



The Royal Academy
of Engineering

Genomic Medicine

Response from The Royal Academy of Engineering to the House of Lords Science
and Technology Sub-committee II

April 2008

Introduction

The Royal Academy of Engineering is pleased to contribute to the House of Lords Science and Technology sub-committee's inquiry into Genomic Medicine. This response has been compiled using contributions from key Fellows of the Academy.

The Academy is content for its input to be made public and would be pleased to provide supplementary evidence if required.

Responses to specific questions

1. What is the state of the science?

- 1.1 Genomic science has been driven by the completion of sequencing of the genome. There is strong correlation in research between post-genomic analysis and the relationships between disease and specific genes.
- 1.2 A thorough analysis of the consequences arising from the multiple and systems-wide aspects of genomics is needed. This progress is hindered by technical constraints and a lack of incentives to encourage collaborations exploring the synergistic effects across disciplinary boundaries.

2. Who is taking the lead in the consideration and co-ordination of research and the development of new technologies?

- 2.1 Research councils are taking the lead. This has been driven by academics on research council panels, international academics and other interested bodies, including several charities (e.g. the Wellcome Trust and the Leverhulme Trust).
- 2.2 There seems to be poor coordination between the various research councils and also with the NHS. This is partly because research councils such as BBSRC, EPSRC and MRC have developed strong independent strategic agendas of their own. However, interactions between research councils are improving.

3. How does research in the UK compare internationally? How much collaboration is there?

- 3.1 Research in the pure laboratory-based life sciences is strong in the UK. However, life sciences and genomics in the USA are increasingly moving towards quantitative methods and analysis. This aspect of genomic research is not yet strong in the UK. To make the most of post-genomic developments in a clinical setting, it is vital that quantitative system-wide approaches, using better diagnostics and mathematical models, are developed to help diagnosis and treatment.
- 3.2 The Royal Academy of Engineering would like to see a greater involvement of engineering and engineering-based ideas to help drive the quantitative revolution necessary to make full use of genomic developments in medicine.
- 3.3 The BBSRC in particular have invested in quantitative research, but there is some resistance from research groups who focus on qualitative analysis of isolated genomic effects. Collaboration is patchy and there is still a tendency for research groups to work in isolation in order to demonstrate excellence.

3.4 There are some perceptions that the NHS research budget is unfocused and too targeted towards clinical care.

4. What are the current research priorities?

4.1 We believe that others will be better suited to describing research priorities in detail. However it is worth mentioning that systems biology is a relevant acknowledged priority of the BBSRC. This was also an area that The Royal Academy of Engineering and the Academy of Medical Sciences highlighted as a key priority in the report *Systems Biology and Medicine: a vision for engineering and medicine*¹.

5. What is the role of industry?

5.1 There is some very good industrial involvement occurring, particularly when specific gene targets are found that can be rapidly commercialised. However industry will usually seek two-year returns at most and so industrial involvement in more fundamental research areas is weak. It is unrealistic to expect industrial support for front line clinical medicine in the public sector except where technological solutions can be developed and sold.

6. What are the implications of developments in genomic technologies for the training of medical specialists and other health professionals?

6.1 Molecular based medicine, which has arisen because of the major developments in molecular and cellular biology and the advances in ICT and computing over the last 60 years, has led to a whole new era of medicine developing.

6.2 Current medical school curricula are not directly related to these developments. Molecular and cellular biology and genetics are taught, but they are usually not taught in relation to the rapidly developing fields of genomics, proteomics and the other 'omics' fields. A thorough review of the requirements for building human resources in these areas is needed.

7. In what way is science and clinical policy decision-making informed by social, ethical and legal considerations?

7.1 The Royal Academy of Engineering, in collaboration with Engineering Council UK and a number of the leading professional engineering institutions, has created a Statement of Ethical Principles² to which it believes all professional engineers and related bodies should subscribe. The Statement is fully compatible with the principles in the UK Government Chief Scientific Adviser's Universal Ethical Code for Scientists³.

7.2 The principles within the Statement and Universal Ethical Code for Scientists should guide the work of scientists and engineers working with genomics.

7.3 The sharing of sensitive personal data is covered by a complex framework of laws, conventions and guidance. Legal considerations include:

¹ http://www.raeng.org.uk/news/publications/list/reports/Systems_Biology_Report.pdf

² http://www.raeng.org.uk/policy/ethics/pdf/Statement_of_Ethical_Principles.pdf

³ http://www.berr.gov.uk/dius/science/science-and-society/public_engagement/code/page28030.html

- the European Convention on Human Rights (ECHR) Article 8 which guarantees “respect for private life, home and correspondence”;
- the Council of Europe Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data⁴;
- Directive 95/46/EC of the European Parliament and of the Council of 24 October 1995⁵ (the EC Directive) on the protection of individuals with regard to the processing of personal data and on the free movement of such data;
- the UK Data Protection Act 1998⁶, which purports to implement the EC Directive; and
- the judgment of the European Court of Human Rights in the recent case *Copland v the UK*⁷ which ruled on the need for specific statutory authorisation and effective monitoring of any sharing of personal data without consent.

7.4 The combined effect of these laws appears to require that processing of personal data requires informed and explicit consent by the data subject or explicit and detailed authorisation by legislation coupled with effective monitoring to deter and detect abuse.

8. 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?

8.1 Genomic data is or may be sensitive personal data as it may reveal, for example, familial relationships, ethnic origins, medical conditions, predisposition to medical conditions or possible personality traits. Even if the data is anonymised (i.e. not linked to the individual’s name, address or other identifying attributes) it may still be or become personal data if it is unique to one identified or identifiable natural person.

8.2 ‘Processing’ is defined very widely by the EC Directive, as “any operation or set of operations which is performed upon personal data, whether or not by automatic means, such as collection, recording, organisation, storage, adaptation or alteration, retrieval, consultation, use, disclosure by transmission, dissemination or otherwise making available, alignment or combination, blocking, erasure or destruction”. So even ‘anonymisation’ of personal data is processing, and requires authorisation.

8.4 Data security includes data integrity (accuracy and protection from deliberate or accidental corruption), confidentiality (protection from unauthorised disclosure) and availability (assurance that the data will be available for authorised use when required and that access will not be prevented by unreliable systems or a denial-of-service attack).

8.5 Data security is difficult to achieve, expensive to implement effectively, and may have serious implications for the usability of systems that process the data. Security is difficult to achieve because of the need to analyse the

⁴ European Treaty Series, Nos. 108 and 181

⁵ http://ec.europa.eu/justice_home/fsj/privacy/docs/95-46-ce/dir1995-46_part1_en.pdf

⁶ http://www.opsi.gov.uk/acts/acts1998/ukpga_19980029_en_1

⁷ EuCtHR judgment of 3 April 2007, which became final on 3 July 2007

possible ways in which security could be breached and to then create adequate barriers to such breaches. In the case of computer-based systems, no practical amount of testing will provide a high degree of confidence that the systems are secure, so confidence in security has to be based on rigorous analysis. This is generally impracticable for commercial software because the necessary information is not available.

8.6 Security is expensive because the assurance activities require significant effort from skilled staff. This is because the data may need physical protection during collection, processing, archiving and destruction, and because commercial off-the-shelf systems may be inadequate. Security conflicts with ease of use because it is necessary to authenticate legitimate users in order to protect against unauthorised disclosure. Personal data must be held securely; otherwise the data controller cannot guarantee that there will be no unauthorised processing.

9. Is other medical information recorded in a suitable format to allow optimal interpretation of genomic data? How should genomic data be brought together with other health information?

9.1 Data formatting of medical and biological information is a complex issue. This has to be seen in the context of the various levels of the human organism which essentially comprises systems, viscera, tissues, cells, proteins and genes. This structure is often referred to as the Biological Continuum. At the upper levels of the Biological Continuum (i.e. systems, viscera and tissues) there are now well established international standards for data formatting. The key standard among these is the Digital Imaging and Communications in Medicine (DICOM) standard.

9.2 At the cellular level, there are standards for both optical and electron microscopy. However at the lower levels, i.e. the protein and gene levels, there is a wide range of standards. A significant amount of international work is occurring to integrate and co-ordinate these standards.

9.3 Genomic data should be brought together with other health information. This integration forms the basis of what is now termed 'molecular based medicine' and it is only effective if data is considered, wherever possible and appropriate, across the Biological Continuum.

10. Is genomic information published, annotated and presented in a useful way? Should there be a common, public database? If so, who should fund, and have responsibility for, such an initiative?

10.1 Any genetic information database should be developed through international collaboration. Genomics present universal problems where data and understanding would need to be coordinated and shared internationally.

10.2 If a public database were developed we would recommend that the issues surrounding data security (as explained in section 8.) are taken into consideration.

11. 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?

- 11.1 Genomic information is unlikely to be particularly useful at the level of the non-specialist. It is very unlikely that the individual will be able to understand and manage pure genomic information data.
- 11.2 However, the interpretation of diagnostic data derived from genomic information will be useful for individualised medical advice. This is comparable to diagnostics derived from other medical technologies.