AHCDC Genotyping Reference laboratory: Annual report for 2019

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

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 and Shawn Tinlin. 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 2019, as part of the requirements of accreditation status by the Institute of Quality Management in Healthcare (IQMH) (ISO 15189) the laboratory completed an Internal Audit.

In June 2019, 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, 2019:

Hemophilia A Referrals:
males and females **178**

Disease Severity

Severe FVIII deficiency	-	**10** cases
Moderate FVIII deficiency	-	**29** cases
Mild FVIII deficiency	-	**6** cases
Unknown	-	**83** cases
Carrier testing	-	**50** cases (unknown severity)

Referring Clinic (% of cases)

ON	**61.2**	%	BC	**10.7**	%
QC	**5.1**	%	NB	**0.6**	%
AB	**11.2**	%	NS	**6.7**	%
SK	**1.1**	%	NL	**3.4**	%
MB	**0**	%			

Hemophilia A reports generated: 142 (total in 2019) – 110 (for samples submitted in 2019)

Hemophilia B Referrals:
males and females **36**

Disease Severity

Severe FIX deficiency	-	**4** cases
Moderate FIX deficiency	-	**6** cases
Mild FIX deficiency	-	**4** cases
Unknown	-	**12** cases
Carrier testing	-	**10** cases (unknown severity)

Referring Clinic (% of cases)

ON	**75**	%	NS	- **0**	%
AB	**14**	%	NL	- **2.7**	%
BC	**2.7**	%	MB	- **2.7**	%
QC	**0**	%	SK	- **2.7**	%
NB	**0**	%			

Hemophilia B reports generated: 29 (total in 2019)

von Willebrand Disease Referrals:

Type 1 (incl 1C)	**5** cases
Type 2 (A,B,M)	**64** cases
Type 2N	**24** cases
Type 3	**7** cases
Unknown	**12** cases

Referring Clinic (% of cases)

ON	**53.6**	%	NS	- **9.8**	%
AB	**16.1**	%	NL	- **0.9**	%
BC	**7.1**	%	MB	- **0.9**	%
QC	**3.6**	%	SK	- **3.6**	%
NB	**4.5**	%			

von Willebrand Disease reports generated: 125 (total in 2019) – 84 (for samples submitted in 2019)

Rare Bleeding Disorders Referrals:

Factor V	**4**	cases
Factor VII	**5**	cases
Factor X	**4**	case
Factor XI	**4**	cases
Factor XII	-	case
Factor VIII/V	-	case
Factor XIII	**-**	cases
Fibrinogen	**2**	cases
Gp1Ba	**1**	cases

Rare Bleeding Disorders reports generated: 24 (total in 2019) – 19 (for samples submitted in 2019)