

## UKHCDO Genetic Testing Network

### Background:

The UKHCDO 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 20<sup>th</sup> May at the Manchester Royal Infirmary and 4<sup>th</sup> November at the Birmingham Children's Hospital in 2010. The last meeting was on the 12<sup>th</sup> May 2011 at the John Radcliffe Hospital in Oxford. It is hoped to hold a satellite / working party meeting at the BSHT / UKHCDO meeting in Brighton in October.

The UKHCDO Genetic Working Party, attended by the Chair of the GTN, met on 3<sup>rd</sup> March, 17<sup>th</sup> May, 14<sup>th</sup> September and 30<sup>th</sup> November in 2010 and on 8<sup>th</sup> March and most recently 14<sup>th</sup> June in 2011.

### Current activities:

1. Laboratory Audit: The latest round of Genetic Laboratory Audits is now complete. Due to the bad weather the audit process ran over into 2011. Data from the individual audit visits will be collated and reviewed. Data from the previous rounds of audits (2005 & 2007) has been submitted to the International Society for Thrombosis & Haemostasis (ISTH) meeting in Japan in July 2011. It is proposed that in future the lab audit will be incorporated into the UKHCDO Triennial audit process.
2. Haemophilia Best Practice Guidelines: The BPGs for Haemophilia A and Haemophilia B have been reviewed and updated by the original authors – the originals were published in 2005. They have been submitted to the CMGS Best Practice Guidelines Website. Once they are published on the site the Network will submit a letter to the journal Haemophilia (where the original guidelines were published) informing of the updated versions. The Network has also produced BPG for the molecular analysis of VWD.
3. Haemophilia Genetics NEQAS scheme: The Haemophilia Genetics EQA scheme, run by UK NEQAS in Sheffield, continues with bi-annual distributions (last distribution was Exercise 18 in May 2011). The scheme continues to highlight variation across

the Network and to provoke much debate. The scheme currently includes F8, F9 and VWF gene analysis – usually restricted to only partial gene analysis. There was discussion about the possibility of expanding the scheme to include other genes involved in inherited bleeding disorders. Since not all the labs participating in the scheme provide analysis for other possible genes, it is proposed that this would be an additional & optional part of the scheme and not replace one of the standard bi-annual distributions. It is hoped that this may be introduced soon as there is much enthusiasm for the idea.

4. National Haemophilia Database / Local Database: The proposed expansion of the National Haemophilia Database to include genetic mutation data is on-going with three pilot sites, Manchester, Oxford and Nottingham, looking at this. A portal is being developed to allow Labs to upload mutation data to the NHD. The Network is also discussing with the UKHCDO improved laboratory access to the database. This project is on-going.

The IT team have also developed a web-based Local database which, it is planned, could interface with NHD. Some labs are very keen but others are already 'locked in' to IT systems and would find it difficult to change.

5. Locus-specific mutation databases: The network has raised and discussed concerns over the future of locus-specific mutation databases. These are an invaluable resource but vulnerable as many are 'one man operations' e.g. F8 & F9. It was suggested that ISTH might be persuaded to take stewardship. There are also on-going discussions with regards to possible commercial sponsorship to support the maintenance and upgrading of some of these sites.
6. UKHCDO Genetics Working Party: Two members of the network sit on the UKHCDO GWP, Dr Mike Mitchell and Dr Tony Cumming. The GWP is currently involved in re-writing the 'Clinical Genetic Services for Haemophilia' document first published in 2004. Mike and Tony are centrally involved in this project, with particular emphasis on laboratory aspects, and liaise with the Network as appropriate.

Dr Mike Mitchell  
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