



AHCDC Genotyping Reference Laboratory

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. The testing in the laboratory is performed by two technologists: Jayne Leggo and Shawn Tinlin. The laboratory is located on the second 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.

Methodologies:

Samples for analysis are sent to Kingston as either whole blood or genomic DNA. Extracted DNA is amplified by PCR and the amplified fragments are analyzed by either a heteroduplex screening method or directly by DNA sequence analysis. All heteroduplex-positive PCR fragments are sequenced to identify the primary sequence changes.

Reporting Turn around times:

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 in 1-3 months. With subtle mutations that require extensive sequencing, results are reported in several months.

Laboratory Activity. January 1st – December 31st 2008:

Hemophilia A Referrals: 175

83 males and 92 females

Disease Severity

Severe FVIII deficiency	22 cases
Moderate FVIII deficiency	4 cases
Mild deficiency	70 cases
Unknown	79 cases

Referring Clinic (% of cases)

Ontario	31%
Alberta	21%
BC	15%
Quebec	13%

Nova Scotia 13%
Manitoba 3%
Saskatch 3%
Newfound 1%

Hemophilia A Reports Generated - 168

Hemophilia B Referrals: 44

18 males and 26 females

Disease Severity

Severe FIX deficiency 5 cases
Moderate FIX deficiency 5 cases
Mild FIX deficiency 10 cases
Unknown 24 cases

Referring Clinic (% of cases)

Ontario 34%
Nova Scotia 25%
BC 16%
Alberta 7%
Quebec 7%
Manitoba 7%

Hemophilia B Reports Generated - 47

Von Willebrand Disease Referrals: 49

Type 1 3
Type 2A 1
Type 2B 7
Type 2M 6
Type 2N 21
Type 3 3
Unknown 8

Referring Clinic (% of cases)

Ontario 35%
BC 24%
Alberta 16%
Nova Scotia 12%
Quebec 6%
Saskatch 6%

von Willebrand Disease Reports Generated - 32