AHCDC Genotyping Reference laboratory: Annual report 2013

Director: David Lillicrap **Technologists:** Jayne Leggo 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: Jayne Leggo 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.

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 and XII 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 (e.g. 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.

Laboratory Activity: January 1 to December 31, 2013:

Hemophilia A Referrals: 210

99 males and 111 females

Disease Severity

Severe FVIII deficiency	14 cases
Moderate FVIII deficiency	8 cases
Mild FVIII deficiency	61 cases
Unknown	16 cases
Carrier testing	111 cases

Reverse pages

Referring Clinic (% of cases)

ON	39%	BC	11%
QC	15%	NB	6%
AB	16%	MB	5%
NS	8%		

Hemophilia A reports generated: 221

Hemophilia B Referrals: 35

15 males and 20 females

Disease Severity

Severe FIX deficiency	3 cases
Moderate FIX deficiency	7 cases
Mild FIX deficiency	4 cases
Unknown	1 case
Carrier testing	20 cases

Referring Clinic (% of cases)

ON	39%	NB	6%
AB	34%	NL	6%
BC	9%		
QC	6%		

Hemophilia B reports generated: 31

von Willebrand Disease Referrals: 117

Type 1 (incl. 1C)	6 cases
Type 2 (A, B,M)	64 cases
Type 2N	25 cases
Type 3	10 cases
Unknown	12 cases

Referring Clinic (% of cases)

ON	34%
NS	10%
AB	20%
QC	18%
NB	3%
BC	12%
MB	2%
SK	1%

von Willebrand Disease reports generated: 110

Rare Bleeding Disorders Referrals (2006-2013): 11

Factor V	5 cases
Factor VII	1 case
Factor X	1 case
Factor XI	2 cases
Factor XII	1 case
Factor VIII/V	1 case

Rare Bleeding Disorders reports generated: 10