

Response from the Genetic Interest Group

1. Introduction

The Genetic Interest Group (GIG) is the UK Alliance of approximately 140 patient support groups for those affected by or at risk from all forms of genetic disorders. Our membership includes those groups supporting individuals and families with rare disorders caused by mutations in a single gene, and those with common complex disorders where gene- environment and gene-lifestyle interactions play a role in precipitating the condition in question. GIG's members are supportive of high quality biomedical research and its translation into products and services that address unmet health needs for those currently affected by severe, intractable or incurable disorders. GIG welcomes the establishment of the Lords' Science & Technology Sub-Committee, and is grateful for the opportunity to submit evidence to it. We have concentrated our input on those issues where we feel we have relevant expertise.

2. Policy Framework

2.1 Whilst recognising the often conflicting interests of public and private sector research in the field of genomic medicine, GIG feels that there is a need for greater integration in research activities and programmes undertaken by the academic and clinical communities and those of the private sector. We believe this will create the framework for making best use of limited human and material resources, and secure a focus on addressing significant unmet health needs relating to serious diseases and major health scourges. Closer integration would allow for the creation of mechanisms for effective translation of novel insights into the underlying biology of disease into effective products and services for those who need them. However, in encouraging effective translation of this new knowledge care must be taken to ensure that those with complex, expensive to treat, or rare conditions are not left out in the cold by commercial pressures aimed at securing a return on investment in the short term at the expense of medium to long term gain.

2.2 Greater patient and family involvement in the evaluation of research proposals and their potential to deliver improvements in the circumstances of those at risk would help create an ethical and regulatory framework that not only took account of the potential harms arising from *doing* genomic research, but also of the harms associated with *not doing* it – notably the balance that needs to be struck between individual risk and lost opportunities.

3. Research & Scientific Development

3.1 GIG has no particular expertise on which to base an evaluation of UK science in relation to developments elsewhere. However, while it is clear that our understanding of genomics and of personalised medicine has advanced substantially in recent years, it is also clear that, with some notable exceptions such as Herceptin and the HER2 receptor in breast cancer, we are some way off seeing the widespread clinical applications of this new knowledge. This does not mean that we are sceptical about the potential benefits

that may flow from genomics. Rather it is an expression of our anxiety that the time-scale for investment, and the time-scale for potential return on public sector investment may not coincide with the time-scale for delivery of real clinical benefits for patients. It is important that this promising area of R & D is allowed sufficient time to flourish against a background of stringent peer review to create the impetus for progress and the effective use of resources (whether publicly or privately provided).

3.2 The potential integration of NHS records for clinical and research purposes proposed under the NPFIT programme offers an unparalleled opportunity to create a research resource of global significance. GIG welcomes this potential, but we would stress the importance of respecting the rights of the individual to privacy and a personal opt out from particular uses of their identifiable personal data. There also needs to be a systematic programme of public engagement to explore public concerns about the potential for misuse of personal health and genomic data by government, by industry and by others whose intent may not be in accord with those to whom data and samples relate.

4. Data Use & Interpretation

4.1 Large scale public sector tissue and sample databanks have been bedevilled by a lack of inter-operability on a range of issues that would potentially have permitted the sharing of data and/or samples and resulted in the achieving of the critical mass necessary to address common complex diseases much more quickly. These issues may be administrative – do you record dates of birth in the European (dd/mm/yy) or the US (mm/dd/yy) format for example. They may be technical – are samples held in freezers, dried at room temperature or in paraffin blocks? Or they may relate to laws and governance – such as the terms of the original consent for example. The international P3G Consortium (Public Population Projects in Genomics) is attempting to address some of these issues in order to create conditions for harmonisation and the best possible opportunities for data and sample sharing where appropriate, but there is some way to go before this is achieved, even with respect to publicly funded collections such as UK Biobank and others across the world.

4.2 The position with regard to “personal” collections of samples and data held by academics and clinicians in university and hospital freezers may even more unsatisfactory, although the Human Tissue Act and the Patient Safety Agency have gone a significant way to addressing this. The collections represent an invaluable resource which may currently lie dormant because the project for which they were assembled has ceased. This is, we believe, likely to be particularly the case with respect to the many rare genetic conditions where the uncertainties of the research funding process often result in a stop/start programme, with duplication and repetition wasting resources that could be better used in securing faster progress towards understanding of the disease in question.

4.3 GIG believes there should be a systematic inventory of samples and data collections languishing in university and hospital stores with a view to creating a comprehensive database of what is held where and by who for what purposes. This would re-activate use of many dormant collections, leading to further insights based on continued use of existing resources rather than the need to start from scratch again. Patients and families

would generally support such an initiative, seeing it as a positive use of their commitment to improving understanding of currently intractable diseases through the systematic application of genetic and genomic knowledge.

5. Translation

5.1 Translation of basic research into products and services that benefit patients and families is clearly essential if new knowledge is not to languish unexploited in the pages of the academic literature. Whilst we have no specific expertise to offer with respect to solutions to the problems associated with translation we believe that attention needs to be given to the following issues:

5.1.1 incentives to address disease areas not seen as immediately economically attractive to commercial investors. In particular this would include rare genetic diseases, paediatric formulations of innovative therapies and a sustained approach to addressing technical issues associated with advanced therapies and tissue engineered products (gene and stem cell therapies are but two examples of these);

5.1.2 an appropriate intellectual property framework that balances public, private and citizen interests in the proper exploitation of new knowledge;

5.1.3 a framework for pricing and reimbursement that is not biased against innovative interventions in favour of cheaper (generic but possibly less effective) treatments that do not deliver the desired health gain for patients and families and which subtly discourage sustained investment in cutting edge R & D.

6. Biomarkers & Epidemiology

6.1 GIG sees this as an essentially technical question best addressed by others.

7. Use of genomic information in a healthcare setting

7.1 For patients living with intractable complex diseases potentially able to benefit from advances in genomics, concern is centred on the rate of progress in basic scientific knowledge, its translation into novel interventions and the likelihood that they will be able to access these based on their need; not on where they live, their ability to pay or some other non-clinical issue.

7.2 In addition to the creation of incentives and an IP framework mentioned above, there needs to be a systematic programme of continuing professional education and development. This is critical to ensuring that the NHS is able to deliver state of the art care to patients that reflects current scientific understanding and best clinical practice – wherever they live in the UK and irrespective of administrative boundaries and local commissioning arrangements.

7.3 Care also needs to be taken to ensure that the private health care sector does not undermine the credibility of genomic medicine through the premature marketing of cleared interventions (such as inadequately researched predisposition genetic testing sold over the counter) in ways that might dissuade those with real, addressable health needs from seeking the support they need – either from the NHS or reputable and ethical private sector providers.

7.4 GIG believes that there is a need for an appropriate regulatory framework. This should strike the balance between legal necessity (in relation to the need to prove quality, safety and efficacy for example) and proportionately policed professional, industry based and voluntary codes of practice applied to the promotion of research and the systematic application of its outcomes for improving human health.

8. GIG would be happy to supply further comments in writing or any of the above parts or to extend on them in person to the committee if this would be helpful to its deliberations.

11th April 2008
