

National Genetics Reference Laboratory Manchester

Response to the House of Lords Science and Technology Committee Call for Evidence: Genomic Medicine

The National Genetic Reference Laboratories in Manchester and Wessex were created in 2002 by the Department of Health to promote and take forward development of genetic laboratory science in NHS and to act as a dedicated resource to support molecular and cytogenetic communities. They were re-funded in 2007 for a further five years, with Wessex focusing their work programme on technology development, validation, molecular cytogenetics and reference reagents, and Manchester focusing on informatics, technology assessment and quality assurance. Both NGRLs are embedded in NHS service laboratories and each have a dedicated staff of 5-6 scientists, technicians and informaticians. The comments below are from and on behalf of NGRL Manchester, and derive from their particular areas of expertise.

Data Use and Interpretation

- *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?*

There are many potential users and uses of genomic information, as well as many providers and resources. Projects like Gen2Phen (Genotype-to-phenotype databases: a holistic solution, www.gen2phen.org), a recently funded EU Framework 7 project led by the University of Leicester, would contend that genomic data are not presented usefully: they are not coordinated in terms of entering, storing, integrating and searching for data. Gen2Phen aims to develop tools, resources and standards to address these issues. Common public databases within the UK are perhaps not the answer as the design, scope and abilities of different databases are so varied, and there are already many initiatives to collect and publish data. Databases also tend to be international and this becomes more important the rarer a disease is. One difficulty that database projects have is establishing the funding mechanisms to develop and maintain their resources: methods include grant funding, sponsorship and subscription. For medicine to rely on these resources they must be viable in the long term, and the nature of many internet-based resources means that the user is completely reliant on the supplier maintaining the service: any break in funding for the service provider can be catastrophic for the users who have little or no fall-back position. Although the applications may be used internationally, when they are grant funded this is likely to be at a national level, so users in different countries may have limited say in policy decisions on their continued funding.

If the NHS relies upon internet-based resources it must therefore consider how it can ensure that they remain available. This could include making contributions of e.g. funds, data and expertise, or becoming a stakeholder in the provision and management of key resources. Another alternative, or a parallel action, would be to maintain internet servers for NHS users with 'mirror' implementations of important resources, and archives of previous data sets.

- *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?*

An issue here is with the coding and classification of phenotype data. As the National Programme for IT has developed, genetics has considered some of these issues (e.g. during the Do Once and Share project for Clinical Genetics, http://www.bshg.org.uk/documents/official_docs/DOAS_final_printed_report%5B1%5D.pdf, and the development of the Output Based Specification for NHS Genetics Service Information Systems, <http://www.ngrl.org.uk/Manchester/Downloads/OBS.pdf>) and found that clinical centres all use their own classification of diseases and that mandated coding standards like Snomed-CT and HL7 require, and are in the process of, international development in order to be applicable. Elsewhere the Rare Disease Task Force, led by Orphanet in Paris, are leading projects which are concerned with rare genetic diseases, including input to the ICD-11 development. This recognises that ICD-10 does not currently provide sufficient detail and accuracy for unambiguous classification of inherited diseases. Coding standards need to be in place for genomic medicine to converse fully with the NHS medical systems that are being developed, and thereby avoid being an isolated speciality. An issue that must be addressed is the representation of family data which is not possible within the current NPfIT systems which assume treatment of individuals.

A coordinated effort to bring together initiatives in Connecting for Health, SNOMED-CT, HL7 and other coding systems and projects with the genetics community, supported by informatics expertise, is a goal that NGRL Manchester has identified and is working towards.

21 April 2008