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 genetic analysis of all patients with inherited bleeding disorders and for results to be recorded on NHD
4. Support enrolment of patients in NIHR UK Biobanking initiative
5. Provide oversight of gene therapy trials for haemophilia in the UK

Meetings and main work streams

The working party met by teleconference in October 2019.

The following guideline has been published this year: *Recommendations for the clinical interpretation of genetic variants and presentation of results to patients with inherited bleeding disorders. Haemophilia. 2019 Jan;25(1):116-126.*

There are three main work streams at the moment.

Reconfiguration of Genetic Services by NHS England

Following a tendering process, four Genomics Laboratory Hubs (GLH) were designated as preferred providers for genetic analysis of heritable bleeding, platelet and thrombotic disorders by NHS England in September 2018. These are Wessex and West Midlands GLH (led by Oxford), South London GLH (led by St Thomas'), North West GLH (led by Manchester) and Yorkshire and North East GLH (led by Newcastle/Sheffield). An approved list of genes that can be tested using this service is at https://panelapp.genomicsengland.co.uk/panels/545/. This is essentially the same as the tier 1 list approved by the International Society on Thrombosis and Haemostasis Scientific and Standardization Committee (ISTH SSC) for Genomics in Haemostasis. Requestors from within NHS England will not be charged for testing of genes on this list if the sample is sent to the GLH. NHS England will re-imburse GLHs for the tests that they carry out. The expectation is that GLHs will be ready to start providing this service in April 2020.

In the meantime the Haemophilia Genetics Laboratory Network (which includes the four GLHs, other current providers of genetic testing in England and representatives from Wales, Scotland and Ireland) will continue to meet with the aim of ensuring that the service meets the needs of clinicians. To guide this process a workshop for stakeholders in the provision

of non-malignant haematology genetics services took place in July 2019 and there will be further meetings in 2020.

Consent

The Joint Committee on Genomics in Medicine published new guidance on consent and confidentiality in July 2019 (https://www.rcplondon.ac.uk/projects/outputs/consent-and-confidentiality-genomic-medicine). The main changes are that the consent process should cover the possibilities of variants of uncertain significance and incidental findings. The working party will revise the UKHCDO form and information sheet to make it consistent with this guidance.

Creation of new category of Heritable Platelet Disorders on NHD

In conjunction with the Data Mangement Working Party the NHD will be revised so that Heritable Platelet Disorders will be adequatley capture. The aim will be to facilitate entry of genetic data, laboratory data and clinical features of syndromes.

Other work streams

The working party monitors the capture of genetic information in the NHD. The table overleaf shows the capture of genetic diagnosis for haemophilia A and B as of November 2018. Compared with last year the percentages of total patients with haemophilia and a genetic diagnosis have increased. From 27 to 30% for haemophilia A and from 11 to 21% for haemophilia B.

Health Education England provide factsheets on heritable conditions for non-specialists, predominantly in primary care. We will update the factsheet on haemophilia and produce a new sheet on von Willebrand Disease.

Dr Keith Gomez,
Chair, Genetics Working Party
October 2019