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 October 2017 and September 2018.

The working party has produced a new good practice paper entitled *Recommendations for the clinical interpretation of genetic variants and presentation of results to patients with inherited bleeding disorders*. This has been submitted for publication.

The provision of genetic testing services in England has been revised by NHS England with a consolidation of services into four hubs. We are currently in the transition period and over the next year, the testing of samples will be transferred to the hubs. There should not be any break in service provision, but for many members the testing laboratory will change. On behalf of UKHCDO the Genetics Working Party responded to the NHS England stakeholder consultation regarding potential issues regarding the quality of genetic reporting. We have been used to a service tailored to the needs of UK haemophilia clinicians and the working party will monitor the service to assess how this is maintained.

The ThromboGenomics platform continues to be the main platform providing testing for genes in rare bleeding and platelet disorders. The long-term provision of this service is unclear following the NHS England service reconfiguration, but the current plan is for it to continue perhaps with a testing fee. Genomics England 100K project is due to close later this year and it is not clear whether a similar testing platform will be introduced to replace it.

There are now 11 centres enrolling patients into the NIHR rare diseases project. As of June 2018, 869 patients had been enrolled including 340 from Liverpool and 236 from Oxford.

A proposal for revising the registration of heritable platelet disorders has been submitted to the Data Management Working Party. This should allow capture of the genetic variants and other phenotypic features.

The table below shows the capture of genetic diagnosis for haemophilia A and B as of end of April 2018. There is regional variability in submission of data with some examples of incomplete submission. This is partly due to staffing issues in laboratories and will be monitored by the Genetics Laboratory Network.

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

Diagnosis	Region	Factor VIII / IX level (iu/dl)				
		< 1	1 & 5	> 5	40+	N/K
		Patients with a genetic diagnosis (% of registered patients)				
Haemophilia A	East Midlands	72 (41.9%)	11 (16.7%)	65 (20.8%)	10 (7.4%)	1 (10.0%)
	East of England	28 (19.7%)	7 (8.0%)	39 (9.8%)	4 (2.7%)	0 (0.0%)
	London	95 (27.5%)	22 (29.3%)	81 (19.2%)	9 (5.9%)	0 (0.0%)
	North East	12 (17.4%)	8 (21.1%)	36 (29.3%)	20 (16.8%)	0 (0.0%)
	North West	55 (26.8%)	28 (31.5%)	92 (25.1%)	23 (16.7%)	1 (20.0%)
	Northern Ireland	43 (58.9%)	18 (60.0%)	66 (45.2%)	0 (0.0%)	0 (0.0%)
	Scotland East	7 (8.4%)	4 (11.1%)	17 (10.0%)	0 (0.0%)	0 (0.0%)
	Scotland West	2 (3.0%)	2 (6.3%)	3 (2.3%)	3 (2.5%)	0 (0.0%)
	South East	99 (31.0%)	29 (36.7%)	82 (14.3%)	20 (11.2%)	0 (0.0%)
	South West	33 (23.1%)	3 (4.4%)	19 (5.7%)	5 (7.7%)	0 (0.0%)
	Wales	32 (39.0%)	9 (26.5%)	34 (18.7%)	4 (9.3%)	0 (0.0%)
	West Midlands	37 (21.5%)	3 (5.1%)	15 (6.1%)	1 (1.0%)	0 (0.0%)
	Yorkshire and the Humber	18 (12.2%)	4 (3.4%)	20 (5.5%)	8 (6.3%)	0 (0.0%)
	Total	**533 (26.4%)**	**148 (18.3%)**	**569 (15.1%)**	**107 (7.3%)**	**2 (2.8%)**
Haemophilia B	East Midlands	7 (23.3%)	3 (14.3%)	4 (8.2%)	0 (0.0%)	0 (0.0%)
	East of England	1 (2.6%)	2 (7.1%)	1 (1.2%)	0 (0.0%)	0 (0.0%)
	London	4 (6.5%)	4 (7.1%)	8 (6.7%)	1 (2.2%)	0 (0.0%)
	North East	2 (16.7%)	1 (9.1%)	1 (3.3%)	1 (3.6%)	0 (0.0%)
	North West	8 (22.9%)	13 (35.1%)	14 (21.9%)	10 (21.7%)	0 (0.0%)
	Northern Ireland	3 (33.3%)	1 (25.0%)	4 (23.5%)	0 (0.0%)	0 (0.0%)
	Scotland East	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%)
	South East	7 (11.5%)	8 (14.3%)	11 (10.4%)	3 (9.4%)	0 (0.0%)
	South West	1 (5.6%)	2 (11.1%)	6 (9.2%)	0 (0.0%)	0 (0.0%)
	Wales	3 (27.3%)	13 (56.5%)	9 (27.3%)	0 (0.0%)	0 (0.0%)
	West Midlands	5 (16.1%)	6 (35.3%)	2 (3.0%)	0 (0.0%)	0 (0.0%)
	Yorkshire and the Humber	2 (7.7%)	0 (0.0%)	3 (6.3%)	3 (13.0%)	0 (0.0%)
	Total	**43 (12.0%)**	**53 (15.7%)**	**63 (8.2%)**	**18 (5.8%)**	**0 (0.0%)**

Patients registered with the NHD 2017/18

Haemophilia A includes females with VIII deficiency & haemophilia A carriers
Haemophilia B includes females with IX deficiency, haemophilia B carriers, FIX Leyden & FIX Leyden carriers

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