Genetic Laboratory Network

Background

The UKHCDO GLN (formerly GTN) was formed in 2002, arising out of the UKHCDO Genetic Working Party, with the aim of improving collaboration between laboratories and of ensuring quality and equity of service across the U.K. The network currently comprises 13 laboratories, 12 across the UK plus Dublin, involved in the molecular genetic analysis of haemophilia and related inherited bleeding disorders (many of the laboratories are also involved in other areas as well).

Representatives of the Network sit on the UKHCDO GWP, attend meetings of the UKGTN and attended those Haemophilia Alliance meetings that took place during 2014.

Meetings

The UKHCDO GTN holds bi-annual meetings and met on 21st November 2013 in London and 27th May 2014 in Manchester. The next meeting is scheduled for late October or November in Cardiff.

Chair & Secretary

Steve Keeney (Chair) and Vince Jenkins (Secretary) continue in these positions.

Current activities

1. UK Genetic Testing Network (UKGTN) revised remit – UKHCDO response:

The UKGTN have revised their remit to include *"Any genetic test provided by a UKGTN member laboratory for NHS service provision for rare disorders that usually affects fewer than 1 in 2000 as described in the UK Rare Disease Strategy"*. Steve Keeney has coordinated the overall opinion of the UKHCDO via the GLN and GWP on this proposal and has fed back to the UKGTN that there are no objections to this process. This was fed back formally at the UKGTN CSAG meeting on 16th of September. Individual GLN labs that are not registered with the UKGTN have been advised that they may wish to consider applying for membership if they perceive it is in their interest.

2. Laboratory Audit:

The latest round of the Haemophilia Centre Triennial audit programme was the first where the genetic lab audit was incorporated to form an integrated audit process. The success of the merged audit programme will be reviewed, with Marian Hill representing the UKHCDO-GLN.

3. National Haemophilia Database Genetics Portal

The inclusion of a portal to the National Haemophilia Database for the upload of genetic mutation data is now in routine use across the GLN. At the last check (August 2014) there were 1324 mutation entries across F8, F9 and VWF mutation data entries from the 12 labs registered.

4. Locus Reference Genomic (LRG) development

Steve Keeney has input to the development of LRG reference sequences for the main bleeding disorder genes. The F9 LRG is now live, F8 is in final stages of preparation, and VWF is in development.

5 Bleeding Disorder Genetic Analysis Best Practice Guidelines

The BPGs for Haemophilia A and Haemophilia B are still considered current, although nearing a review. The VWD genetic analysis guideline update is underway.

6 Haemophilia Genetics NEQAS scheme

The Haemophilia Genetics EQA scheme, run by UK NEQAS in Sheffield, continues with bi-annual distributions. The results for each round of the scheme are reviewed and discussed at the following network meetings and any relevant comments fed back to the steering group. The scheme currently includes F8, F9 and VWF gene analysis.

7. Participation in other groups

A representative from the network attends the:

- Clinical and Scientific Advisory Group UK Genetics Testing Network (see item 1 above)
- The Haemophilia Alliance meeting, attended by Bim Theophilus on behalf of the GLN, has now been replaced by the Clinical Reference Group for Haemophilia. No role for a scientific or laboratory member has been created in the new group.
- Various members sit on relevant UKHCDO working parties - Two members of the network sit on the UKHCDO GWP and have had input to the draft updated 'Clinical Genetic Services for Haemophilia' document, now submitted.

Dr Steve Keeney
Chairman, UKHCDO Genetic Laboratory Network
September 2014