

Genetics Working Party

Membership

Keith Gomez	Chair
Nicola Curry	
Gerry Dolan	
Steve Keeney	Representing Genetics Laboratory Network
Mike Laffan	
Megan Sutherland	Representing Genetics Laboratory Network

Remit

1. Continue role as oversight committee for issues related to genetics in haemophilia
2. Annual guideline review
3. Support ThromboGenomics programme and enrolment of patients in NIHR UK Biobanking initiative
4. Support genetic analysis of all patients with inherited bleeding disorders and for results to be recorded on NHD
5. Provide oversight of gene therapy trials for haemophilia in the UK

Meetings and work streams

The working party met by teleconference in July 2017 and agreed to meet by teleconference every 3-4 months. The next meeting is scheduled for October 2017.

There is a steady increase in genetic testing of patients with inherited bleeding disorders for whom analysis was not previously accessible using the ThromboGenomics and other next generation sequencing platforms. This has led to changes in the ways in which genetic results are handled in the clinic. The guidelines for genetic services were published relatively recently in 2015 and deal with the framework for service provision rather than the handling of results. While these do not require updating at this stage, the working party will produce a position statement with recommendations on the interpretation of genetic results in the context of new guidelines for reporting and how genetic information should be conveyed to patients.

The laboratory guidelines for the molecular analysis of Haemophilia A and B and von Willebrand disease require updating to incorporate the use of next generation sequencing and guidelines on variant classification and reporting.

Dr K Gomez,
Chair, Genetics Working Party
September 2017