

**House of Lords Science and Technology Committee
Call for evidence: Genomic medicine**

Response from: Association for Clinical Cytogenetics (ACC)

The ACC is the professional body representing Clinical Cytogenetics.

Background - Introduction

Cytogenetics is the study of the whole genome for chromosomal abnormalities their aetiology and risks; and as such, investigates general phenotypic abnormalities such as mental retardation and developmental delay, dysmorphism, infertility and reproductive problems and a spectrum of congenital abnormalities spanning the range of organ systems such as heart, neuro-muscular, etc. Referrals come from all age groups: prenatal diagnosis, children, adolescents and adults. Cytogenetics investigates the presence of chromosomal abnormalities that are constitutional (germ-line) and acquired (somatic in cancer) and provides both diagnosis and prognosis. Investigations include examination of the whole genome by microscopic analysis of the chromosomes, together with complementary molecular cytogenetic analysis targeting explicit syndrome related chromosome abnormalities or cancer markers at the submicroscopic level. Whilst this methodology has been employed for decades, the recent development of whole genome cytogenetic analysis by microarray technology, which gives very high-resolution sub microscopic resolution, is starting to radically change laboratory practice.

Response

The ACC endorses the response from the Joint Committee on Medical Genetics and would like to emphasise the following important issues:

Policy Framework

The rapid increase in knowledge, technology and consequent capability means it is not possible to predict and plan accurately for the longer term. However, current scientific awareness does allow a reasonable prediction of trends.

The infrastructure of specialist regional genetics centres and the culture of close collaborations between clinicians, laboratories and centres has supported effective genetic service development and delivery and must be maintained. Benefits would be gained by improved connectivity for bioinformatics and IT.

The UK is one of the leaders in both genetics R&D and service. The recognition and investment from the government via the White Paper 'Our Inheritance, Our Future' ensured this position was maintained.

NHS processes can be protracted and care must be taken to ensure timely implementation of improvements to service, which are implemented and funded in an equitable manner across the UK.

The NHS Genetics Team (England) and GenCAG are involved with policymaking. Information and recommendations are obtained from various genetics groups such as UKGTN, JCMG and professional bodies including ACC together with input from academic/clinical experts either independently or as part of an expert working group. These advisory groups can be specialist and it is increasingly important to ensure that feedback is evaluated in a broad strategic context and incorporated into a 'joined up' national genetics strategy.

As genetics involves the wider family, commonality across UK regions and countries is preferable.

European (or wider) involvement currently relies on individuals identifying and joining appropriate committees and networks. This area needs to be strengthened so there is structured and comprehensive involvement of a national position.

Research and Scientific Development

We value the government's recognition of genetics in medicine and support via the genetics knowledge parks and reference laboratories. We also support the establishment of external oversight groups for these bodies to ensure appropriately targeted work plans and effective strategies for timely dissemination of outcomes to inform UK service laboratories. Also these oversight groups should be able to provide independent external advice to DH

The fast pace of change means there is a need for faster validation and effective feedback of findings to the diagnostic community from designated R&D groups.

We support the proposal from the Royal College of Pathologists that the government and DH should give greater priority to the evaluation of diagnostic tests; to evaluate the expected benefits of new tests for patients vs. cost, and make recommendations for the funding of cost-effective new tests in an equitable manner across the UK.

We emphasise the point made by the Joint Committee for Medical Genetics, that the NHS Research and Development funding, which is coordinated by the National Institute of Health Research, excludes all proposals linked to laboratory medicine. And other sources are not addressing this funding gap. This is a weakness within the current system that needs to be rectified.

Data Use and Interpretation

We are in an era of producing vast quantities of genetic data on patients.

Genetics IT systems should support:

- Managed collation of UK data to increase specific datasets (eg normal variation within our ethnically diverse population) and hence assist with interpretation – there should be mechanisms to ensure these datasets are quality managed.
- Appropriate networking across genetics and with NHS patient administration systems
- Suitable storage of data, since genetics problems span generations

Data access and use must be controlled to ensure confidentiality of what are very sensitive data. There must be clarity as the purpose of these data, in that they are obtained with informed consent to benefit the health of patients and their families; and hence are not available to other parties for commercial or potentially disadvantageous applications (e.g. assessing insurance risks).

Translation

New technologies such as cytogenetic microarray analysis are already proving themselves with an increase in abnormality detection and improved diagnosis. However, this technology and the new high-throughput DNA sequencing technology, are also identifying non-pathogenic genetic imbalance and variability. The pace of clinical integration will not be limited by the rate of technology development, but by our ability to accurately interpret the resulting data and recognise normal variations, and potentially false positive results.

The White Paper investment has meant that UK Genetic Centres are well-equipped for cytogenetic microarray analysis, and high-throughput molecular genetic analysis, and as such can become kernels of equipment and expertise around which facilities for the wider applications of genomic medicine within Pathology could be developed in collaboration with these other disciplines.

Information is being steadily accrued in various international internet databases. Collation of quality controlled UK data into a NHS database may assist in reviewing commonality and phenotype and allow patterns of significance to be seen.

The use of Imatinib in chronic myeloid leukaemia illustrates the value of identifying the genetic basis of disease and the development of targeted drug therapy. This link between genetics and therapeutics will increase in the future.

Diagnostic clinical scientists employed in UK NHS laboratories are ideally placed for translation into service as they have the knowledge and skills pertinent to both the science and the clinical setting. Experience of diagnostics, including the strict quality requirements, and clinical applications ensures the development work is focused and appropriate. However, funding for this work is unclear.

The pace of change for both technology and knowledge in genetics is extremely fast. Care must be exercised in strategic planning to ensure investment is in longer term, high impact technologies rather than short term, new methodologies.

Use of genomic information in a healthcare setting

To date, the clinical handling of genomic information has been limited to clinical geneticists and other specialists. As there is increasing transfer of genetic testing into mainstream medicine, there needs to be an increasing programme of genetics education for a broader spectrum of clinicians and appropriate support staff.

Current clinician and laboratory regulation ensures appropriate handling of genetic information. Regulation, including validation of genetics training, should also ensure appropriate handling by the wider medical community.

Regulatory standards must be applied to both NHS and private sectors.

Testing must be led by clinical need and not influenced by technological capability.

The public will need a greater education of genetic testing and the reassurance of a safe and controlled use of their genetic information as a key part of the roll-out of genetic testing across medicine.