Sample Collection in Canada

LifeLabs Genetics is proud to offer equal access to genetic testing across Canada.

**Ontario, Saskatchewan, and British Columbia**
Find the closest LifeLabs collection centre at http://locations.lifelabs.com
Book an appointment online at www.lifelabs.com or by calling either 1-877-849-3637 (ON) or 1-888-333-0222 (SK) or 1-800-431-7206 (BC)

**Quebec**
Please contact Biron at 1-800-463-7674
Veuillez communiquer avec le numéro ci-haut
If you do not have access to a Biron collection centre, please contact us for other arrangements

**Other Provinces and Territories**
Please contact us to be directed to the closest Innomar collection centre

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**Accepted Sample Types**

- **EDTA tube**
  - Sample size: 2-8mL
  - For newborns and small children, 2ml is sufficient
  - Please note: when ordering more than one panel, please contact us for confirmation of the required volume of blood to be submitted.

- **Amniotic fluid**
  - Sample size: 10mL
  - Clear – no blood contamination
  - Maternal Sample: 1mL EDTA blood or 1ug purified DNA for maternal contamination analyses

- **Purified DNA**
  - Sample size: 5-10µg
  - We accept purified DNA with a ratio of absorbance at 260 and 280nm (A260/280) in the range from 1.6 - 2.1.

- **Filtercard**
  - Sample size: 50µL of EDTA blood per circle (circles must be completely saturated)
  - 10-20 filled in circles, depending on the type of analysis ordered
  - Maternal Sample: 1mL EDTA blood or 1ug purified DNA for maternal contamination analyses

- **Tissue sample**
  - Sample size: 2 x T25 flasks of amniocyte or chronic villi cultures near confluence and sterile
  - Cultures must be topped off with sterile medium immediately prior to shipping
  - Maternal Sample: 1mL EDTA blood or 1ug purified DNA for maternal contamination analyses

- **Saliva**
  - Sample size: 3-5mL
  - Please note: when ordering more than one panel, please contact us for confirmation of the required volume of blood to be submitted.

All frozen or prenatal samples should be sent directly to our sequencing partner, Centogene. Please contact us at Ask.Genetics@LifeLabs.com for more information.
Fixed Panels

Ordering instructions

1. Use the LifeLabsGenetics.com search tool to identify the panel(s) that best represents the patient’s clinical indications and download the hereditary testing requisition form from the Healthcare Providers tab.

2. The Ordering Checklist on page 1 can be used to ensure you’ve completed all the necessary pages of the requisition.

3. In the Testing for Single Gene(s) or Fixed Panel(s) section on page 2 of the requisition form, write the panel name(s) and test code(s) found on the LifeLabs Genetics website and select the methodologies (sequencing, deletion/duplication, etc.).

4. You are only required to complete the first page of the Informed Consent.

For all testing, we strongly encourage you to provide the patient’s in-depth medical history to ensure the most comprehensive analysis and interpretation of the genetic testing results. This can be done through any of the following three ways:

- Writing in the Relevant Medical and Family History section on page 1
- Completing the checkboxes on page 5
- Providing the patient’s hospital charts
**CONTRACT #**  LL: K012-01

**Report to Physician #**
- Physician OHIP# (Ontario):
- Physician MSC# (British Columbia):
- Other Provinces: 999

**Ordering Physician Name**

**Ordering Physician Address & contact info:**
- Address
- Tel:
- Fax:

**Physician Signature:**

**Confirmation of Patient Consent:** I confirm that this patient has given consent to testing as may be required by applicable law, which indicates that: the patient has been informed about the details associated with the genetic test(s) ordered below including its risks, benefits and limitations; we will ensure that test results will be interpreted to the patient in an appropriate manner, and that the patient will not receive the results without accompanying counseling; and the patient was informed that she has the right to revoke her consent at any time.

**Physician Signature:**

**Copy to:**
- Genetic Counselor
- Other Healthcare Provider
- Copy to name
- Tel:   Fax:

**Bill to:**
- Contract # K012-01 (patient does not pay at time of collection)
- Patient Sex:  
  - Female
  - Male

**Patient Name (Last, First):**

**Patient Address:**

**Health Card #:**

**Patient Tel:**

**Patient Information:**
- African/African American
- Caucasian
- French Canadian or Acadian
- Middle Eastern
- Northern European e.g. British, German
- South Asian e.g. Indian, Pakistani
- East Asian e.g. Chinese, Japanese
- Ashkenazi Jewish
- Other/Mixed Caucasian
- Native American
- Hispanic
- Southern European e.g. Italian, Greek
- Southeast Asian e.g. Filipino, Vietnamese
- Pacific Islander

**Relevant Medical and Family History**
- No additional information available

**Billing Status**
- Ministry of Health Approved (Approval letter attached)
- Ministry of Health Approval Pending
- Institution (Complete information below)
- Private Pay (Complete additional form)

**Institution Billing ONLY**
- Institution Name: ___________________________
- Contact Name: ___________________________
- Address:
  - Phone: (    )
  - Fax: (    )
- Email: _______________________________

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**For samples not collected at a LifeLabs location, please ship all NON-PRENATAL samples to:**

- British Columbia: LifeLabs • Attn. Specimen Management • 3680 Gilmore Way • Bumaby BC • V5G 4V8
- All Other Provinces: LifeLabs • Attn. Specimen Management • 37 Voyager Court N. • Toronto ON • M9W 6J2

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**Ordering Checklist**
- Sample Type
  - Known variant
    - Single gene
    - Fx Panels
    - Ex Panel
    - ProGx Panels
    - Whole Exome Sequencing (WES)
    - Whole Genome Sequencing (WGS)
  - Must complete pages 1, 2, & 3
    - Physician, patient, & test information (p1-2)
    - Informed consent (p3)
  - Must complete pages 1-5
    - Physician, patient, & test information (p1-2)
    - Informed consent (p3-4)
    - Clinical features checklist (p5)
  - Must complete pages 1-8 (if applicable)
    - Physician, patient, & test information (p1-2)
    - Informed consent (p3-4)
    - Clinical features checklist (p5)
    - Parental 1 & 2 requisitions (p6-7)
  - Additional Family Member requisition (p8)
  - IL TC
  - Mnemonic
    - Blood-Adult (2 x 4ml EDTA)
    - Blood-Pediatric (1 x 2ml EDTA)
    - Purified DNA (single genes 1-10ug, Panels: 10-100ug)
    - Filter card *
    - Other:** _________

**Available by request. Please contact LifeLabs Genetics.**

**Other sample types are permitted. Please contact LifeLabs Genetics for details.**

Please contact LifeLabs Genetics before shipping prenatal samples. Samples should be shipped directly to Centogene.

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**Date Sample Collected:**

**Time Collected:**

**Collector Name:**
<table>
<thead>
<tr>
<th><strong>Patient Name:</strong></th>
<th><strong>Patient DOB (MM/DD/YYYY):</strong></th>
</tr>
</thead>
</table>

### Testing for known variants:

<table>
<thead>
<tr>
<th>Gene: ___________________________</th>
<th>Mutation (HGVS): ____________</th>
</tr>
</thead>
</table>

Familial Report attached  □ Yes  □ No

### Testing for Single Gene(s) or Fixed Panel(s):

Please use the online catalogue to find test code & names [www.lifelabsgenetics.com/hereditary-conditions](http://www.lifelabsgenetics.com/hereditary-conditions)

**Test Code(s):**

**Test Name(s):**

#### Single Genes
- □ FULL analysis (by NGS Panel Plus+CNV)
- □ Sequencing (by NGS Panel Plus)
- □ Deletion/Duplication Testing
- □ Repeat Expansion

*Depending on coverage optimization, sequencing may be performed via Sanger ("Full Sequencing"), if NGS Panel Plus is unavailable. Similarly, CNV analysis may be performed by MPA of qPCR ("Deletion/Duplication Testing"), if the option of "+CNV" is unavailable.*

### Expanded Panel

Please contact LifeLabs Genetics if you require a Reference Number for your request.

**Test Code(s) / Reference Number(s):**

**Test Name(s):**

- □ Sequencing + Deletion/Duplication (by CentoDxPlus + CNV)
- □ Sequencing only (by CentoDxPlus)

### Progressive Panels

Please contact LifeLabs Genetics to receive a Reference Number for your request.

**Test Code(s) / Reference Number(s):**

**Test Name(s):**

#### ProGx Panels (by NGS Panel Genomic)

*Reflex available – Please contact us*

- 30x average read depth
- ~99% of targeted bases covered at >20x

### Whole Exome Sequencing (WES)

- □ Gold
  - 100x average read depth
  - 97-98% of targeted bases covered >10X
  - Turnaround time is 6-8 weeks
  - No prenatal testing available
- □ Platinum
  - 100x average read depth
  - 97-98% of targeted bases covered >10X
  - Turnaround time is 2-3 weeks
  - Prenatal testing is available

### Whole Genome Sequencing (WGS)

- □ WGS
  - 30x average read depth
  - ~99% of targeted bases covered at >10X
  - Turnaround time is 4-6 weeks
  - Prenatal testing is available
  - Del/Dup included

### Whole Exome Sequencing (WES)Additional analyses

- □ Solo
  - Solo implies analysis of index patient only; we recommend trio analysis for enhanced diagnostic accuracy.
- □ Trio
  - Trio implies analysis of index patient, along with the parents.
- □ Trio Plus
  - "Trio Plus" indicates "Trio" plus additional relatives. All trio samples have to be received simultaneously to start testing. If not, each sample from the same family will be charged as a solo.

### Additional analyses

- Del/Dup (+CNV and repeat expansion, if applicable)
- Sequencing (by NGS Panel)
- Deletion/Duplication Testing
- Repeat Expansion

### Reporting and data exchange

- □ .fastq
  - Raw data (.fastq and .bam files) are available only for a limited time and must be downloaded from the server within 1 month after the customer is informed of the completion of the analysis or after the final medical report has been issued.
- □ .bam
  - Data selected above with annotated and filtered variant report (Excel table)
- □ .vcf
  - Research Report (includes potential disease-causing variants in candidate genes for which there is not yet sufficient published evidence)

Eg: Specify genes of interest for Ex or ProGx panels
INFORMED CONSENT (ALL)

GENETIC TESTING CONSENT

A genetic test studies the inherited substance (DNA) using a molecular-genetic analysis of characteristics, which may be the cause of the disease that has occurred or is suspected in you or your family. The study material is usually a blood sample. Normally there are no health risks when taking a blood sample. Sometimes patients can experience bruising (hematoma) at the drawing site; very rarely, there could be nerve damage. Another risk that cannot be fully excluded exists in the extremely unlikely possibility of the samples being swapped. Every effort is made to avoid this and other mistakes.

DNA sequencing analyzes your DNA and compares it to the reference human genome. Variations (changes) are identified by comparing data with medical databases and looking for scientific links, all of which will be reported to your physician. While there are always certain variations, depending on the individual being tested and the available data, CENTOGENE and/or LifeLabs Genetics adhere to the guidelines set out by the American College of Medical Genetics (ACMG). A medical report may include information that is considered to be of direct and immediate relevance, either to your own health or to that of family members who share part of your genetic background.

Possible results of genetic testing include:
- Positive: Indicates a genetic variant was identified in a specific gene and that variant is pathogenic (likely highly likely to be causal of the disease-related condition).
- Negative: If no disease-causing variant is found, genetic changes responsible for the disease or a tendency to have a disease may still exist and cannot usually be fully excluded.
- Variant of Uncertain Significance: Sometimes, gene variants are proven but their significance is not clear. This is stated in the results and discussed with you by your physician. This category of variant is not reported for fetal samples or samples from deceased persons.

In addition, CENTOGENE and LifeLabs Genetics also make use of its own mutation database (CentoMD®), which encompasses over 12,000 mutations collected from a global population. Our medical colleagues may recognize other genes that might be of medical significance, and these can be reported as well. Should you not wish to receive this information in your report, it is possible to opt out of this service. A comprehensive explanation of all possible causes of diseases due to genetic reasons is not possible. It is also not possible to exclude every disease risk for you and your family members, especially your children, utilizing genetic analyses.

I consent to the carrying out of the genetic analysis indicated on these pages, on me or the person I am custodian for.

I understand that my specimen for DNA analysis will be sent to LifeLabs for genetic testing. I am aware that correct information about the relationships between my family members is important. I agree that my specimen and personal health information may be shared with LifeLabs. I understand that LifeLabs will contact me for a new specimen, if a test result cannot be provided from the original specimen. I agree to the carrying out of the genetic analysis indicated on these pages, on me or the person I am custodian for.

1. I understand that, once the requested test(s) has/have been completed, personal data and remaining sample will be stored at the testing laboratory for 20 years.
2. I agree that my de-identified sample may be used for product development or research purposes. I understand that I will not receive any royalties, resultant payments, benefits, or rights to products or discoveries.
3. I consent to the storage and use of my pseudonymized (encrypted) or de-identified test results in a statistical database for scientific purposes and to facilitate and improve the diagnosis of genetic changes and diseases in other patients.
4. I consent that my de-identified results stored in the database are being provided to physicians, scientists and researchers for the purposes of researching genetic diseases and improving their diagnostics and treatment.

I do not want my remaining sample or data from my results to be stored and/or used for product development or research purposes.

Please destroy any remaining sample once the final report has been issued. By ticking this box I disagree with points 1, 2, 3, & 4 listed above.

You can withdraw your consent to the analysis at any time in full or in part without stating reasons. You have the right not to be informed about test results (right not to know), to stop the testing processes that have been started at any time up to being given the results and to request the destruction of all test material and all results collected up to that time.

Patient/Substitute Decision Maker: _______________________; Date: _______________________

Printed name: _______________________; Relationship to person being tested: _______________________

OR: I certify that verbal consent was obtained from the patient/substitute decision maker for the requested genetic testing.

Signature of Physician: _______________________; Date: _______________________
Ex Panels and Whole Exome Sequencing

The exome is the collection of the DNA sequences of the genes that determine the production of proteins, which your body needs in order to function properly. So far, the exome is where the vast majority of causative mutations have been identified by scientific research. Whereas most genetic tests focus on a single gene or a set number of predetermined genes, WES examines thousands of genes simultaneously. Ex Panel focuses on approximately 6,700 genes for which scientific research has identified mutations that are directly related to the development of specific diseases or disorders. Whole Exome Sequencing studies the >20,000 genes that make up our genome, including those ~6,700 genes investigated by Expanded NGS Panels.

Whole Genome Sequencing

Our DNA is composed of exons (studied by sequencing the exome) and introns, among other regions. Introns were previously not considered to contain important genetic information, as these regions do not directly determine the function of proteins. Recent research has provided evidence that some of these regions may be involved in the development of certain rare diseases and disorders. Whole Genome Sequencing analyzes all parts of the >20,000 genes that make up our genome.

Incidental or Secondary Findings

CENTOGENE and LifeLabs Genetics adhere to the guidelines set out by the American College of Medical Genetics (ACMG), which allow for reporting specific types of medically actionable or incidental findings (PMIDs: 23788249 and 25356965). Medically actionable findings may be associated to a predisposition to increased cancer risk, a carrier status of recessive diseases, or a predisposition to late-onset diseases, among others. This list of genes from the ACMG is constantly being updated to include all such instances of mutations that are seen as being relevant to patients. Patients are required to select whether or not they would like to receive information on the 59 genes or classes of genes outlined in these recommendations, which are known to be medically actionable. Incidental findings are not reported for fetal samples or samples from deceased persons.

Confirmation of Findings

CENTOGENE and/or LifeLabs Genetics use Sanger sequencing to confirm all pathogenic variants that do not pass the quality control parameters of next-generation sequencing. Structural variants are confirmed by orthogonal methods, such as MLPA or qPCR.

Use of Parental Samples for Large Scale Testing

Biological parental samples are used to improve the interpretation of the final results in exome and genome testing. Trio analysis, testing and bioinformatic analyses on parental samples are done in parallel to the analysis of the index patient. We check the parents’ materials only with regard to the patient’s condition and issue parental reports accordingly. If additional analyses on the parental samples are required, such as complete exome analyses or analyses of the 59 genes or classes of genes outlined in the ACMG guidelines, please contact us as additional charges may apply. If several family members are tested, accurate interpretation of the results depends on the assumed relationships being correct. If doubt is created by the genetic analysis about the apparent relationships, we will not inform you. An exception will be made if it is absolutely necessary for the completion of the requested test.

Technical Limitations

1. Exome testing does not analyze all genes in the human genome. Some genes cannot be examined because of various technical reasons. For the targeted exome and the whole exome, respectively, approximately 5% and 3% of the targeted exons may not be well covered due to various technical reasons.
2. You may have a mutation in one of the genes included in the test, but it is not always possible to detect all mutations with these methods. This means that a patient can be affected with a certain condition, but that this testing does not identify or reveal it.
3. Exome and Genome testing encompasses many different genes and looks for a variety of conditions and diseases. These tests may reveal genetic information about you or a family member that is new and is not necessarily related to your reasons for ordering such a test. Such information could reveal details about diseases that will only develop in the future or for which there is no known treatment or cure.

Consent to Exome or Genome Testing

(Ex and ProGx Panels and Whole Exome and Whole Genome) (In addition to the general informed consent on page 3)

The initials of the physician confirm the patient has been informed of all the information on this page.

HCP

The initials of the patient confirm he/she has chosen to receive information on the 59 genes or classes of genes outlined in the ACMG recommendations described above.

Patient

For private pay testing, the initials of the patient confirm, if an exome or genome test is cancelled prior to test set-up, he/she will be charged a processing fee and will receive a cancellation report. Once testing is initiated, the full price of the analysis will be charged.

Patient