

Clinical Immunology and Allergy Section

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

Clinical Immunology is a rapidly advancing specialty with both clinical and laboratory diagnostic components. Clinically the main groups of patients cared for by Clinical Immunologists are those with primary immunodeficiency (i.e. genetic defects of the immune system rendering the patient susceptible to infection of which there are now around 200), patients with allergic disorders where Immunologists provide over half of the specialist services for the UK and patients with inflammatory or immune mediated disorders.

The Clinical Immunology laboratory provides diagnostic testing across a range of diseases including primary (genetic) and secondary immunodeficiency (e.g. HIV), allergy testing, connective tissue diseases (e.g. rheumatoid arthritis, SLE, vasculitis) and others for NHS patients.

The diagnostic laboratory has an increasing need to perform molecular diagnosis for primary immunodeficiency patients. This allows accurate diagnosis with implications for family testing, detection of carrier status, prenatal testing, genetic counselling as well as informing treatment decisions such as bone marrow transplantation and gene therapy. Currently only a very small number of genetic tests for primary immunodeficiency are available within UK-GTN and this has not kept pace with the rapid expansion in knowledge regarding the genetic causes of primary immunodeficiencies.

Clinical Immunology R&D also involves the use of Immunodeficiency micro arrays allowing the expression of a focussed gene set to be studied to aid the molecular characterisation of disease states in immunodeficiency. It may in future be possible to use micro arrays to not only improve the precision of molecular classification of disease states but also to aid more personalised clinical management decisions and to suggest new treatment strategies.

There are as yet no NHS Clinical Immunology laboratories which are able to address issues relating to the analysis of complex diseases such as allergy.

Policy Framework

The Government's 2003 White Paper *Our Inheritance our Future* has strengthened specialist genetic services with the UK Genetic Testing Network (UK-GTN) and modernised laboratory services with a round of capital funding.

There is however no cohesive support which allows the translation of research based genetic tests into clinical practice.

In Clinical Immunology initiatives may be funded by patient organisations and even individual patients, there is then a gap in provision to allow these developments to progress. This results in tests which are either not available at all in the UK (the case for the majority of known genes for primary immunodeficiency) or offered in a research setting outside an accredited laboratory environment with variable turnaround times, lack of stability and planning as testing depends on the continuation of research funding, staff and research direction/interest.

There is an opportunity to establish molecular diagnosis for a much more comprehensive range of immunodeficiency diseases which are widely under-diagnosed even in 'first world' settings. Given the lack of testing within an accredited environment the potential exists to derive income from referral for testing from outside the UK.

Recommendations

- NHS R&D and NIHR support the evaluation and when appropriate translation of individual genetic tests in the NHS and that this is linked to pathology modernisation. The current NHS culture that if a test cannot be shown to save money in the business case it cannot be introduced requires review.
- Training in bioinformatics will be required for clinicians involved in the interpretation of complex datasets such as those generated by micro array analysis and in the reporting of molecular tests.
- The establishment of a number of networked centres with both the Clinical Immunology and molecular expertise will be required to provide comprehensive testing.
- Realisation of the benefits of genomic medicine will require public trust and this will in turn require greater engagement of the public by government.
- The molecular genetic information generated should be available in a standardised coded manner within the stable platform of a secure NHS database and shared appropriately with other networked centres.
- All of the above will require targeted stable long term investment.

British Society for Immunology
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