

Genetic Testing Network

Background

The UKHCDO Genetic Testing Network (GTN) was formed in 2002, arising out of the UKHCDO Genetic Working Party (GWP), 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 U.K. plus Dublin, involved in the molecular genetic analysis of haemophilia (*F8 & F9 genes*) 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 and attend meetings of the Haemophilia Alliance and the UKGTN.

Meetings

The UKHCDO GTN holds bi-annual meetings and met on 13th November 2012 in Manchester and on 16th May 2013 in Edinburgh. The next meeting is scheduled for November 2013 in London (date TBA).

Chair & Secretary

At the meeting on 24th May 2012 Mike Mitchell and Marian Hill formally stood down as Chair & Secretary. Steve Keeney (Chair) and Vince Jenkins (Secretary) continue in these positions (formally appointed at the 24th May meeting 2012).

Current activities

1. Laboratory Audit

The 2010/11 audit round was the last 'stand-alone' genetic lab audit. The genetic lab audit has now been incorporated in to the UKHCDO Triennial Audit programme led by Dr. David Perry, with Marian Hill liaising on behalf of the Network. Audit paperwork for future audits was reviewed by Steve Keeney and Marian Hill. The final combined Clinical and Laboratory audit pro-forma has subsequently been used in the current round of haemophilia centre audits which are undergoing collation. The success of the merged audit programme to be reviewed once this round is complete.

2. Bleeding Disorder Genetic Analysis Best Practice Guidelines

The BPGs for Haemophilia A and Haemophilia B are still considered current. VWD genetic analysis guidelines are due an update and this is underway.

In future all BPGs, including revisions, must be approved by the appropriate Steering group and possibly referred to the British Committee for Standards in Haematology (BCSH). VWD genetic analysis guidelines are due an update and this will be taken forward in 2013.

3. 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 network meetings. The scheme currently includes *F8*, *F9* and *VWF* gene analysis – usually restricted to only partial gene analysis. Discussion over the utility and wider appeal of exercises involving the rarer bleeding disorders has not led to the selection of any likely candidates.

4. National Haemophilia Database Genetics Portal

The inclusion of a portal to the National Haemophilia Database for the upload of genetic mutation data was piloted in Manchester, Birmingham and Cardiff and has now been rolled out across the laboratory network. Labs have been asked to prioritise the inclusion of post year 2000 PUPs to aid examination of inhibitor development risk in this cohort. At the last check (4/9/13) there were 698 mutation entries across *F8*, *F9* and *VWF* mutation data entries from the 11 labs registered (proportion of these who are post 2000 PUPs not known).

5. Late amniotic fluid pre-natal diagnosis survey

Steve Keeney has circulated a request for current practice in this area. If deemed appropriate on collation of results, and after further discussion at the next meeting, this may form the basis of a network publication.

6. Bio-informatics/Locus-specific mutation databases

Steve Keeney is coordinating, on behalf of the network, development of Locus Reference Genomic (LRG) reference sequences for the main bleeding disorder genes. The planned development of standardised formats for mutation databases through EAHAD and ISTH entities is an on-going, long term project.

7. Participation in other groups

A representative from the network attends the:

- Clinical and Scientific Advisory Group UK Genetics Testing Network
- The Haemophilia Alliance
- 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.

Dr Steve Keeney
Chairman, UKHCDO Genetic Testing Network
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