BIOSCIENCES FEDERATION

Genomic Medicine

A joint response to the Lords Science & Technology Committee

April 2008

Introduction

The **Biosciences Federation** (BSF) is a single authority representing the UK's biological expertise, providing independent opinion to inform public policy and promoting the advancement of the biosciences. The Federation was established in 2002, and is actively working to influence policy and strategy in biology-based research – including funding and the interface with other disciplines - and in school and university teaching. It is also concerned about the translation of research into benefits for society, and about the impact of legislation and regulations on the ability of those working in teaching and research to deliver effectively. The Federation brings together the strengths of 44 member organisations (plus seven associate members), including the Institute of Biology**. The Institute of Biology** is an independent and charitable body charged by Royal Charter to further the study and application of the UK's biology and allied biosciences. It has 14,000 individual members and represents 37 additional affiliated societies (see Appendix). This represents a cumulative membership of over 65,000 individuals, covering the full spectrum of biosciences from physiology and neuroscience, biochemistry and microbiology, to ecology, taxonomy and environmental science.

Policy framework

1. The Department of Health, primarily under the aegis of the National Institute for Health and Clinical Excellence (NICE) and the National Horizon Scanning Centre (NHSC), together with the Department for Innovation, Universities and Skills are responsible for setting and reviewing policy in the area of genomic medicine. However, it is not clear to us how effective these departments are in anticipating the relevance of scientific developments to future policy.

2. Scientific advice on policy development in this area is provided by research funders, particularly the Medical Research Council and the National Institute for Health Research (NIHR), learned and professional societies, and the scientific advisory bodies in the relevant government departments such as the Human Genetics Commission (HGC) and the NHSC.

3. The UK framework does not compare favourably with that of the US, where the Food and Drug Administration (FDA) appears to be more proactive and forward-thinking than its UK counterparts. The National Human Genome Research Institute is an extremely valuable asset to the US framework. Given time, we would expect the Office for Strategic Coordination of Health Research (OSCHR) to significantly enhance the UK policy framework, provided that the advisory capacity of this body is joined –up with the regulatory mechanisms of the relevant government departments and their agencies.

4. We believe that it should be possible to respond to policy issues through the improved use of existing structures, and that there is no clear need for additional regulatory or advisory bodies at this stage.

5. The increasing application of genomic information raises significant ethical issues surrounding prediction of future morbidity and mortality, and subsequent genetic discrimination. The Council of Europe Convention on Human Rights and Biomedicine[1] explicitly prohibits any form of discrimination on the grounds of genetic heritage and provides that, "tests which are predictive of genetic disease or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes and subject to appropriate genetic counselling". The UK Government has not signed the Convention and so its requirements are not currently binding. The Federation welcomed the Government's Discrimination Law Review and its proposals for a Single Equality Bill. However, we disagree with its view that "there is currently no need to legislate to prohibit discrimination on grounds of genetic predisposition".[2]

Research and Scientific Development

6. In terms of specific technologies, genomic medicine falls into three broad categories: development of new gene-based diagnostics for common conditions; introduction of pharmacogenetics or 'personalised medicine'; creation of novel therapies such as gene therapy and the use of adult/embryonic stem cells to treat disease. Gene-based diagnostics, particularly in the sub-classification of cancers, are developing rapidly through the use of microarray technology. In contrast,

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[1] *Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine*, Council of Europe, 1997. http://conventions.coe.int/treaty/en/treaties/html/164.htm

[2] *Discrimination Law Review: A Framework for Fairness: Proposals for a Single Equality Bill for Great Britain*, Department for Communities and Local Government. http://www.communities.gov.uk/documents/corporate/pdf/325332

pharmacogenomics and gene therapies are at a very early stage, and stem cell therapies are barely off the ground.

7. We believe that it is the research funders that are taking the lead in the consideration and co-ordination of research and the development of new technologies. The Wellcome Trust invests heavily in the genetic research of common diseases and its Case Control Consortium has demonstrated that meaningful data can be obtained from genome-wide association studies. Genetic risk factors for common diseases are now being identified at a pace which was not thought possible a short time ago.

8. UK research is extremely competitive internationally, largely due to the establishment of Research Centres of Excellence and the ease of knowledge transfer within and between research councils and Higher Education Institutions. However, effective coordination of research at a global level is essential to maximise the use of funding streams. The UK's involvement in international partnerships has been backed to a considerable extent through funding from research charities and other non-governmental sources. There is significant scope for the MRC and other public funders to take a more proactive role in supporting international initiatives.

Data Use and Interpretation

9. Existing public genome databases are annotated and presented in a sufficiently useful manner for the majority of researchers. The Federation commends the work of the ENCODE Consortium, the European Bioinformatics Institute and the Wellcome Trust Sanger Institute in the UK in collaboration with the National Centre for Biotechnology Information (NCBI) and the University of California, Santa Cruz in the US, and others to identify and characterise conserved DNA elements, and define gene structures, alternative transcripts and regulatory regions. However, the Federation recognises that much remains to be done to determine the function of DNA sequences identified through this and other research programmes. Such work is currently heavily supported by the Wellcome Trust and should be a priority area for UK public investment.

10. Genomic data should be brought together with other health information through the NHS Research Capacity Programme within the Connecting for Health initiative. However, the Federation does not believe that medical information is currently recorded in a suitable format to allow much more than basic interpretation of genomic data let alone optimal interpretation!

11. We believe that Generation Scotland serves as an excellent model of how patient records can be harnessed in the context of translational medicine, and how large-scale genetics studies could be integrated alongside clinical genetics and associated public engagement activities.

12. We consider that genome data should be afforded the same level of security and privacy as existing medical records provided that the existing security of such information is fit for purpose.

Translation

13. The opportunities for diagnostics, therapeutics and prognostics are potentially great but development will inevitably be demand-led. The requirements to deliver safer and more effective medicines to patients, together with the potential for industry to make better decisions during drug development by the use of genetic information are the main drivers for this approach.

14. The genotyping of participants in clinical trials will bring both benefits and risks. On the one hand, it would be possible to recruit for trials only those patients who, according to their genotype, are likely to benefit from the drug, so that clinical trials could become smaller, cheaper and faster to run. However, such trials would yield information on a smaller segment of the population, with the risk that some side effects would be undetected.

15. The commissioning of the Cooksey report and the subsequent creation of the Office for Strategic Coordination of Health Research (OSCHR) has been a driver for the translational research which will underpin genomic medicine. However the Federation believes that translation to clinical practice is currently driven by industry, the NHS, and the entrepreneurial spirit of individuals.

16. In principle the joined-up nature of the UK health system is an asset to translation and, if properly exploited, is likely to put the UK in a more competitive position than that of the US. The integration of electronic medical records with genomic information will provide a valuable resource for pharmacovigilence studies, currently captured by the yellow card system.

Biomarkers and Epidemiology

17. Although genome-wide association studies are only a point of entry to the identification of biomarkers, they are not dependent upon current biological knowledge but provide a “hypothesis-free” approach to finding novel genetic factors and therapeutic targets to enable patient stratification and aid personalised medicine. Outcomes will need further analysis and replication before becoming informative.

18. Biobanks should be able to link this genetic information with prospective information on environmental exposures etc., increasing understanding of the interplay between genetic and environmental factors in complex diseases. Such information is essential if the dream of 'personalised medicine/medical advice' is to become a reality.

Use of Genomic Information in a Healthcare setting

19. Genomic information will inevitably increase the complexity of disease classification, as is being proven by the sub-classification of cancers, but will allow a sounder understanding of the genotype-phenotype relationship in complex diseases. This will lead to the molecular sub-classification of diseases, each of which may have different therapeutic targets.

20. We are concerned that as no legislation governs the scientific validity and clinical utility of commercial genetic tests sold direct to the public in the UK, this area is wide-open to "quackery".

21. It is essential that genomic information is provided to patients by medical professionals able to communicate the clinical implications and risk, with access to specialist counselling services where appropriate. In particular, it is likely that the provision of genomic and pharmacogenetic tests will in the future be provided at the point of primary care, rather than through specialist genetic services. Guidelines for good clinical practice must be in place, together with Continuing Professional Development (CPD) programmes. We support the recommendations of the 2003 Genetics White Paper for the training of medical professionals in this area and welcome the progress made in the 2008 Genetics Review.[3]

Contact

We should be happy to provide additional information to the Committee. Any queries regarding this response should in the first instance be addressed to Dr Caroline Wallace, Policy Coordinator, Biosciences Federation, 3rd Floor, Peer House, 8-14 Verulam Street, London WC1X 8LZ email:cwallace.bsf@physoc.org.

Taskforce Members

This response was written by a BSF Task Force comprising Dr R Dyer (Biosciences Federation; Chair), Dr J Bradshaw (University of Edinburgh), Dr A Flannery (AstraZeneca), Professor Tony Harmer (University of Edinburgh), and Dr C Wallace (Biosciences Federation).

[3] *Genetics White Paper Review 2008*, Department of Health. http://www.dh.gov.uk/en/Publichealth/Scientificdevelopmentgeneticsandbioethics/Genetics/DH_084147#_1

Appendix

Member Societies of the Biosciences Federation

Association for the Study of Animal Behaviour
Association of the British Pharmaceutical Industry
AstraZeneca
Biochemical Society
Bioscience Network
British Andrology Society
British Association for Psychopharmacology
British Biophysical Society
British Ecological Society
British Lichen Society
British Mycological Society
British Neuroscience Association
British Pharmacological Society
British Phycological Society
British Society of Animal Science
British Society for Developmental Biology
British Society for Immunology
British Society for Matrix Biology
British Society for Medical Mycology
British Society for Neuroendocrinology
British Society for Plant Pathology
British Society for Proteome Research
British Toxicology Society
Experimental Psychology Society
Genetics Society
Heads of University Biological Sciences
Heads of University Centres for Biomedical Science
Institute of Animal Technology
Institute of Biology
Institute of Horticulture
Laboratory Animal Science Association
Linnean Society
Nutrition Society
Physiological Society
Royal Microscopical Society
Royal Society of Chemistry
Society for Applied Microbiology
Society for Endocrinology
Society for Experimental Biology
Society for General Microbiology
Society for Reproduction and Fertility
Universities Bioscience Managers Association
UK Environmental Mutagen Society
Zoological Society of London

Associate Member Societies

BioIndustry Association
Royal Society
Wellcome Trust
Merck, Sharp & Dohme
Medical Research Council
Biotechnology & Biological Sciences Research Council
Association of Medical Research Charities

Additional Societies represented by the Institute of Biology

Anatomical Society of Great Britain & Ireland
Association for Radiation Research
Association of Applied Biologists
Association of Clinical Embryologists
Association of Clinical Microbiologists
Association of Veterinary Teachers and Research Workers
British Association for Cancer Research
British Association for Lung Research
British Association for Tissue Banking
British Crop Production Council
British Inflammation Research Association
British Marine Life Study Society
British Microcirculation Society
British Society for Ecological Medicine
British Society for Research on Ageing
British Society of Soil Science
Fisheries Society of the British Isles
Freshwater Biological Association
Galton Institute
Institute of Trichologists
International Association for Plant Tissue Culture & Biotechnology
International Biodeterioration and Biodegradation Society
International Biometric Society
International Society for Applied Ethology
Marine Biological Association of the UK
Primate Society of Great Britain
PSI - Statisticians in the Pharmaceutical Industry
Royal Entomological Society
Royal Zoological Society of Scotland
Scottish Association for Marine Science
Society for Anaerobic Microbiology
Society for Low Temperature Biology
Society for the Study of Human Biology
Society of Academic & Research Surgery
Society of Cosmetic Scientists
Society of Pharmaceutical Medicine
Universities Federation for Animal Welfare

Additional Societies represented by the Linnean Society

Botanical Society of the British Isles

Systematics Association