NHS

UK Genetic Testing Network

UK GENETIC TESTING NETWORK STEERING GROUP
FOURTH MEETING, 1 APRIL 2003
KING'S FUND, LONDON

Present:

Peter Farndon (Chairman), *Clinical genetics, GenCAG*
Naomi Brecker, *NHS Genetics, DoH*
Anne Green, *Biochemical Genetics Network (Observer)*
Tony Cumming, *UK Haemophilia Centres Doctors Organisation (Observer)*
John Barber, *Cytogenetics, National Genetics Reference Laboratory - Wessex*
Jo Whittaker, *Molecular Genetics, CMGS*
Rob Elles, *National Genetics Reference Laboratory – Manchester*
Kendal Bird, *Commissioner, GenCAG*
Ron Zimmern, *Public Health Genetics Unit, GenCAG*
Louise Newport, *NHS Genetic, DoH*
Dianne Kennard, *NHS Genetics, DoH*
Robin Winter, *Clinical Genetics, JCMG*
David Baty, *Scotland*
Rachel Butler, *Wales*
Reg Gerrar, *Genetics Branch, DoH*
Maureen Boxer, *Lead Scientist, GTN Team*
Jacquie Westwood, *Project Director, GTN Team*
Cressida Miller, *Laboratory Co-ordinator, GTN Team*
Jane Deller, *Commissioning Co-ordinator, GTN Team*

Apologies: Alastair Kent, *Patient Interest, GenCAG,* Fiona Stewart, *Northern Ireland,* Ann Curtis, *Molecular Genetics, CMGS*

In attendance: Liz Woodeson, *Head of Genetics, Embryology & Assisted Conception Branch, DoH*

1. Welcome and introductions

1.1 The Chairman opened the meeting by welcoming the presence of Liz Woodeson and the return of Louise Newport (Senior Scientist) to the group.

2. Minutes of the last meeting

2.1 Minutes were agreed in principle, with some minor amendments to be forwarded by the chairman. Subject to these minor alterations the minutes will be published on the Department of Health website (http://www.doh.gov.uk/genetics/gtn.htm).

3. Matters arising from the minutes

3.1 There were no matters arising from the last minutes that were not on the agenda.

4. Network updates

4.1 National Metabolic Biochemistry Network

A verbal report was presented that an interactive website for the network is currently under development and training workshops for it's use. Best practice guidelines are also being developed. A questionnaire to determine the current level of service provision for biochemical genetics is due to be carried out in May and a workshop will be held in October 2003 to look at bringing together the different External Quality Assurance schemes (EQA) and identify any gaps in provision. Issues were raised concerning clinical scientist training posts in biochemical genetics. It was agreed that the UK GTN Steering Group chairman would compose a letter to the Chief Scientist at the DOH to highlight these issues.

4.2 The Haemophilia Genetics Laboratory Network

A verbal report was presented which highlighted that network participants had met three times since November 2002, that a specific request form is being developed and that issues surrounding patient consent and confidentiality and the storage of DNA samples are being discussed and a patient consent form is being developed. The Work Load Units, developed by the Clinical Molecular Genetics Society (CMGS), will be piloted for three months by network participants and then reviewed. A meeting will be held in May to update haemophilia genetic guidelines and to look at developing links with the EQA. The UK Haemophilia Centre Directors Organisation has recently carried out audits of their own centres. However, these audits did not include regional genetic laboratories and therefore it is planned that they will do so in the future.

5. Work Programme

5.1 The revised Outputs to the UK GTN Steering Group Terms of Reference were endorsed and will be published on the DoH GTN website.

6. Education and Training Programme

6.1 GenCAG paper 'Education and training in genetics for commissioners and managers of NHS services' was enclosed for information. Proposed Terms of Reference for the delivery of an Education and Training Programme for commissioners and managers were debated. It was agreed that this paper would be expanded to show an outline of the content of the workshop once it has been agreed. Subsequently this paper will then be placed on the DoH GTN website.

6.2 The South Directorate of Health and Social Care has agreed to be the pilot site for the education programme, with a workshop in the South West of England in early July. A meeting with the co-ordinating team and local champions from the South is planned for 7 April to discuss the format of the workshop, to agree local speakers and to determine the target audience. Further workshops are planned in the Midlands and Eastern DHSC (September 2003), North DHSC (November 2003) and London (February 2004). A meeting for the co-ordinating group to discuss the whole programme in more depth is arranged for late May. It was stressed that the 4 workshops are intended to be the first of an ongoing programme and evaluation of year one will occur in March 2004.

6.3 After evaluation of the programme, the co-ordinating group will liase with relevant parties in Wales, Scotland and Northern Ireland to share lessons learnt from the programme.

7. Referral Criteria for the reference laboratories

7.1 Background information to 'The Very Rare Disease Testing Service'

Provision of a clinical diagnostic service for very rare inherited diseases was identified by the DoH as being a responsibility of the National Genetics Reference Laboratories (NGRL). In connection with this the NGRL propose to set up a testing service for extended family members of patients who have had a genetic mutation detected in a research laboratory and for which there is no testing service available on the NHS. The number of index cases that will be handled in the first year has been restricted to 25 due to limited resources. Members of the Steering Group voiced a concern that restricting the service to only 25 index patients was under-estimating the real demand for such testing. Consequently it was agreed that the Lead Scientist (of the GTN executive team) would identify the current number of requests for this type of service from NHS laboratories and report back to the Steering Group in due course. In addition, the Lead Scientist will also investigate the number of requests for a mutation scanning service of ultra rare diseases, which is presently not available on the NHS. The rare disease testing service will be audited one year after its implementation.

A paper entitled 'Rare Disease Testing Service' which provides full details of the proposed service was circulated to members of the Steering Group. This paper was endorsed with the proviso that a section on clinical utility should be added and that reference should be made to the gene dossier.

7.2 Application forms for the 'Very Rare Disease Testing Service'

The application forms, which will be available in electronic format via the NGRL website (online in May 2003), were endorsed. It was agreed that all referrals should be through a Clinical Geneticist based in a UK Regional Genetics Centre.

8. Process for setting up the UK GTN

8.1 Process for laboratories to join.

Minor amendments were made to the revised paper that sets out the process for laboratories to join the UK GTN. The Lead Scientist agreed to present, to the next Steering Group meeting in July, any problems encountered in the assessment of laboratories for participation in the network.

The criteria that laboratories will need to work towards in order to participate in the network were clarified as the quality criteria that are set out in the "Framework for Delivering the UK Genetic Testing Network" paper.

8.2 Laboratory Application Pack

The laboratory application pack will compose of an application proforma, a summary of the paper "Framework for Delivering the UK Genetic Testing Network" and a covering letter. These 3 documents were drafted for members to approve.

Application Proforma

Amendments to the draft application proforma, inviting laboratories to be providers on the GTN, were suggested. It was agreed that the Chairman would endorse the amendments.

Summary of the Framework for Delivering the UK Genetic Testing Network

A summary of the "Framework for Delivering the UK Genetic Testing Network" was prepared as part of the application pack. It was endorsed on condition that some minor amendments will be incorporated into this document.

Letters to laboratories

A draft letter inviting laboratories to make an application to participate on the UK GTN and a draft letter inviting laboratories to express an interest in applying to become participants at a later date were circulated for comment by the Steering Group Members.

It was agreed that laboratories that do not currently fulfil all the quality criteria would be considered to join the network on condition that they have plans in place to work towards the expected standards in an agreed timescale. It was agreed that the letters should be amended to highlight this point. It was agreed that the intention of the application proforma is to determine current activities rather than an ideal that laboratories may be aspiring towards.

8.3 Commissioning.

An example of the letter and proforma that was sent to the commissioning representatives on GenCAG, requesting relevant commissioner contact details, was presented for information. Members were advised that some of the proformas had already been returned. A UK wide directory of commissioner contacts will be compiled following the submissions being returned.

Communication

8.4 Project Plan.

The mechanisms for delivering the UKGTN should be seen as enhancing the existing processes for current genetic services. Focus is on delivering key targets set out in the NHS plan and ministerial commitments, including meeting patient needs, improving patient access, enhancing the quality of services and providing good value within available resources. The emphasis is to build capacity within a whole systems service approach in a collaborative manner.

A preliminary project plan showing the year 1 work plan for the UK GTN executive team was presented. Members requested that the project plan be made available to the rest of the genetics community. However, it was decided that it should first be seen by GenCAG members (at their next meeting on 8 May) and by members of the Friends of SCGs before it is made widely available.

8.5 DoH website and future arrangements

The Steering Group was informed that a GTN page has been set up on the DoH website (www.doh.gov.uk/genetics/gtn.htm). It documents the minutes of the first two GTN Steering Group meetings, a list of GTN Steering Group Members and the GTN Steering Group Terms of Reference. In the future an independent GTN website will be developed. Initial proposals have been gathered from a number of development companies in order to set up a UKGTN website. It was agreed that an evaluation panel should be set up to commission the company who will develop the website. The Evaluation panel should include the Lead Scientist, a DoH representative and an IT expert.

9. Framework for Delivering the UK Genetic Testing Network

9.1 *Background*

A paper is being drawn up to provide a statement that draws together all the strategic approaches that will drive the delivery for the UKGTN. The paper has been called "Framework for delivering the UK GTN" and is near completion. The Steering Group members discussed content of the section that will detail the process for evaluating and prioritising network tests. It was noted that when requesting NHS funding for tests to be offered on the network commissioners would prefer the GTN Steering Group to present to them a number of tests, with costs, that have been prioritised.

9.2 *Criteria for the evaluation of genetic tests for NHS service*

A slide presentation was given which highlighted a potentially large number of disorders within one medical speciality that would warrant genetic tests. The London Dysmorphology Database was scanned for all syndromes where genes had been found (473 syndromes) and these were then categorised into 3 levels of seriousness. Furthermore each of these categories of seriousness were graded for commonality. It was concluded that there could be a total of 293 dysmorphology syndromes where it would be reasonable to expect genetic tests. This exercise emphasised the need for a robust system to be in place to decide which tests should be given funding on the network. In connection with this it was also suggested that gene tests should be packaged together according to clinical speciality in one place. This model would allow for specialists in a particular clinical field to work together in one place and thus foster expertise in that medical speciality.

A paper entitled 'Criteria for the Evaluation of Genetic Tests for NHS Service' was circulated. The paper detailed the suggested process for determining which genetic tests could be put forward to commissioners for consideration for NHS funding. It was decided that the Evaluation Panel should be the UK GTN Steering Group and the paper was amended accordingly. A gene dossier has been developed to initiate the evaluation

process. Additions to the gene dossier were agreed and the document was then accepted.

9.3 A précis of 'Criteria for the Evaluation of Genetic Tests for NHS Service' will be added to the "Framework for Delivering the UK Genetic Testing Network" document.

10. Clinical Users Group

10.1 Previously it was considered that one Clinical User Group could be established to identify new genetic tests and give a view about priorities. It was suggested that rather than having one over arching Clinical User Group there should be a number of virtual Clinical User Groups, such as those covering different specialities and collaborative advisory groups. The Steering Group was interested to hear of work by a group of ophthalmologists on ophthalmology DNA tests.

It was agreed that the clinician who organised the ophthalmologists' meeting should be contacted and asked to attend the next Steering Group meeting in order to share experiences.

11. UK GTN Tests

11.1 Inter-Genetic Centres Referrals for Genetic Tests
A preliminary report providing information on tests that are imported by laboratories for the year 2001-2002 was tabled. The ten diseases for which most samples were reported as out of region referrals were identified. The Lead Scientist is to produce a more in depth report, using information from the laboratory application proformas and CMGS audit data, which will aim to highlight the top twenty diseases for which most samples are reported as out of region referrals. The Steering Group agreed that genetic tests for these diseases would be the first to be appraised via the evaluation process, including completion of gene dossiers.

11.2 Role of GTN in the IT developments for tracking systems for samples
The Steering Group agreed that the development of sample tracking for the relevant users of the network would play an essential role in the GTN. The Steering Group members were informed that a number of organisations are developing proposals for sample tracking systems. The Lead Scientist agreed to scope the systems that are available and to contact Dr Stenhouse to follow up the paper 'Sample Exchange for UKGTN' tabled at the UK GTN Steering Group meeting held on 28 January 2003.

11.3 Data standards
A paper entitled 'Data standard in Genetics' informed the Steering Group members of the work that the Manchester reference laboratory is leading on in this area. Data standards are essential to ensure the compatibility of genetic laboratory databases in different laboratories.

11.4 Currencies
The Steering Group was updated on the activities of the CMGS Work Load Units Working Group and the Steering Group thanked CMGS for doing this work. The

WLUs are to be implemented by 1st April 2003 in molecular genetic laboratories. There are currently no WLUs for medium and high throughput techniques and therefore a best practice meeting will be held in order to resolve this issue. It was agreed that when completing the section on the gene dossiers concerning WLUs, for techniques that are high throughput, it should be stated that WLUs are currently unavailable.

The Cytogenetic laboratories successfully piloted the Cytogenetics WLUs and the findings were reported back to GenCAG. An issue concerning the capital element in the WLU needs to be addressed before this scheme can be accepted by commissioners.

There are issues surrounding the casemix and complexity for clinical currencies in genetics and different models of determining them are being used to help resolve this.

12. Department of Health

12.1 It was noted that the DoH report: 'Summary of Responses to the Review into Commissioning Arrangements for Specialised Services' reinforces collaborative arrangements for commissioning. Copies of the full report can be found at http://www.doh.gov.uk/commissioning/specservsresponse03.pdf. Further guidance from the DoH is expected to be published shortly.

12.2 The Steering Group members will be notified by the Department of Health, via email, when the Genetics Green Paper is published.

13. Any Other Business

13.1 A letter had been received from a clinician expressing concerns about inequity of access to BRCA testing and that this will be discussed at the next GenCAG meeting on 8th May 2003. Data on clinical selection of patients for BRCA testing and on the tests currently carried out in laboratories have already been collected from 18 laboratories across the whole of the UK by the CMGS. The Steering Group asked if the report of the analysis of this data could be presented to the next Steering Group meeting in July. It was noted that NICE is also doing work on BRCA testing and that NICE will be contacted to determine the issues that they are addressing.

14. Dates and venues of the next meetings

8th July 2003
10.00 a.m. (for 10.30 a.m.) to 4.00 p.m., room 281D, Skipton House, London.

7th October 2003
10.00 a.m. (for 10.30 a.m.) to 4.00 p.m., room 136B, Skipton House, London.