

Genetics Working Party

Membership

John Pasi	Chair
Keith Gomez	Secretary
Tony Cumming	
Mike Laffan	BCSH representative
Dianne Marshall	UK RCN Haemophilia Nurses Association
Alan Fryer	Joint Committee for Medical Genetics
Heather Skirton	Association of Genetic Nurses and Counsellors
Mike Mitchell	Chairman Laboratory Network
Christopher Ludlam	

Professor Christopher Ludlam retired from the group over the last year and we would like to thank him immensely for his work with the GWP.

Remit

The purpose of the GWP is to advise and inform the UKHCDO on relevant issues in genetics.

The Genetics Laboratory Network (representing the laboratories providing services in genetic analysis to haemophilia centres) remains a sub-group of this working party and reports to the UKHCDO via the GWP.

The GWP will maintain a clinical governance role overseeing the provision of genetic services in haemophilia services.

Meetings and work streams

Aside from the regular work of the GWP in providing a governance and feedback forum, identified work streams for this current iteration of the GWP are:

Guideline review

The existing guidelines have been updated and are in final draft form. They will be circulated for comments and have been written to act as a practical guide

Audit programme

Following proposals for including the genetics audit within the triennial audit, the audit programme has now been merged as part of the triennial programme. Hence, rather than a stand alone programme the genetics audit will take place in the current round of audits with an additional scientific auditor for the relevant sites.

Capture of genetic information on HCIS and UKHCDO NHD

Work has continued on delivery of a module for HCIS for use as a genetics register and ensure appropriate capture of genetic information on NHD for research purposes in liaison with the Inhibitor Working Party and Data Management Group. Discussions still remain on this point.

Future of Working Party

The Working Party has now completed a three year cycle.

As genetics is a rapidly moving field, there is a clinical governance responsibility to ensure that UKHCDO is kept up to date with advances in clinical genetics practise in general and especially in relation to congenital bleeding disorders. The Organisation needs to have a focus for such a responsibility and a 'formal' link with the specialty of clinical genetics. One of the key roles of the GWP has been to provide this link and a multidisciplinary forum in haemophilia for considering genetic services. UKHCDO only represents physicians and other disciplines must be included in advising about service provision. The GWP as a multidisciplinary forum has been greatly enhanced and enlivened by having members from other non-haemophilia specialties with different backgrounds.

The Working Party considers there remains a need for a committee to be appointed to take continuing responsibility for the following topics:

1. Clinical governance oversight of developments in clinical genetics
2. Multidisciplinary forum for review of haemophilia genetic services
3. Collaboration with the Haemophilia Laboratory Network
4. Audit of clinical and laboratory genetic services
5. Oversight, with Data Management Committee, of mutation data on NHD
6. Other aspects of genetic services to be agreed with the Advisory Group

Prof KJ Pasi
Chairman, Genetics Working Party
September 2012

UKHCDO Genetic Testing Network

Background

The UKHCDO 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 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 29th November 2011 at the City Hospital in Belfast and 24th May 2012 at Queen's Medical Centre, Nottingham. The next meeting is scheduled for 13th November 2012 in Manchester.

The UKHCDO Genetic Working Party, attended by the Chair of the GTN, met on 16th November 2011 and most recently on 27th March 2012.

New Chair & Secretary

At the meeting on 24th May 2012 Mike Mitchell and Marian Hill formally stood down as Chair & Secretary to be replaced by Steve Keeney (Chair) and Vince Jenkins (Secretary).

Current activities:

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 is currently being reviewed by Steve Keeney and Marian Hill.

Marian Hill presented a poster highlighting the findings from the Audit rounds in 2005, 2007 & 2010 at the International Society for Thrombosis & Haemostasis (ISTH) meeting in Japan in July 2011, on behalf of the Network.

Haemophilia Genetic Analysis 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 are published on the CMGS and UKHCDO web-sites. Discussions are on-going re a possible editorial in Haemophilia about the BPGs.

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.

Haemophilia Genetics NEQAS scheme: The Haemophilia Genetics EQA scheme, run by UK NEQAS in Sheffield, continues with bi-annual distributions (last distribution was Exercise 20 in May 2012, a partial F8 gene sequence). The scheme is highly valued by the Network and provokes much debate. The scheme currently includes F8, F9 and VWF gene analysis – usually restricted to only partial gene analysis. There has been discussion, over the past two years, about the possibility of expanding the scheme to include other genes involved in inherited bleeding disorders. So far there is no progress with this. It is hoped that this may be introduced soon as there is much enthusiasm for the idea.

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. Due to differing requirements of each centre involved in the pilot for this it has proved difficult to standardise and is unlikely to be adopted across multiple centres.

Prenatal Diagnosis Survey: The Network performed a survey, led by Vince Jenkins, of Prenatal Diagnosis Services across the network. This was a useful snapshot of the current status and, once again, highlighted variation across the network. Vince Jenkins presented the results of the survey in a PowerPoint presentation at the meeting in Nottingham – the presentation was distributed following the meeting along with the Minutes.

Locus-specific mutation databases: Significant progress has been made with regards to securing the existing LSMDs, specifically HADB and Haemophilia B – both of which were particularly vulnerable. EAHAD is co-ordinating the maintenance and update and will, hopefully, provide future funding to support these databases. It is planned that all the databases will be standardised to the Leiden Open Variation db format (LOVD), as is the case for the VWD database. It is hoped that other databases such as FXI, RBD, FXIII and FVII will be included.

UKHCDO Genetics Working Party: Two members of the network sit on the UKHCDO GWP, currently Mike Mitchell and 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 Steve Keeney,
Chairman, UKHCDO Genetic Testing Network
September 2012