

HOUSE OF LORDS SCIENCE AND TECHNOLOGY COMMITTEE

Genomic Medicine

Evidence for the Inquiry to provide an assessment of Genome Technologies and their actual and potential impact on clinical practice in the post-genome era.

Submission by: London Specialised Commissioning Group (organisation submission)

Date: Monday 21 April 2008

NHS COMMISSIONING PERSPECTIVE

Background

1. Genetic services are defined as a specialised service in the National Definition Set, set out by the Department of Health. Commissioning arrangements for specialised services are overseen by the National Specialised Commissioning Group. There are ten Specialised Commissioning Groups (SCGs) in England and their responsibilities are defined in the 'Review of Commissioning Arrangements for Specialised Services – May 2006', Chaired by Lord Warner.
2. This submission is from the London SCG in association with East of England SCG and South East Coast SCG that is responsible for commissioning (planning, procurement, quality assurance and evaluation) for NHS services.

Policy Framework

3. The UK Genetic Testing Network (UKGTN) influences policy by evaluating new tests. NHS commissioners are made aware of new tests available through the UKGTN processes which inform commissioners of indicative new funding levels required as developments occur. This enables commissioners to agree appropriate funding levels within the commissioning planning cycle. The UKGTN evaluation process includes testing criteria which promotes appropriate referrals. This reassures commissioners that testing will be carried out only when there is clinical need and clinical base.
4. Scientific advice to assist on policy development is provided by the national genetic reference laboratories who are members of the UKGTN.
5. Commissioners do not directly fund developments to translate research into NHS mainstream services. Commissioners would expect laboratories to provide a business case for their own Trust management to support and secure development funding. However, some laboratories have difficulty in securing funding when competing against other service priorities.
6. The National Institute of Clinical Excellence (NICE) provides technical appraisal, clinical guidance and procedures for commissioners although they tend not to look at diagnostics, but the treatment element. However exceptions are made when there is a political imperative, for example, breast screening. In the main, new

tests are not evaluated by NICE. The UKGTN takes on this role for genetic tests for inherited disorders where nucleic acid is the analyte.

7. Laboratories need to be clinically appropriate working within quality assurance scheme like NEQAS and CPA accreditation.
8. Commissioners are not always aware of what is happening in research until developments are ready to become a mainstream service. For example, the Sanger Centre is developing a national database for microarray testing using the three laboratories to undertake the testing and provide results. The consequential implications of large scale and high throughput test provision are not clear to either service providers or commissioners and will require careful monitoring.
9. Social, ethical and legal consideration is taken into consideration when setting policy. The Human Genetics Commission is the UK's government advisory body on new development in human genetics and the impact on individual lives.
10. No other country has a similar system as the UKGTN gene dossier process for introducing new genetic testing. In the USA there is a more open approach where the public can easily access testing. For example, the USA offer open access for paternity testing but, rightly so, this is not available within the NHS.
11. Discussions are currently taking place in Europe concerning the IV Directive and the possibility that genetic tests may move from category one to category two products. Category one products are self regulated whereas category two products need to be passed by a formal regulator. This could have an impact on the tests that are developed in-house. The implication is that some laboratories may need to use commercially available kits which are likely to be more expensive and may not provide the exact testing requirements. This will have an impact on costs for commissioners.
12. NHS policy recognising the important role of the commissioner as set out in the aims of World Class Commissioning. This commissioning perspective should be seen in this context and we would encourage the review to take the opinions from commissioners as well as academic, clinicians, scientists and patients. It is important to ensure that services continue to develop based on evidence in an appropriate way. In order for this to be achieved commissioners need to be informed to ensure the resources are planned for the services required.

Research and Scientific Development

13. There is investment in research but the rate of change for developments is slow because of the links with the NHS and the availability of mainstream funding. Focus in the NHS is on common diseases because of critical mass however we also do need to consider the implications because genetic conditions will be for rarer conditions within each specialty.
14. The national reference laboratories have a role in developing new technologies but the pace is slow and as a consequence much of the work is repeated in local laboratories. Foundation Trusts see this as an opportunity to develop services. For example, Guy's & St Thomas' were able to access development monies to develop rapid QFPCR screening that reduced costs for commissioners.
15. Financial support is usually through charitable grants or the Medical Research Council. Research is usually taken forward by interested parties as there does not

appear to be a policy framework to support research. There is no mechanism for commissioners to measure capacity in the system or to identify value for money other than initiatives led by commissioners.

16. Commissioners are aware that laboratories are developing a whole sequencing process for testing but the implications for this are not clear. It is apparent that there will be a need to assess integration and reconfiguration of laboratories both between additional molecular and cytogenetic providers and their relationship to additional pathology services as indicated within Pathology Modernisation. The NHS needs to be responsive and promote efficiency by reconfiguration.

17. Commissioners have supported robotic systems in laboratories to improve throughput. The time limiting step now appears to be in the interpretation of the assay.

Data Use and Interpretation

18. Technology is far more advanced than the software to interpret results which impacts on the time that patient's receiving their results.

19. Genetic recording systems (often bespoke) hold personal family data with no way of cascading family information. Family data is not part of connecting for health and the diagnostic element and results (positive/negative) is not adequately recorded on systems. There is insufficient coding and classification for the rare diseases and as a consequence the NHS is not capturing the valuable data source about these conditions.

20. The need for mainstream specialties to access genetic tests is growing and needs careful consideration as part of a care pathway in order to conform with the 18 week referral to treatment policy. We need to be clear about the implications for genetic services as the referral pathways may lead to the main specialty dealing with the rarer conditions. Already we are having discussions about the design of a care pathway for familial cancer to ensure that patients and those at high and moderate risk are accessing breast screening services in a timely manner.

21. There are storage issues about holding confidential family information when consent is based on an individual's agreement. Genetics is an NHS service benefiting families rather than individuals and because of the complexities, the British Society of Human Genetics (BSHG) have produced guidance on consent and confidentiality on genetic testing and the sharing of genetic information.

22. Families may choose not to take up testing when offered because of concerns about this information having to be declared.

Translation

23. It is clear that more tests are going to be requested as more genetic conditions are identified in mainstream specialties and these need to be part of an agreed clinical pathway. There are concerns that mainstream specialties do not always consider the extended family and these needs to be taken account of in the future.

24. It is difficult to identify who is responsible for translation to clinical practice. Arrangements are informal and usually developed through the interest of individuals or influenced by patient groups.

25. There is an issue about the fast moving pace in genetics. The Genetics White Paper provided an opportunity for laboratories to update their services but if this is left to individual organisations the pace would be slower and moving at a different rate according to individual organisation's priorities. These impacts on test turnaround times and efficiencies.

26. As far as we are aware there is no regulation on 'over the counter' testing and there is little evidence-based information to support undertaking these tests. People who purchase 'over the counter' genetic tests may not fully understand the result provided due to the company not providing counselling or fully explaining the results. This may lead to unnecessary anxiety and patients entering the NHS system in order to have their results fully explained.

27. In South East England, commissioners have set up a clinical network inclusive of academic leaders:
Imperial College
King's College London School of Medicine
Institute of Child Health/Institute of Neurology/University College London
Queen Mary's College

28. The purpose of this is for commissioners to be informed about developments and their implications on NHS services.

Biomarkers and Epidemiology

29. As the NHS systems are not capturing data it is unable to predict incidence and prevalence of the population. The limited data that is collected does not record numbers of positive/negative tests by disease and there is no benchmarking data to monitor this activity against. Some testing is not commissioned through genetic clinical services but through local specialties, such as paediatrics and is often not separately identifiable.

Use of Genomic Information in a Healthcare Setting

30. The NHS National Genetics Education and Development Centre is working with a range of groups throughout the UK to facilitate the integration of genetics education into all levels of education and training for all NHS health professionals.

31. Regulatory code should be developed with the Royal Colleges so that specialties understand the implications of ordering genetic tests, including consent issues, care pathways and outcomes.