Genetic Laboratory Network (UKHCDO-GLN)

Background

The UKHCDO GLN was formed in 2002, arising out of the UKHCDO Genetic Working Party (UKHCDO 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 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 attend meetings of the UK Genetic Testing Network (UKGTN) and the UKHCDO GWP.

Meetings

The UKHCDO GLN holds bi-annual meetings and met on 22 November 2016 in Birmingham and 17 May 2017 in Manchester. The next meeting is scheduled for November 2017 in London.

Chair & Secretary

Steve Keeney stepped down at the November 2016 meeting and Megan Sutherland was appointed to the Chair prior to the May 2017 meeting. Catriona Keenan continued her role as Secretary.

Current activities

1. NHS England genetic laboratory re-designation exercise. This process is developing rapidly and although it has not directly impacted the configuration of bleeding disorder laboratory services it is likely to do so. As such the network discusses this process at each meeting.

Current estimated timelines for the re-designation process:

- Service specifications review complete and signed off - 29 September
- Directory of test/testing strategy signed off – 22 November
- Published OJEU – 30 November
- Invitation to Tender (ITT) issued – 4th December
- ITT closes – 29th January 2018
- ITT outcome announced - 13 April
- Meetings with successful bidders commence – 14th April
- Mobilisation phase later in 2018

2. Laboratory Audit – ISO 15189: Clinical Pathology Accreditation (CPA) have implemented the ISO 15189 quality standard applicable to laboratories. The GLN continues to share practical advice and knowledge as the revised inspection process is applied to member laboratories, sharing examples of good practice, and implementation of an informal sample exchange scheme between members of the GLN for disorders that are not

covered by UK NEQAS. The majority of laboratories have now successfully undergone the ISO inspection process with the surveillance visit process now underway.

3. **National Haemophilia Database Genetics Portal**: This concerns the inclusion of a portal to the National Haemophilia Database (NHD) for the upload of genetic mutation data, in use across the GLN. Members of the GLN are now able to search the NHD for mutations that been found previously in other centres, however the patient details are not shown. Members are also able to search for patients to confirm which centre they are registered at and if a mutation has been detected prior to contact for release of relevant details if appropriate (no further information regarding the mutation is made available).

4. **Bleeding Disorder Genetic Analysis Best Practice Guidelines:** The VWD genetic analysis guideline update is ongoing. The BPGs for Haemophilia A and Haemophilia B are nearing review.

5. **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 meeting and any relevant comments fed back to the steering group. The scheme currently includes F8, F9 and VWF gene analysis. The GLN have discussed the use of Locus Reference Genomic (LRG) sequences when reporting genetic variants and the recommendations for their use by different EQA schemes. At present, it has been decided to watch the maturation of the LRG sequences until a time at which they are indicated for incorporation into genetic reports.

6. Participation in other groups

A representative from the network attends the:

- Clinical and Scientific Advisory Group UK Genetics Testing Network
- Representatives of the Network input to the UKHCDO GWP.

7. General

The GLN has discussed the application of next generation sequencing approaches to inherited bleeding disorders. GLN members who are currently using this approach are analysing panels of genes and filtering results according to the clinical indication. There has been discussion about whether Sanger sequencing still has a place for the analysis of certain inherited disorders and it was considered necessary for circumstances such as carrier analysis and targeted mutation analysis.

Megan Sutherland,
Chair, UKHCDO Genetic Laboratory Network
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