UK GENETIC TESTING NETWORK CLINICAL & SCIENTIFIC ADVISORY GROUP
EIGHTH MEETING, 17 MARCH 2015
BMA HOUSE, Tavistock Square, LONDON

Present:

Rosalind Skinner, *Chair*
Jacquie Westwood, *UKGTN Director*
Anita Bruce, Association of Genetic Nurses and Counsellors
Peta Campbell, *UKGTN Project Team*
Hilary Burton, *PHG Foundation*
Trevor Cole, *Royal College of Physicians and Chair, UKGTN Rare Disease Service Improvement Group*
Jane Deller, *UKGTN Project Team*
Deirdre Evans, *Scotland*
Frances Flinter, *Chair, Medical Genetics Clinical Reference Group (Observer)*
Dominique Gray-Williams, *Welsh Health Specialised Services Committee*
Jacqui Hoyle, *UKGTN Project Team*
Steve Keeney, *Chair, UKHCDO-GLN*
Alastair Kent, *Genetic Alliance UK*
Mark Kroese, *UKGTN Pubic Health Advisor*
Nick Lench, *Chair, Joint Committee on Genomics in Medicine*
John Livingston, *Royal College of Paediatrics & Child Health*
Jackie McCall, *Northern Ireland*
Shehla Mohammed, *UKGTN Clinical Advisor*
Roger Mountford, *Chair, UKGTN Laboratory Membership & Audit Working Group*
Gail Norbury, *Royal College Pathologists*
Colin Pavelin, *Department of Health*
Fiona Stewart, *Chair, UKGTN Genetic Test Evaluation Working Group*
Allison Streetly, *Public Health England (part meeting)*
Jo Whittaker, *UKGTN Scientific Development Advisor and Chair, UKGTN Development Working Group*

Apologies:

Jill Clayton-Smith, *President, Clinical Genetics Society*
Ann Dalton, *Association for Clinical Genetic Science*
Andrew Devereau, *Personal Invitation*
Angela Douglas, *Association for Clinical Genetic Science*
Sian Ellard, *UKGTN Scientific Advisor*
Christine Lavery, *Patient Representative*
Ruth Newbury-Ecob, *Clinical Genetics Society*
Imran Rafi, *Primary Care Genetics Society*
Karen Temple, *Clinical & Academic Genetics, Personal Invitation*
Susan Walsh, *Patient Representation, Chronic Granulomatous Disorder Society*
Fiona Williams, *Patient Representation, CLIMB, National Information Centre for Metabolic Diseases*

Resignations:

Jill Clayton-Smith, *President, Clinical Genetics Society*
Sian Ellard, *UKGTN Scientific Advisor*
Frances Flinter, *Chair, Medical Genetics Clinical Reference Group (Observer)*
Jacqui Hoyle, *UKGTN Project Team*

1.0 WELCOME AND APOLOGIES

1.1 The Chair extended a warm welcome to members to the seventh meeting of the Clinical & Scientific Advisory Group (CSAG) and in particular to those who were attending for the first time.

Several members have stepped down from the group. Ruth Newbury-Ecob has taken over from Jill Clayton-Smith as President of the Clinical Genetics Society, Fiona MacDonald has taken over from Sian Ellard as UKGTN Scientific Advisor who has resigned to concentrate on laboratory work commitments and Jacqui Hoyle would be resigning from UKGTN at the end of March. Frances Flinter was stepping down as her tenure as Medical Genetics CRG Chair was

due to expire. The Chair thanked those committee members who were stepping down and said she was very appreciative of their support.

2.0 MINUTES OF LAST UKGTN CSAG MEETING (16.09.14)

2.1 The minutes from the previous meeting held on 16 September 2014 were agreed as an accurate record.

2.2 Matters Arising

2.3 New test recommendations 2015/16 – feedback from NHSE

NHSE's decision on funding priorities for 2015/16 remains delayed until the consultation on their prioritisation process has been completed. A decision on UKGTN's new test proposals approved by CSAG in September 2014 is likely to be made in the summer. In the meantime, the tests have been added to the UKGTN website.

2.4 Principles/case for standardisation of Next Generation Sequencing gene panels for tests for endorsement submitted to Joint Committee Genomic Medicine (JCGM)

The paper that outlines a mechanism for defining consensus gene sets for NGS panel tests has been shared with various groups including the JCGM. The principles are agreed and this is subject to wider consensus with Genomics England taking steps to standardise core genes within the 100,000 Whole Genome Project.

2.5 UKGTN Conference: Achieving Ambitions in Genomic Medicine

The UKGTN Director expressed her thanks to Alastair Kent for chairing the conference and to the speakers for the presentations, contributions and support. Due to the efforts of all those involved, the UKGTN team received very positive feedback.

3.0 DEPARTMENT OF HEALTH POLICY UPDATES

3.1 UK Rare Disease Forum

The Genetic Alliance UK member reported on progress that has been made since the last meeting towards meeting the commitments contained in the UK Strategy for Rare Diseases.

Unfortunately, due to problems with the technology, it had not been possible to show two videos to raise awareness of rare diseases. One is aimed at parents and patients on how to approach their GP if they suspect their child has a rare condition and the other at GPs about what to do when parents feel their child has a rare condition. There is also a video from the National Screening Committee on Homocystinuria. The videos are available on line.

- Patient video - http://youtu.be/puU6OOC7E_Q
- GP video - http://youtu.be/1CNGk5lovv4

The Forum receives updates on implementation from each home nation. Alastair Kent commended Wales on their plan for implementation as it has clear lines of accountability.

Fiona Marley has been appointed head of Highly Specialised Services at NHS England and is leading on the development of rare disease and age transition annexes to be included in all relevant service specifications provided by CRGs.

Ed Jessop is involved in work to identify areas where early diagnosis is difficult and where delays in diagnosis could result in sub-optimal care and be harmful to the patient.

John Burn, in his capacity as an NHS England Board non-executive director, has been involved in board-to-board meetings of NHS England and the Health and Social Care Information Centre (HSCIC) where rare diseases and genomics were discussed.

A tender exercise undertaken by Heath Education England (HEE) for a modular MSc in Genomic Medicine was near completion.

Public Health England (PHE) work to establish a rare disease register is on track. Discussions were also taking place with counterparts in Northern Ireland, Wales and Scotland on being part

of this work. DH is working with PHE to set up an expert working group to advise on the rare disease registry and other genomic databases. This working group will include representatives from all four countries in the UK.

Genomics England – ethical approval from the Health Research Authority Ethics Committee for the main phase of the programme was due to be confirmed shortly.

The first call for European Reference Networks (ERNs) is likely to be in December 2015 with approval of the first ERNs by July 2016.

To mark Rare Disease Day (28 February), events have been held across the UK. In England, a reception was hosted by Liz Kendall MP in the House of Commons and speakers included Earl Howe.

3.2 Update on Progress on Formal Review of UKGTN

There was no new relevant information to share on the outcome of the review of UKGTN. The Department of Health representative said he expected the position would become clearer once the NHS England Genomic Laboratory Redesign procurement was approved.

4.0 NHSE ITEMS

4.1 New Test Recommendations 2016/17

The new test recommendations for 2016/17 were endorsed by the group. The Chair of the Genetic Test Evaluation Working Group (GTEWG) wished to thank colleagues for their hard work. It was recognised this is a huge commitment on all members and the independent review of Next Generation Sequencing validation has been particularly helpful. She also commended laboratories for the quality of gene dossiers and information provided which was in most cases excellent. The Chair of CSAG thanked Fiona Stewart for her dedication to this working group.

4.2 Financial implications for new tests to the Clinical Priorities Advisory Group

CSAG members endorsed the paper on estimated costs and cost savings for all tests recommended across all UK nations to be recommended to the Clinical Priorities Advisory Group.

4.3 Medical Genetics CRG (MGCRG) – update on activity

The dashboard is fully operational again. Work is taking place to standardise turnaround times across molecular and cytogenetic activity.

Guidance to Regional Genetic Centres (RGCs) on Direct to Consumer Testing – this draft paper was produced by the MG CRG Chair in consultation with the UKGTN Director and Ann Jarvis and is in response to a query raised where individuals who have arranged their own genetic tests privately or via other health systems subsequently approach RGCs for advice, counselling and confirmatory testing. Overall, CSAG members welcomed the guidance. The Genetic Alliance UK member said he found the guidance useful and looked forward to it being approved by the MG CRG. He said it would be helpful to have a consistent approach and asked whether similar policies were emerging in other countries in the UK. Representatives from Scotland and Northern Ireland said they hoped to be able to follow the same guidance.

The paper was endorsed by the group and will go back to the MG CRG for sign off.

4.4 NHSE Genomic Service Laboratory Redesign – feedback from provider event on 2 March

The UKGTN Scientific Development Advisor presented the update on workstreams at the event in Leeds as the UKGTN Director was unable to attend. She reported that there was no doubt that the conference provided an opportunity for participants to provide feedback although there was little new information to share.

5.0 DEVOLVED COUNTRIES

5.1 Northern Ireland

The representative from Northern Ireland provided the update.

Precise details of the Health Service budget are still awaited.

UKGTN test proposals approved at the September 2014 meeting have been placed on the service priorities list. Due to financial pressures it was noted that there are a number of competing bids for limited resources.

Commissioning arrangements are under review.

Northern Ireland engaged in Rare Disease Day with the main event held in Enniskillen and a series of road shows to raise the profile of rare diseases.

Involvement in the 100,000 Genomes Project including a Genomics Medical Centre is considered essential.

5.2 Scotland

The representative for Scotland provided the update.

There has been frenetic activity over the last 3 months in Scotland in response to the opportunity to bid for Medical Research Council (MRC) funding on genomics, and the possibility of NHS Scotland joining centres in England as a Genomics Medicine Centre. Work is continuing and the main current focus is on developing an outline strategy for the introduction of exome and genome sequencing for clinical care. The emerging view is that, clinically, exome sequencing is of considerably greater value at present than whole genome sequencing. The only areas in which whole genome sequencing is considered to be of current clinical value is in Deciphering Developmental Disorders and rare inherited conditions.

The need for bioinformatics skills and training is recognised and part of the NHS Scotland genomics strategy needs to address the training needs.

The submission date for the final bids for the MRC funding is in April and the intention is to submit a joined up proposal from research, NHS and industry.

The work underway in UKGTN on how it will evaluate exome and genome tests is being closely watched because it is fundamental to the planning of future centres of expertise.

The Molecular Pathology Evaluation Panel has now been operating in shadow form for several months and will go live on 1 April 2015. The web address will be circulated once the website is up and running.

5.3 Wales

There was no new information to share since the last meeting.

6.0 PROFESSIONAL ORGANISATIONS

6.1 Clinical Genetics Society (CGS)

There was no update from the CGS.

6.2 Association for Clinical Genetic Science (ACGS)

There was no update from the ACGS.

6.3 Association for Genetic Nurses and Counsellors (AGNC)

There was no new information to share since the last meeting.

6.4 Royal College of Pathologists

Briefing note provided – see attachment A.

7.0 UKGTN NEW DEVELOPMENTS

7.1 New genetic tests submitted by 31.01.15 for evaluation in 2015

Members were provided with a summary of the submissions which met the 31.01.15 deadline. The recommendations will be presented for endorsement to CSAG in September. The GTEWG held its first meeting of the current cycle last week and was pleased to welcome new members including three genetic counsellors and one scientist to the group.

7.2 **UKGTN/ACGS Workshop 10 February 2015**
Towards Clinical Genome Sequencing : Quality Assurance of Tests
Members were provided with the agenda for the workshop which UKGTN held in association with the ACGS to discuss the introduction of exome sequencing into the genetic test evaluation process and guidelines for best practice. The event provided an opportunity to debate some difficult issues and presentations are available on the UKGTN website. The ACGS will share updated best practice guidelines in due course.

7.3 **UKGTN/ACGS Policy for Organisational Responsibilities**
The UKGTN and ACGS have worked jointly in the development of the GenUs for a number of years. As both organisations contribute, the GenU system is considered jointly owned with each organisation having different responsibilities for the system. Members noted the policy for organisation responsibilities.

7.4 **Health & Social Care Information Centre (HSCIC) – update on molecular genetic test data collection 2013/14**
Members were advised that there had been a great deal of progress since the last meeting. HSCIC Statement of Need had been approved in September 2014 and since November NHSE has been negotiating the draft Directions with HSCIC and UKGTN. These are being finalised with aim for sign off by HSCIC Board in July.

7.5 **UKGTN/British Heart Foundation at British Cardiovascular Society annual conference on 8 June 2015 – Presentations 'The New Cardiac Genetic Testing Panels: Implications for the Clinical Cardiologists'**
Members were advised that UKGTN had secured sessions at this event to highlight new genetic tests available for cardiologists. Details of the sessions were shared with the group. Feedback will be provided at the September meeting.

8.0 **UKGTN BUSINESS**

8.1 **UKGTN Biennial Report published in December 2014**
Members were provided with hard copies of the UKGTN biennial report. The report was launched at the UKGTN conference in December. It is also available to download from the UKGTN website.

8.2 **End of Year Report – UKGTN Work Programme 2014/15**
Members noted progress against the end of year 2014/15 work programme.

8.3 **Draft 2015/16 UKGTN Work Programme**
Members reviewed the draft work programme for 2015/16. The draft will be sent to Working Group Chairs for ratification. Thanks were expressed to working group Chairs for their commitment to this work programme.

8.4 **UKGTN Policy for Data Sharing**
Members noted the policy on data sharing. The paper has been taken to the UKGTN Laboratory Membership and Audit Working Group and amended to reflect comments received. It will be available on the UKGTN website.

8.5 **UKGTN Risk Register**
The updated UKGTN Risk Register was endorsed by the group.

9.0 **A.O.B.**

9.1 **Dates of next meeting**

- 24 September 2015

The Royal College of Pathologists
Pathology: the science behind the cure

ATTACHMENT A

Report for the UK Genetics Testing Network Clinical and Scientific Advisory Group (CSAG) 17 March 2015

Specialist Advisory Committee for Genetics & Reproductive Science (last meeting 8th December 2014)

- A Bioinformatics Symposium was held on 29th October and was attended by 62 participants which had excellent feedback.
- Training and examinations – The College is developing e-learning materials and FRCPath examinations are being updated to accommodate the first tranche of MSC trainees that will be ready for Part 1 this year. Guidance has been issued on the future availability of the different examination formats. Examinations are being developed in Molecular Pathology and thought is being given to those for Bioinformatics/Health Informatics. FRCPath will form the exit examination for HSTT in Genetics.
- Consultations – the committee had responded to the College proposal to manage implementation of recommendations of the Pathology Quality Assurance Review. These relate to the a) external quality assurance schemes, surveillance and standard setting (KPIs) and b) personal proficiency assessment (revalidation). There is also a public consultation of a review of the European working time directive which will link in with proposals for full seven day working in England.
- EU IVDD and in-house exemptions – there remain concerns with the latest re-drafts. On one hand there appear to be some loosening in text. Specifically the change from accredited to standard EN 1ISO 15189 to compliant with the standard or other equivalent provision. On the other hand, there are additional requirements on the Health Institution that seem overly burdensome and would threaten the viability of many rare disease tests.

RCPath Council (last meeting 22nd January 2015)
Dr Suzy Lishman is the new College President.
A position statement on Lab centralisation wihtin Microbiology that appears pertinent to the Genomics service re-design has recently been published in the Bulletin and Health Serice Journal.
British Infection Association's Clinical Service Committee Network

Lessons from Micro.pdf

Pathology Catalogue Executive Team (last meeting 9th March 2015)
See attached Diagnostics Strategy paper on the National Laboratory Medicine Catalogue

NLMC Diagnostics Strategy Group Paper

Funding appears to have been secured till June and it is hoped that future funding will be secured under the auspices of the Professional Records Standards Body
The first test set of Genetic tests have been SNOMed CT coded and XLabs have developed the tool to upload the current spread sheet of tests into the catalogue
The Editorial Principles have been updated to include Genetics that has previously been an appendix.
The key task now is completing the schedule of tests.
Gail Norbury
Chair SAC for Genetics & Reproductive Science, NLMC Lead for Genetics
13/3/15