AHCDC Genotyping Reference laboratory: Annual report for 2020

Director: Dr. David Lillicrap

Co-Director: Dr. Paula James

Senior Clinical Scientists: Drs. Laura Swystun**,** Orla Rawley and Mackenzie Bowman

Technologists: Shawn Tinlin, Gina Jones, Christine Brown, Aomei Mo.

Introduction:

A central, reference variant testing laboratory was initiated at Queen's University in Kingston in November 2000, with funds from Health Canada. Testing in the laboratory is performed by two technologists: Gina Jones (MLT) and Shawn Tinlin. Two additional technicians Christine Brown and Aomei Mo joined the laboratory during Gina Jones' medical leave in 2020. The laboratory is located on the fourth floor of the Richardson Laboratory building in the Department of Pathology and Molecular Medicine at Queen's University.

The Laboratory is supervised by Dr. David Lillicrap and Dr. Paula James is the laboratory co-director. Since 2015, Dr. Laura Swystun has held the appointment of a Senior Clinical Scientist, and in 2017, Dr. Orla Rawley joined the laboratory as a Senior Clinical Scientist. Dr. Swystun began a leave of absence July 2019 with Dr. Rawley and Dr. Mackenzie Bowman filling in during her leave.

Facility Purpose:

The objective of this core AHCDC facility is to provide a national service for genetic analysis of inherited bleeding disorders. Almost all of this testing is for hemophilia A and B and von Willebrand disease, although infrequent testing has been performed for other less prevalent inherited bleeding disorders such as factors V, VII, X, XI, XII, XIII and fibrinogen deficiencies.

Methodologies:

Samples for analysis are sent to Kingston as genomic DNA or whole blood from which DNA is extracted. DNA is amplified using PCR and the amplified fragments are analyzed by DNA sequence analysis.

Report turnaround time:

When requested (eg. in instances of prenatal testing) and when the pedigree-specific variant is known, a result can be reported in 7-10 days. In most instances, results are reported to the referring clinic within 1-3 months. Reports for rare bleeding disorders are issued after 3-6 month periods.

Laboratory Accreditation and Licencing:

During 2020, as part of the requirements of accreditation status by the Institute of Quality Management in Healthcare (IQMH) (ISO 15189) the laboratory completed a self-assessment.

In June 2020, the laboratory's Ontario Ministry of Health and Long-Term Care license for genetic testing of the hemophilias and von Willebrand disease was renewed.

Laboratory Activity: January 1 to December 31, 2020:

Overall, referrals for genetic testing decreased in 2020 due to the onset of the COVID-19 pandemic. Turn-around times also temporarily increased due to the shutdown of Genome Quebec at the beginning of the pandemic.

Hemophilia A Referrals:
males and females **149**

Disease Severity

Severe FVIII deficiency	-	**17** cases
Moderate FVIII deficiency	-	**56** cases
Mild FVIII deficiency	-	**5** cases
Unknown	-	**49** cases
Carrier testing	-	**22** cases (unknown severity)

Referring Clinic (% of cases)

ON	**48.0**	%	BC	**10.2**	%
QC	**4.7**	%	NB	**2.0**	%
AB	**10.2**	%	NS	**10.2**	%
SK	**0.0**	%	NL	**10.9**	%
MB	**0.7**	%			

Hemophilia A reports generated: 112 (reported in 2020) – 34 (reported in 2021)

Hemophilia B Referrals:
males and females **29**

Disease Severity

Severe FIX deficiency	-	**2** cases
Moderate FIX deficiency	-	**6** cases
Mild FIX deficiency	-	**4** cases
Unknown	-	**2** cases
Carrier testing	-	**15** cases (unknown severity)

Referring Clinic (% of cases)

ON	**51.7**	%	NS	**6.9**	%
AB	**17.2**	%	NL	**0.0**	%
BC	**3.4**	%	MB	**3.4**	%
QC	**0.0**	%	SK	**3.4**	%
NB	**3.4**	%			

Hemophilia B reports generated: 21 (reported in 2020) – 7 (reported in 2021)

von Willebrand Disease Referrals:

Type 1 (incl 1C)	**4** cases
Type 2 (A,B,M)	**42** cases
Type 2N	**2** cases
Type 3	**2** cases
Unknown	**12** cases

Referring Clinic (% of cases)

ON	**58.1**	%	NS	**3.2**	%
AB	**14.5**	%	NL	**0.0**	%
BC	**11.3**	%	MB	**0.0**	%
QC	**3.2**	%	SK	**1.6**	%
NB	**4.8**	%			

von Willebrand Disease reports generated: 48 (reported in 2020) – 14 (reported in 2021)

Rare Bleeding Disorders Referrals:

Factor V	**3**	cases
Factor VII	**6**	cases
Factor X	**1**	case
Factor XI	**10**	cases
Factor XII	**0**	case
Factor VIII/V	**0**	case
Factor XIII	**3**	cases
Fibrinogen	**0**	cases
Gp1Ba	**1**	cases
ADAMTS 13	**2**	cases
Protein S	**1**	cases

Rare Bleeding Disorders reports generated: 27 (reported in 2020).