

# **National Inherited Bleeding Disorder Genotyping Laboratory (NIBDGL)**

## **Routine Genotyping Consent:**

Since the establishment of the NIBDGL, 24 yrs ago, we have requested patient consent to proceed with genotype analysis. This was required, at least in part, because the laboratory was not a licensed or accredited facility and thus all testing was regarded as “research”.

However, since 2018, the laboratory has been fully licensed by the Ontario Ministry of Health and Long-Term Care, and fully accredited by Accreditation Canada Diagnostics.

With this as background, we have been reconsidering a renewal of our institutional consent document that we ask our clients to complete. On review, we see that this consent is entirely focused on the use of samples for research purposes, either in the short-term or on long-term banked samples. However, all our referred-in samples are received for diagnostic testing, and when samples are banked this is only for the potential benefit of later diagnostic analysis. Samples have never been used for research purposes, and we have no plans for future research projects involving genotyping.

**Based on this knowledge, effective immediately, we will no longer require the submission of a completed consent form when samples are sent for diagnostic genotyping at the NIBDGL.**

If anyone has plans for research use of the genetic information generated by the NIBDGL they will need to generate their own consent protocol to cover the ethics requirements of their specific research process.

## **Genotypic Testing in Minor Females in Families with X-linked Recessive Traits:**

As everyone is aware, the status of females who are heterozygous for variant alleles of X-linked recessive traits (ie. hemophilia A and B) is an important issue that has been overlooked in past decades. Now, with the recognition that ~30% of these individuals manifest some form of bleeding, some form of testing (either factor level evaluation or genotyping) should be considered to evaluate this clinical problem.

In the past, we have requested the completion of a document confirming that an appropriate discussion has occurred between the medical team and the parent(s) of an underage/minor female who are requesting genetic analysis. If this discussion has occurred, we will proceed with the genotyping as requested.

In recent discussion with our local clinical genetic group, it seems that this issue has now been resolved in the genetics' community, and aside from documentation in the minor's medical record no additional record of consent is required.

**As a result of this recent consultation, we will no longer be requesting supplementary documentation to enable genotyping testing in minors who may be heterozygotes for recessive bleeding disorders.**

**Other NIBDGL updates:**

We have a new fax number: 343-344-2733

Our requisitions have been updated and are available on our website:  
<https://www.nibdgl.ca/forms>