Genetics Working Party

Membership

Keith Gomez	Chair
Nicola Curry	
Gerry Dolan (stood down March 2020)	
Steve Keeney (stood down March 2020)	Representing Genetics Laboratory Network
Mike Laffan	
Megan Sutherland	Representing Genetics Laboratory Network
Kate Talks (joined Sept 2020)	
Jayashree Motwani (joined Sept 2020)	
Gavin Ling (joined Sept 2020)	

Remit

1. Provide oversight of issues related to genetics in haemophilia
2. Review guidance on genomic testing and update as required
3. Support genetic analysis of all patients with heritable haemostatic disorders and for results to be recorded on NHD

Meetings and main work streams

The working party last met by teleconference in October 2019. The 3 year term came to an end at the March advisory committee meeting and was reformed with new members joining following the advisory committee meeting in September 2020

There are three main work streams at the moment.

Reconfiguration of Genetic Services by NHS England

The four Genomics Laboratory Hubs (GLH) providing testing for haemostatic disorders under the national contract in England, at no charge to NHS requestors, are:

- Wessex and West Midlands GLH (led by Oxford)
- London South GLH (led by St Thomas')
- North West GLH (led by Manchester)
- Yorkshire and North East GLH (led by Newcastle/Sheffield).

The genes that can be tested are listed in the following panels:

- Bleeding and platelet disorders (R90) https://panelapp.genomicsengland.co.uk/panels/545/
- Thrombophilia (R97) https://panelapp.genomicsengland.co.uk/panels/516/

Oversight of the service is provided through Non-Malignant Haematology Genomics workshops that met in January and July 2020. The transition period for testing to move to this framework commenced on April 1st 2020. In the last few months most hubs have been validating their panels which has been delayed by redeployment of staff away from genomic testing during the pandemic.

Consent

A revised information sheet and consent form for genomic testing to cover bleeding and thrombotic disorders is now available at http://www.ukhcdo.org/wp-content/uploads/2020/02/Genomics-Information-Sheet-and-Consent-Form_02-2020.docx

A UK working group for discussion of consent in children was being formed prior to the pandemic and work is expected to continue on this next year.

Creation of new category of Heritable Platelet Disorders on NHD

In conjunction with the Data Mangement Working Party the NHD has been revised with a new category of Heritable Platelet Disorder to replace the following categories: Platelet defects (misc), Glanzmann thrombasthenia, Bernard Soulier Syndrome, Severe Platelet Disorders – Other, Heritable Platelet Function Disorder and Platelet-type Pseudo von Willebrand Disease. The new category went live on 21st September.

Other work streams

The working party has updated factsheets provided by Health Education England on haemophilia and von Willebrand Disease. These are aimed at non-specialists, predominantly in primary care and can be accessed at https://www.genomicseducation.hee.nhs.uk/news/new-conditions-factsheets/.

The table shows the capture of genetic diagnosis for haemophilia in the NHD. Having seen annual increases in previous years the numbers are relatively unchanged compared with 2019.

Dr Keith Gomez,
Chair, Genetics Working Party
October 2020

Number of patients with a genetic diagnosis by region (based on patient's postcode and not registered centre)

Diagnosis	Region	Patients with a genetic diagnosis (% of registered patients)									
		Factor VIII / IX level (iu/dl)									
		< 1		1 & 5		> 5		40+		N/K	
		All Variants	Validated Variants	All Variants	Validated Variants	All Variants	Validated Variants	All Variants	Validated Variants	All Variants	Validated Variants
Haemophilia A	East Midlands	88 (49.4%)	68 (38.2%)	9 (15.3%)	8 (13.6%)	85 (25.8%)	66 (20.1%)	21 (11.8%)	10 (5.6%)	1 (12.5%)	1 (12.5%)
	East of England	27 (18.4%)	26 (17.7%)	14 (16.1%)	9 (10.3%)	41 (10.3%)	36 (9.1%)	4 (2.5%)	3 (1.9%)	0 (0.0%)	0 (0.0%)
	London	109 (30.4%)	107 (29.9%)	30 (36.6%)	28 (34.1%)	93 (20.8%)	90 (20.1%)	11 (6.7%)	9 (5.5%)	0 (0.0%)	0 (0.0%)
	North East	10 (15.9%)	8 (12.7%)	8 (20.5%)	8 (20.5%)	42 (29.0%)	37 (25.5%)	23 (16.8%)	20 (14.6%)	0 (0.0%)	0 (0.0%)
	North West	64 (29.0%)	59 (26.7%)	30 (35.7%)	26 (31.0%)	103 (27.3%)	86 (22.8%)	43 (28.9%)	24 (16.1%)	1 (14.3%)	1 (14.3%)
	Northern Ireland	42 (56.0%)	42 (56.0%)	18 (56.3%)	18 (56.3%)	64 (41.8%)	64 (41.8%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	Scotland East	7 (8.8%)	7 (8.8%)	4 (10.8%)	4 (10.8%)	19 (10.5%)	16 (8.8%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	Scotland West	2 (3.1%)	0 (0.0%)	2 (6.3%)	2 (6.3%)	3 (2.2%)	3 (2.2%)	3 (2.3%)	3 (2.3%)	0 (0.0%)	0 (0.0%)
	South East	131 (40.4%)	103 (31.8%)	37 (43.5%)	30 (35.3%)	131 (21.5%)	88 (14.4%)	51 (23.8%)	19 (8.9%)	0 (0.0%)	0 (0.0%)
	South West	39 (26.4%)	36 (24.3%)	8 (11.9%)	2 (3.0%)	37 (11.0%)	21 (6.3%)	13 (16.5%)	5 (6.3%)	0 (0.0%)	0 (0.0%)
	Wales	36 (40.0%)	34 (37.8%)	9 (31.0%)	8 (27.6%)	39 (19.6%)	38 (19.1%)	4 (6.9%)	4 (6.9%)	0 (0.0%)	0 (0.0%)
	West Midlands	39 (21.9%)	37 (20.8%)	5 (8.2%)	4 (6.6%)	15 (5.8%)	13 (5.0%)	2 (1.9%)	1 (1.0%)	0 (0.0%)	0 (0.0%)
	Yorkshire and the Humber	22 (14.0%)	19 (12.1%)	4 (3.7%)	3 (2.8%)	32 (8.8%)	20 (5.5%)	13 (8.7%)	8 (5.4%)	0 (0.0%)	0 (0.0%)
Haemophilia A Total		**616 (29.6%)**	**546 (26.2%)**	**178 (22.2%)**	**150 (18.7%)**	**704 (17.9%)**	**578 (14.7%)**	**188 (11.2%)**	**106 (6.3%)**	**2 (2.8%)**	**2 (2.8%)**
Haemophilia B	East Midlands	10 (33.3%)	8 (26.7%)	4 (22.2%)	1 (5.6%)	7 (12.7%)	6 (10.9%)	2 (9.5%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	East of England	0 (0.0%)	0 (0.0%)	3 (10.7%)	2 (7.1%)	2 (2.5%)	1 (1.2%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	London	8 (12.1%)	3 (4.5%)	6 (11.1%)	4 (7.4%)	9 (7.1%)	7 (5.6%)	2 (4.0%)	1 (2.0%)	0 (0.0%)	0 (0.0%)
	North East	3 (21.4%)	3 (21.4%)	1 (7.1%)	1 (7.1%)	3 (9.7%)	1 (3.2%)	3 (8.1%)	1 (2.7%)	0 (0.0%)	0 (0.0%)
	North West	13 (37.1%)	9 (25.7%)	14 (35.9%)	14 (35.9%)	18 (27.7%)	13 (20.0%)	18 (38.3%)	10 (21.3%)	0 (0.0%)	0 (0.0%)
	Northern Ireland	2 (25.0%)	2 (25.0%)	1 (25.0%)	1 (25.0%)	4 (22.2%)	4 (22.2%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	Scotland East	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	Scotland West	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	South East	21 (33.3%)	10 (15.9%)	15 (26.8%)	6 (10.7%)	24 (20.2%)	11 (9.2%)	13 (28.9%)	3 (6.7%)	0 (0.0%)	0 (0.0%)
	South West	3 (15.8%)	1 (5.3%)	4 (20.0%)	2 (10.0%)	10 (13.7%)	6 (8.2%)	1 (6.3%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	Wales	4 (30.8%)	4 (30.8%)	12 (54.5%)	12 (54.5%)	11 (25.0%)	10 (22.7%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	West Midlands	8 (26.7%)	5 (16.7%)	6 (31.6%)	6 (31.6%)	2 (3.0%)	2 (3.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)	0 (0.0%)
	Yorkshire and the Humber	2 (7.7%)	2 (7.7%)	1 (3.8%)	0 (0.0%)	2 (3.9%)	2 (3.9%)	4 (13.3%)	3 (10.0%)	0 (0.0%)	0 (0.0%)
Haemophilia B Total		**74 (20.2%)**	**47 (12.8%)**	**67 (19.5%)**	**49 (14.3%)**	**92 (11.1%)**	**63 (7.6%)**	**43 (11.9%)**	**18 (5.0%)**	**0 (0.0%)**	**0 (0.0%)**

Patients registered with the NHD 2019/20
Haemophilia A includes females with FVIII deficiency & haemophilia A carriers.
Haemophilia B includes females with FIX deficiency, haemophilia B carriers, FIX Leyden & FIX Leyden carriers
Genetic variants entered on the NHD require a second-person entry as a validation check. This table shows all Genetic variants and validated entries separately by disease severity

Taken from the 2020 UKHCDO Annual Report