



21 April 2008

**British Heart Foundation written evidence to House of Lords Science and Technology Sub-Committee II Inquiry on Genomic Medicine**

**Introduction**

1. The British Heart Foundation (BHF) welcomes the opportunity to submit written evidence to this inquiry. The BHF is the UK's leading research charity in cardiovascular disease and is an active member of the Association of Medical Research Charities (AMRC). Each year we commit some £50-60m to support cardiovascular research in UK universities and health departments.
2. Our research portfolio extends from fundamental laboratory-based molecular, biological and genetic studies to large scale clinical trials of novel and existing preventive and therapeutic interventions. We support research through infrastructure awards for buildings and equipment, project and programme grants for research staff and consumables and, most importantly, research training and career posts for basic and clinical scientists, from PhD students through to Research Professors. Consequently, the BHF has a very legitimate interest in the UK Genomic Medicine environment.

**Questions by the Sub-Committee**

**Policy Framework**

Q1: Who is in charge of setting and reviewing policy in this area?

3. BHF isn't aware of a policy framework regarding Genomic Medicine. Therefore, we don't know who is in charge of setting and reviewing policy in this area.

**Research and Scientific Development**

Q1: What is the state of the science? What new developments are there? What is the rate of change?

Q2: Who is taking the lead in the consideration and co-ordination of research and the development of new technologies?

4. BHF believes that the rate of change is very rapid. The state of the science is right at the beginning of understanding the genome and any susceptibility to disease, as well as any possible responses to medical treatments. We believe that the research community itself should take the lead, particularly the Wellcome Trust.

Q3: How effective is the policy and investment framework in supporting research in this area?

5. BHF isn't aware of any investment framework.

Q4: How does research in the UK compare internationally? How much collaboration is there?

6. We think that research in the UK compares favourably internationally, as there is considerable cooperation of genetic research in the UK, which is the UK's major strength.

Q5: What are the current research priorities?

7. BHF believes that one current research priority is the understanding of the relation of genomic information to disease development, progression and treatment.

#### **Data Use and Interpretation**

Q1: 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?

8. BHF believes that genomic information is generally published in a useful way, however it is not annotated and presented as such. We agree that there should be a common, public database, which should be government funded and linked to similar initiatives in other countries.

Q2: Who should provide the framework for optimal evaluation of data and translational opportunities? What policy and funding mechanisms are in place for recognising and utilising potential opportunities?

9. We believe that medical research funding organisations, such as the Association of Medical Research Charities or MRC are best equipped to provide such a framework.

Q3: 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?

10. Medical information is often not recorded in a suitable format, due to confidentiality issues and difficulties in accessing medical information.

Q4: 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?

11. The potential for abuse is huge. The existing genetic testing methods are not reliable enough to draw any conclusions regarding the possibility of future illness.
12. Further, we believe that it is a concern that people who undergo such testing privately and discover something which causes them concern would then rely on the NHS to provide the support, advice and care which is needed to make the test worthwhile. It should also be noted that private companies may

promote their services to the worried well or more vulnerable groups which could exacerbate inequalities. We are already seeing the abuse of genome data for commercial use through personal testing kits available from companies like 23andMe, DecodeMe and Genetic Health. We would question how the government can assure that such a database is reliable.

13. In regards to the use of genome data in employment, there is no legislation which directly regulates employers' use of genetic tests, unlike in the USA where many states have laws to discriminate genetic discrimination. We believe there is no evidence that employers are currently attempting to carry out routine screening as part of the recruitment process.

#### **Translation**

Q4: How meaningful are genetic tests which use genome variation data? What progress has been made in the regulation of such tests?

14. As explained above, BHF believes that commercially available genetic testing kits are not particularly meaningful for individuals. There is no sufficient regulation of these genetic tests, therefore BHF would welcome better regulation, such as a code of practice relating to genetic testing services.

#### **Biomarkers and Epidemiology**

Q1: In what way do genome-wide association studies contribute to the identification of biomarkers? How is the study of genetic factors and biomarkers integrated for translational purposes?

15. BHF thinks that it is too early to tell in what way genome-wide association studies contribute to the identification of biomarkers. The study is currently not integrated for translational purposes.

#### **Use of genomic information in a healthcare setting**

Q2: 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?

16. We believe that genomic information won't be useful as part of individualised medical advice for a long time (see answer above). There are currently no provisions for ensuring that the individual will be able to understand and manage genomic information. The aspect of use of genomic information is currently unregulated and opportunistic.

Q3: Should there be a regulatory code (mandatory or voluntary) covering the provision of this advice?

17. BHF believes that there should be some guidelines and parameters in place which help doctors and individuals decide how meaningful genetic tests are.

Q4: What are the implications of developments in genomic technologies for the training of medical specialists and other health professionals? Are there any gaps that need addressing? What is the assessment and planning for future needs in capacity?

18. We think there won't be huge implications as future doctors will have a working knowledge of genetics and genomics. However, there are currently gaps of understanding genomic data as it is a specialist area.

**Conclusion**

19. The BHF supports further research in the area of genetics as it could lead to understanding the role of genes and heart disease. Genetic medicine may offer insights into pathogenesis of heart disease and may provide opportunities to develop new treatments and interventions. This potentially offers a new level of "personalised healthcare for all".

20. However, it is only appropriate to think about the usefulness of screening for these genetic variations/defects, once we have fully understood the relevance of genetics. Once we have gained better understanding of disease mechanisms and the ability to identify people at risk before cardiovascular disease manifests itself in the patient, effective genetic screening programmes could be implemented.

**British Heart Foundation**