Lab Use Only

Res Lab #: _____

Date Rec'd: _____

Initials: _____

National Inherited Bleeding Disorder Genotyping Laboratory

Department of Pathology and Molecular Medicine
Queen's University, Kingston, Ontario

AHCDC

von Willebrand Disease Genotype Testing Requisition

Patient Name: _____ **Sex:** Male ☐ Female ☐
(Surname, First Name)

DOB: ____/____/____ **Unique Identifier:** _____ **CBDR#:** _____
YY MM DD eg. Health card #, Hospital #

Date of specimen collection: ____/____/____ **Phlebotomist:** _____
YY MM DD

Referring Clinic: _____ **Report to:** _____

Von Willebrand Disease: ☐ **Type 2**_____ ☐ **Type 3** ☐ **Type 1C**
(subtype if known)

Testing: VWF:Ag _____ IU/mL

☐ **VWF:RCo** ☐ **VWF:G1bM** ☐ **Other**_____ **Value** _____ IU/mL
(Indicate test performed)

Factor VIII:C _____ IU/mL

Multimers _____

Desmopressin trial _____

Have samples from this family been sent to this lab before? Yes ☐ No ☐

If Yes, specify _____

Relationship to this patient _____

Sample Requirements:

6 cc whole blood

EDTA (lavender top) or

ACD (yellow top) or

DNA

Ship to:

Attn: Gina Jones/Samira Kheitan
Department of Pathology and Molecular Medicine
Queen's University, Richardson Laboratory, Room 201
88 Stuart St., Kingston, Ontario K7L 3N6
Tel: 613-533-3187 FAX: 613-533-2907
Email: NIBDGL@queensu.ca

PATIENT INFORMATION AND CONSENT FORM

Molecular Hemostasis Laboratory-Genotyping Research

Principal Investigators:

Dr. David Lillicrap, MD, FRCPC
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Queen's University, Kingston, ON
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Background Information: You are invited to participate in a research study. Your Doctor has requested your sample(s) (or your child's/ward's) be tested in the National Inherited Bleeding Disorders Genotyping Lab (NIBDGL). We are requesting your permission to retain the sample in the NIBDGL for future research use in genetic analyses for inherited bleeding disorders. The request for storage and use of the sample for future research has been reviewed for ethical compliance by the Queen's University Health Sciences and Affiliated Teaching Hospitals Research Ethics Board.

Description of the Research: If you are interested in participating, we ask you give us permission to store your (or your child's/ward's) de-identified sample indefinitely once clinical testing has been completed. Your sample will be stored in a secure lab following the clinical testing for use in potential future research opportunities and will become the property of the study investigator.

Risks: If you are a First Nations person, or an indigenous person and you have contact with spiritual "elders" you may want to talk with them before you agree to participate. Elders have reservations about genetic research.

When you donate your blood or tissue for research, you are sharing genetic information, not only about yourself, but also about biological (blood) relatives - information that could eventually be linked to you. Due to the rapid pace of technological advances, the potential future use (and potential future risks) of genetic information such as loss of privacy and financial, social, emotional or discrimination risks is unknown.

Benefits: It is unlikely that you will benefit directly from this research. However, future studies on inherited bleeding disorders will improve our knowledge of the implications of inherited bleeding disorders.

Inclusion/Exclusion: Any individual who has had a sample sent by their doctor for testing is eligible to have their sample retained for research purposes.

Confidentiality:

All persons associated with this study, including study investigators, coordinators, nurses and delegates (hereby referred to as "study personnel") are committed to respecting your (or your child's/ward's) privacy. No persons, other than those listed in this consent form will have access to your (or your child's/ward's) personal health information or other identifying personal information without your consent, unless required by law. Any personal health information collected from your (or your child's/ward's) medical records, or other information related to you (or your child/ward) will be coded by study ID codes (i.e. be "de-identified"). This means the investigator and study personnel will assign a unique identifier to each participant instead of the subject's name in study documentation. A master linking log with your (or your child's/ward's) medical record number will be kept and stored separately from the data, it will be password protected and stored on an encrypted computer. Only the investigators will have access to the raw data. The data will be maintained indefinitely and will be accessible to the principle investigator and co-investigator. The study personnel here at Queen's University are in control of the study code key, which is needed to connect your personal health information to you (or your child/ward). Our guidelines include the following:

All information that identifies you (or your child/ward), both paper copy and electronic information, will be kept confidential and stored and locked in a secure place that only the study personnel will be able to regularly access.

- Electronic files will be stored securely on hospital or institutional networks or securely on any encrypted portable electronic devices.
- No information that can directly identify you (or your child/ward) will be allowed off site in any form.

By signing this form, you agree to allow the study doctor and his/her staff to collect and use personal data about you (or your child/ward) for the study. This information may include: your (or your child's/ward's) month and year of birth, sex, and personal data on your (or your child's/ward's) physical condition. All information collected will be kept strictly confidential and will only be used for purposes of the study as described in this consent form. Your consent to the use of this information will not expire, but you may withdraw your consent at any time by telling your study doctor. If you withdraw your consent to collect and use your personal information, you may not be permitted to continue to participate in the study and any information that has already been collected will continue to be used as described in this consent form, however, no new information will be collected.

Your (or your child's/ward's) records may be reviewed by:

- Queen's University Health Sciences and Affiliated Teaching Hospitals Research Ethics Board. The REB is a group of scientists and non-scientists who review the ethics of research. The goal of the REB is to protect the rights and welfare of study participants.

These people may look at your (or your child's/ward's) records to make sure the study has been done the right way. They also want to make sure that your (or your child's/ward's) health information has been collected the right way, or for other reasons that are allowed under the law. No information identifying you (or your child/ward) will be transferred outside the site of this study.

The results of the research will include information from many people grouped together so that no one person can be identified. Any responses, records or personal information that could be directly linked to you (or your child/ward) will not be reported or shared with anyone outside of the study team. If information about this study is published, you (or your child/ward) will not be identified.

It is important to understand that despite these protections being in place, there continues to be the risk of unintentional release of information. The study personnel will protect your (or your child's/ward's) records and take all reasonable steps to keep all the information in your (or your child's/ward's) study file confidential. The chance that this information will be accidentally released is small.

Federal and Provincial data protection laws, including the federal Personal Information Protection and Electronic Documents Act, 2000 (PIPEDA) and Ontario's Personal Health Information Protection Act, 2004 (PHIPA), protect your (or your child's/ward's) personal information. They also give you rights to control the use and disclosure of

your (or your child's/ward's) personal information (including personal health information) and require your written permission for this personal information to be collected, used, or disclosed for the purposes of this study, as described in this consent form. You have the right to review and copy your (or your child's/ward's) personal information collected in this study. However, if you decide to be in this study or choose to withdraw from it, your right to look at or copy your personal information related to this study may be delayed until the study is complete.

Voluntary Nature of Participation: Your participation is voluntary. The genetic testing ordered by your doctor will still be performed even **if you do not wish for your sample to be used for research and your decision will have no impact/effect on your current or future health care.** Additionally, you may withdraw at any time and your withdrawal will not affect you or your families' future medical care with your Hematologist or at this hospital. If at any time you choose to withdraw from this study, please contact a member of the study team.
If you choose to no longer take part in the study, the information which has been collected to date will be used in analysis. No further study procedures will be completed and no further information will be collected.

Study Contacts
If at any time during the study you have questions about the study or the study procedures, you should contact the study doctor, Dr. David Lillicrap at (613) 548-1304 (9am-5pm), or contact the research coordinator, Julie Grabell, at (613) 533-6000 ext. 75223 (9am-5pm).

If you have any questions about your rights as a research participant or about ethical issues related to this study, you may contact the Queen's University Health Sciences and Affiliated Teaching Hospitals Research Ethics Board at 1-844-535-2988 during business hours (9am to 5pm) or email HSREB@queensu.ca.

Consent:
I acknowledge that the research study described above has been explained to me and that any questions that I have asked have been answered to my satisfaction. I have been informed of the alternatives to participation in this study, including the right not to participate and the right to withdraw without compromising the quality of medical care for me and for other members of my family. As well, the potential risks, harms and discomforts have been explained to me and I have also been informed of the benefits (if any) of participating in the research study.

It has been explained that I have not waived my legal rights nor released the investigators, sponsors, or involved institutions from their legal and professional duties. I know that I may ask now, or in the future, any questions I have about the study or the research procedures. I have been assured that records relating to me and my care will be kept confidential and that no information will be released or printed that would disclose personal identity without my permission unless required by law. I authorize access to my medical records (or my child's/ward's) as explained in this consent form and I have been given sufficient time to read and understand the above information.

I hereby consent to participate, and will be given a copy of this consent form for my records.

***Note: If you have any questions about the study, you will have a chance to talk to one of the study staff or your regular doctor. Do not sign this form unless you have had the chance to ask questions and have received satisfactory answers.**

I agree that the study investigators can store my (or my child's/ward's) sample(s) for use in future research on inherited bleeding disorders

YES **NO** _____ / _____
Initials Date

_____ _____
Name of Participant (print) Name of person obtaining consent (print)

_____ _____
Name of Person giving consent Signature

_____ _____
Relationship to Participant Date

_____
Signature

Children's Assent Form: Molecular Hemostasis Laboratory-Genotyping Research

I understand that I am being tested for an inherited bleeding disorder. This means that Doctor _____ has explained to me that this is being done to help understand the effects this disease has, or might have, on me.

I understand that it is up to me whether I want my sample to be used in research or not and that it is OK if I decide I do not want to. I also understand that even if I agree today I can change my mind later.

I understand that while the research may not help me directly, it may help other people with the same disease.

I have had a chance to think about my sample being used in research, and have had my questions answered. If I think of any questions in the future, I can discuss them with Drs. Lillicrap and/or James.

I understand that all of the information that is discovered from this research will be kept private.

I agree to be my sample being used for research:

YES **NO** _____ / _____
Initials Date

_____
Name of Child

_____ _____
Signature of Child Date

_____ _____
Signature of Parent/Guardian Date

National Inherited Bleeding Disorder Genotyping Lab
Sample Collection Instructions

Requisition and Consent Forms:

1. Samples must be accompanied by a completed requisition form.
2. Submitted patients must have a documented rationale for testing:
   - For affected hemophilia A or B patients, a clotting factor activity level must be provided.
   - For VWD patients, VWF:Ag, VWF activity (specify test used), FVIII:C, and multimer information must be provided.
   - For carrier testing, a documented family history or a coagulation factor level must be provided.
   - For carrier testing, information on the family variant if available must be provided.
   - For prenatal testing, information on the family variant if available must be provided.
3. For queries regarding carrier testing of women under 16 years of age, please contact lab staff at NIBDGL@queensu.ca
4. Incomplete requisition forms will result in delayed sample testing.

Sample Collection and Shipment:

1. Samples acceptable for testing:
   - Venous whole blood (minimum 6 cc) collected into EDTA (lavender top) or ACD (yellow top) evacuated tubes
   - Expired tubes should not be used.
   - If blood is being drawn from an intravenous line for laboratory testing, two times the dead-space volume should be discarded.
   - When drawing blood specimens for several examinations during a single venipuncture, the "order of draw" shall be: (1) blood culture; (2) coagulation specimens; (3) serum tube with or without clot activator or gel; (4) heparin; (5) EDTA; (6) glycolytic inhibitor.
   - DNA (minimum of 15 µg at 150 ng/µL); for some cases smaller samples are acceptable.
   - Patient ID must be verified and samples labelled using two unique identifiers.
   - Samples shall be collected using routine practices/standard precautions.
   - Materials for sample collection shall be safely disposed of according to institutional protocols.
2. Ship packages on a Monday, Tuesday, Wednesday, Thursday as follows:
   - Shipping temperature: DNA (ambient), whole blood (cold packs), whole blood frozen (dry ice).
   - Place the samples in sealable plastic bags with absorbent material.
   - Include completed requisition and consent form.
   - Ship overnight. Contact courier for complete shipping instructions.
3. Attach the following labels to the outside of the box/package:
   - Dry ice label (if applicable).
   - Return address label (including the contact name and telephone number).
4. Ship to: **Attn: Gina Jones/Samira Kheitan**
   Department of Pathology and Molecular Medicine
   Queen's University, Richardson Laboratory, Room 201
   88 Stuart Street, Kingston, Ontario K7L 3N6
   Tel: 613-533-3187 FAX: 613-533-2907 NIBDGL@queensu.ca

Results:

- Turnaround time is approximately 3 months from the time of sample submission but may be longer for rare genes or for carrier testing if familial variant is unknown.
- If the family-specific variant is known, urgent reporting can be completed within several weeks. Please indicate this on the requisition form.
- For prenatal testing, maternal cell contamination studies are the responsibility of the referring clinic.

- Results will be reported via Canadian Bleeding Disorders Registry (CBDR). If a patient has not consented to their information being entered into CBDR, a paper copy of the report will be sent to the referring clinician listed on the requisition form.