene expression is mutually modulated by human and symbiotic microbial cells. These developments present nothing less than a reconceptualisation of what a human body is, and can therefore potentially revolutionise our understanding of the system that medicine aims to influence.
16. Work in Egenis to date has been concerned primarily with mapping these developments and communicating to scholars in a range of disciplines (including philosophers of biology) the importance of the insufficient attention that has been paid to microbes. Further research is planned by the award holders to expand this work in the direction of its relevance to medical practice.

Data Use and Interpretation

- ***What are the implications of the generation and storage of genome data on personal data security and privacy, and on its potential use or abuse in employment and insurance? How should these be addressed?***
17. The Committee should have reference to the quality work that has already been undertaken in the area of data storage, data security and data use in the genomic context.⁶ Generally, the issues of concern in the employment and insurance context – which turn on genetic determinism, consent, discrimination, and data security – have not changed.

Translation

- ***How meaningful are genetic tests which use genome variation data? What progress has been made in the regulation of such tests?***

⁶ See Nuffield Council, *Genetic Screening: Ethical Issues* (London: NCB, 1993), chs. 6 and 7 Human Genetics Commission, *Whose Hands On Your Genes? A Discussion Document on the Storage, Protection and Use of Personal Genetic Information* (London: HGC, 2001), Human Genetics Commission, *Inside Information: Balancing Interests in the Use of Personal Genetic Data* (London: HGC, 2002),.

18. Again, there has been extensive consideration of the ethical, legal, social and practical aspects of this issue.⁷
19. In addition to *Genes Direct* (2003),⁸ the Human Genetics Commission (HGC) has recently issued a new report entitled *More Genes Direct* (2007),⁹ which identifies key issues in the genetic testing area as (1) pre-market review of tests, (2) quality assurance of testing services, (3) advertising and promotion controls, and (4) access to independent, impartial advice on the part of consumers. The HGC rightly identify a need for stricter controls in this field combined with improved NHS genetics services, and recommends that certain tests should only be offered through qualified health professionals. We endorse this position.
20. There are also on-going concerns about international access to, and internet marketing of, genetic tests. In 2007, the OECD adopted Guidelines aimed at quality assurance, analytic accuracy of tests, and information provision,¹⁰ and EuroGentest is examining the possibility of requiring labs, clinical services and professionals to be (ISO) accredited.¹¹ Generally, it is recognised that there needs to be accelerated cooperation around international standardisation of definitions, harmonisation of quality assurance standards, and controls on advertising and administering tests. Use of the internet makes harmonisation and regulatory cooperation all the more critical.
21. The above supports the claim that there is still much to do in the regulation of this field and in managing/rationalising the provision of genetic testing services.

Use of genomic information in a healthcare setting

- ***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?***
 - ***Should there be a regulatory code (mandatory or voluntary) covering the provision of this advice?***
22. There is already a plethora of advice and guidance in existence about the handling of medical/genetic data. We would caution against yet another iteration unless this comes from a sufficiently high-level, authoritative body and is accompanied by a clear explanation of how this new guidance should be read with existing provisions.

⁷ See ECG, *Ethical, Legal and Social Aspects of Genetic Testing: Research, Development and Clinical Applications* (Brussels: EU, 2004), and the ongoing work of EuroGentest (<http://www.eurogentest.org/>), an EU-funded Network of Excellence with a remit of encouraging harmonisation of standards and practices throughout the EU and beyond.

⁸ HGC, *Genes Direct: Ensuring the Effective Oversight of Genetic Tests Supplied Directly to the Public* (London: HGC, 2003).

⁹ HGC, *More Genes Direct: A Report on Developments in the Availability, Marketing and Regulation of Genetic Tests Supplied Directly to the Public* (London: HGC, 2007).

¹⁰ OECD, *Guidelines for Quality Assurance in Molecular Genetic Testing* (Paris: OECD, 2007).

¹¹ See EuroGentest, *Recommendations for Genetic Counselling Related to genetic Testing (Draft 2)*, 2007.



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