

AHCDC Genotyping Reference Laboratory: Annual Report 2009

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 2009:

Hemophilia A Referrals: 242

140 males and 102 females

Disease Severity

Severe FVIII deficiency	26 cases
Moderate FVIII deficiency	16 cases
Mild deficiency	116 cases
Unknown	84 cases

Referring Clinic (% of cases)

Ontario	26%
Alberta	12%
BC	17%
Quebec	19%
Nova Scotia	15%

Manitoba 8%
Saskatch 3%
Newfound 0.4%
New Brunswick 0.4%

Hemophilia A Reports Generated - 261

Hemophilia B Referrals: 43

25 males and 18 females

Disease Severity

Severe FIX deficiency 7 cases
Moderate FIX deficiency 9 cases
Mild FIX deficiency 17 cases
Unknown 10 cases

Referring Clinic (% of cases)

Ontario 39%
Nova Scotia 7%
BC 30%
Alberta 14%
Quebec 7%
Manitoba 3%

Hemophilia B Reports Generated - 40

Von Willebrand Disease Referrals: 82

Type 1 6
Type 2A 4
Type 2B 16
Type 2M 3
Type 2N 29
Type 3 2
Unknown 22

Referring Clinic (% of cases)

Ontario 55%
BC 13%
Alberta 12%
Nova Scotia 4%
Quebec 4%
Saskatch 3%
Manitoba 3%
Nfld 2%

von Willebrand Disease Reports Generated - 81