AHCDC Genotyping Reference laboratory: Annual report 2016

Director: Dr. David Lillicrap

Co-Director: Dr. Paula James

Senior Clinical Scientist: Dr. Laura Swystun

Technologists: Gina Jones and Shawn Tinlin

Introduction:

A central, reference mutation 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. In 2015, a new appointment was made to the laboratory. Dr. Laura Swystun has joined the laboratory as a Senior Clinical Scientist. Dr. Swystun's initial role involves developing plans for the formal accreditation of the laboratory in 2016, and formulating a strategy for the introduction of new next generation sequencing strategies for the laboratory.

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 and XIII 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 mutation 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. Since Jayne Leggo's retirement in November 2016 report turnaround times have been extended, although STAT samples can still be processed. We are in the process of training a new MLT, Gina Jones and we anticipate that once Gina's training is complete our turnaround times will return to regular rates.

Laboratory Accreditation:

During 2015, a major initiative has been underway, supervised by Dr. Laura Swystun, to prepare the laboratory for a formal accreditation review by the Institute of Quality Management in Healthcare (IQMH). IQMH is internationally recognized as a competent provider of ISO 15189 accreditations, a mark of excellence acknowledged by the global healthcare community. The laboratory is currently in the process of applying for a lab licence under the Ministry of Health and Long Term Care. Once the lab licence application is granted our accreditation application with IQMH will proceed.

Laboratory Activity: January 1 to December 31, 2016:

Hemophilia A Referrals: 172

87 males and 85 females

Disease Severity

Severe FVIII deficiency	37	cases
Moderate FVIII deficiency	50	cases
Mild FVIII deficiency	16	cases
Unknown	5	cases
Carrier testing	65	cases (59 unknown severity)

Referring Clinic (% of cases)

ON	46%	BC	11.4%
QC	19.6%	MB	5.6%
AB	8.7%	NS	8.1%
NL	0.5%		

Hemophilia A reports generated: 143

Hemophilia B Referrals: 35

10 males and 25 females

Disease Severity

Severe FIX deficiency	4	cases
Moderate FIX deficiency	9	cases
Mild FIX deficiency	5	cases
Unknown	4	cases
Carrier testing	13	cases (13 unknown severity)

Referring Clinic (% of cases)

ON	71%	NS	8.6%
AB	8.6%	NL	1%
BC	2.9%		
QC	5.7%		

Hemophilia B reports generated: 32

von Willebrand Disease Referrals: 74

Type 1 (incl 1C)	5 cases
Type 2 (A,B,M)	34 cases
Type 2N	11 cases
Type 3	3 cases
Unknown	21 cases

Referring Clinic (% of cases)

ON	45%
NS	2.7%
AB	30%
QC	5.4%
NB	1.3%
BC	12%
MB	5.4%
NL	1.3%
SK	1.3%

von Willebrand Disease reports generated: 70

Rare Bleeding Disorders Referrals (2006-2016): 36

Factor V	11 cases
Factor VII	6 cases
Factor X	1 case
Factor XI	14 cases
Factor XII	1 case
Factor VIII/V	1 case
Factor XIII	2 cases

Rare Bleeding Disorders reports generated: 32