AHCDC Genotyping Reference laboratory: Annual report 2017

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 times:

When requested (eg. in instances of prenatal testing) and when the pedigree-specific mutation is known, a result can be reported in two weeks. In most instances, results are reported to the referring clinic within 1-3 months. For female carrier testing without clear a known familial history, turnaround times can be extended (3-6 months), although a preliminary report may be issued where appropriate.

Laboratory Accreditation and Licencing:

Since 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.

Our IQMH assessment visit took place March 22/23 2018. The lab was assessed on 335 requirements, and received only 5 minor non-conformance findings, a major achievement. The lab is currently performing corrective actions, and we expect our IQMH accreditation certificate to be awarded within the next 6 months.

The lab has also received provisional licencing by the Ontario Ministry of Health and Long Term Care starting January 1, 2018. A full licence will be granted in conjunction with the receipt of the IQMH accreditation certificate. A copy of our provisional licence can be found on our website at www.nibdgl.ca

Laboratory Activity: January 1 to December 31, 2017:

Hemophilia A Referrals: 164

91 males and 71 females (carrier testing)

Referring Clinic (% of cases)

ON	47%
BC	4.3%
QC	20%
MB	5.0%
AB	10%
NS	8.1%
NL	1.2%

Hemophilia A reports generated: 116

Hemophilia B Referrals: 40

25 males and 15 females

Referring Clinic (% of cases)

ON	47.5%
NS	5.0%
QC	15.0%
BC	10.0%
AB	5.0%

Hemophilia B reports generated: 35

von Willebrand Disease Referrals: 80

Type 1 (incl 1C)	13 cases
Type 2 (A,B,M)	55 cases
Type 2N	7 cases
Type 3	3 cases
Unknown	9 cases

Referring Clinic (% of cases)

ON	45%
NS	9.3%
AB	19%
QC	4%
NB	4%
BC	9.3%
MB	9.3%
NL	1.3%
SK	1.3%

von Willebrand Disease reports generated: 67

Rare Bleeding Disorders Referrals (2006-2017): 46

Factor V	11 cases
Factor VII	12 cases
Factor X	1 case
Factor XI	18 cases
Factor XII	1 case
Factor VIII/V	1 case
Factor XIII	2 cases

Rare Bleeding Disorders reports generated: 44