

30 April 2008

## **Response to the Call for Evidence on Genomic Medicine from the House of Lords Science and Technology Sub-Committee**

In Sheffield the Genetic services are managed and delivered within the Diagnostics Directorate as part of the portfolio of specialist services. They include Cytogenetics, Clinical Genetics, Molecular Genetics, Biochemical Genetics and Newborn Screening.

There are many issues which have already been appropriately covered by the response from the JCMG. However there are some points which may benefit from further clarification or where we significantly differ in our response.

- 1. Configuration of Laboratory Services.** The existence of 23 regional laboratories can no longer be justified on past successes and existing service configurations but must be based on future potential and best value for money. For example the expansion of genetics into mainstream medicine will increase and broaden the strategic interactions of laboratory and clinician, but the introduction of robust electronic communication will facilitate rationalisation and remote laboratories. This complex issue has begun to be addressed by the UKGTN commissioning group regarding the impact of new technologies but will inevitably have to consider wider issues of the services delivered on those technologies and the potential for future development. It is clear that thought must be given to the configuration required now but also in 10-20 years time as too much rationalisation will promote *ad hoc* local development, whilst too little will fail to deliver a cost effective service which competes well with the private sector.
- 2. Future commissioning models.** With the developing NSCG and with enhancement of the role of the UKGTN, there is a real potential for intelligent commissioning: Further development and strengthening of these structures will inform and support Specialist Commissioning at an SHA level. It will be important however that in the near future the commissioning structure is reviewed as genetics becomes more mainstream. There needs to be consideration if there is a point at which, and in what areas, laboratory genetics may no longer be part of specialist provision. Would PCTs be commissioning some types of genetic testing and if so how will this be informed and delivered?
- 3. Training and education in Genetics for Healthcare Scientists and other healthcare professionals.** Modernising Scientific Careers will clearly broaden the understanding of scientists in healthcare and this will ensure that genetics training will be more widely available. This recognises and supports the expanding role of genetics in many disciplines and the need integrate genetic technologies in a cost effective manner into many disciplines. Training and education will be key but also will place significant demands on those who deliver training and education in genetics. This must be addressed. However it will be beneficial to those finally specialising in the genetic disciplines to have a better understanding of other disciplines, enhancing and improving the services in all areas.
- 4. IT initiatives.** There are specific IT requirements that are particularly relevant to Genetics but overall there is a significant need for greater capacity for storage and retrieval of information, faster transfer of information and far better integration. To date the IT agenda has been dominated by poor performance and delivery by the IT

sector which has drastically reduced expectation, often to the point well below what the IT systems can deliver. It may be an appropriate time to start developing what the NHS systems need for the future and making demands on the IT companies for the systems of the future.

In addition to these general points there are some more specific points that have in part been channelled through the JCMG

#### Biochemical Genetics and Newborn Screening

- Enzymic and metabolite assays provide direct genetic information leading to genetic counselling and pre-natal diagnosis in many patients. Indeed, newborn screening for conditions such as phenylketonuria and sickle cell disease depend exclusively on these techniques while disorders such as medium chain acyl CoA dehydrogenase deficiency and cystic fibrosis are primarily identified and often confirmed by these approaches. This means that these diagnostic approaches are overwhelmingly the predominant means of genetic testing in the UK at present with in excess of 3.0 m tests performed annually. The importance of this means of providing genetic information is reflected in the inclusion of metabolic testing for inherited disorders and newborn screening within Specialist Medical Definition set 20 but investment is severely lacking.
- It is important for the patient that these varying approaches are integrated and co-ordinated to provide a seamless service and set of standards for the user. This is not the case at the moment, as an example differing turnaround times requirements apply and are addressed under differing initiatives eg “the six week diagnostic wait” does not apply for some forms of genetic testing.
- Training initiatives and funding for technological development are primarily aimed at molecular, cytogenetic and clinical genetics while workforce planning and training for screening remains unfounded within these initiatives. Some limited but valuable funds are available for biochemical genetics. This may be addressed within Modernising Scientific Careers but it needs to be clearly included in the discussion at the discipline-specific stage of planning.
- Direct support for the exploitation of new technologies such as Tandem Mass Spectrometry and their implementation into practice could significantly extend the range of conditions accessible to newborn screening. This approach is mandated in the USA and has been adopted by many countries in Western Europe, the UK now lags significantly behind developed and developing countries in this regard.
- High throughput whole population screening demands carefully designed IT support and an integrated IT system linking Child Health Records Departments, Maternity Units and Screening Laboratories. This is long overdue. The lack of investment of IT in this area affects both the safety and efficiency of these services.
- Many vulnerable services such as specialist diagnostic service dependent on enzyme assay are poorly resourced and there is little resilience in the system with the real risk that new developments are unavailable in the UK and some existing services may be under threat. There is a real opportunity to develop these services both to serve the NHS and to pioneer healthcare technological innovation as a means of income generation from overseas.