



## National Genetics Reference Laboratory (Wessex)

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

#### Background

1. 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 from NGRL(W) derive from our particular areas of expertise.

#### Policy framework

2. Genetic tests in the UK are currently performed primarily by a network of 23 regional genetics laboratories. Many of these laboratories will be in a strong position to implement genomic tests once these are developed, and indeed many are already undertaking such tests, e.g. microarray comparative genomic hybridisation. However this configuration may not be optimal for all aspects of testing associated with genomic medicine, e.g. if there are greatly increased numbers of tests as a consequence of new screening programmes. It will be important that any new configuration maintains strong links with clinical and research communities as well as strengthening links with other pathology disciplines as part of new care pathways.

3. The rapid pace of technological developments means that laboratory equipment is rapidly outdated. Additional laboratory capital investment will be needed to realise the benefits of genomic medicine, and consideration should be made for this to be available on a recurrent basis.

4. Much of the recent attention has focused on the spectacular successes of genome wide association studies (lead principally by UK researchers) in identifying common, inherited low penetrance susceptibility alleles for a variety of conditions. It should be borne in mind however that many aspects of genomic medicine involve acquired genetic or genomic changes in cancer. An integrated approach to inherited and acquired changes should be considered that includes mechanisms of funding/commissioning of tests.

## **Research and scientific development**

5. Although the UK leads the world in many aspects of basic research, there is a significant gap in funding for development that is not currently being met by the NIHR and HTA programmes, which focus largely on clinical utility and validity of diagnostic tests. Translating research findings into robust, quality assured, fit-for purpose laboratory assays is often a lengthy and costly process that needs to be specifically funded.

6. There is a need for detailed translational research programmes to establish the benefits of new scientific findings in the context of genomic medicine.

## **Data use and interpretation**

7. Emerging technologies will require much more sophisticated information technology to manage and interpret data. The infrastructure needs, access requirements and data volumes are very different to standard NHS requirements and will require dedicated IT staff embedded in testing laboratories as well as capital investment.