

AHCDC Genotyping Reference Laboratory: Annual Report for 2023

Laboratory Director: Dr. David Lillicrap

Laboratory Co-Director: Dr. Harriet Feilotter

Senior Clinical Scientist: Dr. Mackenzie Bowman

Technologists: Gina Jones (MLT), Samira Kheitan, Aomei Mo

Clinical Administrative Assistant: Julie Grabell

Introduction:

A central, reference variant 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 (MLT) and Samira Kheitan. An additional technician, Aomei Mo provides part-time support to the lab. 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. Dr. Harriet Feilotter serves as the Laboratory Co-Director. In 2023, Dr. Mackenzie Bowman resumed a part-time role as Senior Clinical Scientist. Drs. Laura Swystun and Orla Rawley left their role as Senior Clinical Scientists and Queen's University to pursue other professional endeavours. Ms. Julie Grabell has assumed a part-time role to support Dr. Bowman and the lab.

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 testing has also been performed for other less prevalent inherited bleeding disorders such as factors V, VII, X, XI, XII, XIII and fibrinogen 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. *F8* Intron 22 analysis by inverse PCR began in the lab in December 2020.

Report turnaround time:

When requested (eg. in instances of prenatal testing) and when the pedigree-specific variant 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. Reports for rare bleeding disorders are issued after 3-6 month periods.

Laboratory Accreditation and Licencing:

During February 2022, the laboratory underwent a full accreditation assessment by Accreditation Diagnostics Canada (formerly IQMH, Institute of Quality Management in Healthcare) which provides laboratory accreditation to ISO 15189 standards. A four-year renewal of the lab's accreditation certificate was granted on May 24th 2022.

On July 1st 2022, the laboratory's Ontario Ministry of Health and Long-Term Care license for genetic testing for hemophilia A, hemophilia B and von Willebrand disease was renewed for a two-year period.

Laboratory Activity: January 1 to December 31, 2023

Hemophilia A Referrals: 225

- Affected males : 101 cases
- Carrier testing : 123 cases

Disease Severity

- Severe F8 deficiency : 22 cases
- Moderate F8 deficiency : 12 cases
- Mild F8 deficiency : 47 cases
- Unknown : 144 cases

Referring Clinic (% of cases)

- ON 58.6 %
- BC 12 %
- AB 10 %
- NS 8 %
- NB 4 %
- QC 3.5 %
- MB 0.8 %
- SK 0.4 %
- NL 0 %
- Others: 3.5 %

Hemophilia A reports generated: 224 cases in 2023

Hemophilia B Referrals: 43

- Affected males : 17 cases
- Carrier testing : 26 cases

Disease Severity

- Severe F9 deficiency : 3 cases
- Moderate F9 deficiency : 3 cases
- Mild F9 deficiency : 1 cases
- Unknown : 36 cases

Referring Clinic (% of cases)

- ON 53.6 %
- NS 6.9 %
- BC 23.4 %
- AB 6.9 %
- MB 4.6 %
- NB 0 %
- NL 0 %
- QC 0 %
- SK 2.3 %
- Others: 2.3 %

Hemophilia B reports generated: 40 cases reported in 2023

von Willebrand Disease Referrals: 118

- Type 1 (incl 1C) : 7 cases
- Type 2 (A,B,M) : 83 cases
- Type 2N : 11 cases
- Type 3 : 8 cases
- Unknown : 8 cases

Referring Clinic (% of cases)

- ON 41.5 %
- AB 16.1 %
- NS 9.3 %
- BC 14.4 %
- NB 7.6 %
- MB 4.2 %
- QC 5.9 %
- NL %
- SK %
- Others: 1%

von Willebrand Disease reports generated: 123 cases reported in 2023

Rare Bleeding Disorders Referrals: 35

- Factor V: 1 case
- Factor VII: 15 cases
- Factor X: 2 cases
- Factor XI: 11 cases
- Factor XII: 0 cases
- Factor XIII: 4 cases
- Factor VIII/V: 0 cases
- Fibrinogen: 1 case
- ADAMTS 13: 2 cases
- GPIIb/IIIa: 0 cases
- Protein S: 0 cases
- Protein C: 0 cases

Rare Bleeding Disorders reports generated: 31 cases reported in 2023