

ORDERING HEALTHCARE PROVIDER	
Billing #	_____
Name	_____
Address	_____
	No Street
	_____
	City Province Postal Code
Telephone	_____ Fax _____
Physician Signature	_____

COPY TO HEALTHCARE PROVIDER	
Billing #	_____
Name	_____
Address	_____
	No Street
	_____
	City Province Postal Code
Telephone	_____ Fax _____

REQUIRED PATIENT INFORMATION	
Has carrier screening already been ordered for reproductive partner? * <input type="checkbox"/> YES <input type="checkbox"/> NO	
<b>Reason for testing</b> (select all that apply): <input type="checkbox"/> Screening for genetic carrier status <input type="checkbox"/> Family history <input type="checkbox"/> Partner is a known carrier <input type="checkbox"/> Patient / Partner is pregnant <input type="checkbox"/> Egg/sperm donor <input type="checkbox"/> High risk ethnicity <input type="checkbox"/> Other: _____	<b>Ethnicity</b> (select all that apply): <input type="checkbox"/> French Canadian or Acadian <input type="checkbox"/> Ashkenazi Jewish <input type="checkbox"/> Sephardic Jewish <input type="checkbox"/> Caucasian/White <input type="checkbox"/> Asian <input type="checkbox"/> African or African American <input type="checkbox"/> Hispanic <input type="checkbox"/> Mediterranean <input type="checkbox"/> Indigenous <input type="checkbox"/> Pacific Islander <input type="checkbox"/> Other: _____

PARTNER INFORMATION	
REQUIRED if your partner has already performed the Invitae Carrier Screen	
Partner DOB	_____
	MM / DD / YYYY
Date Partner Tested	_____
	MM / DD / YYYY
Partner Name	_____
Partner RQ #	_____
	From original report

COLLECTION INFORMATION	
Sample type	<input type="checkbox"/> Blood (EDTA: 4mL) <input type="checkbox"/> Other _____
Date & Time	_____
Blood Collected	_____
	MM / DD / YYYY HH / MM
Collector Name	_____

LIFELABS LABELS	

PATIENT INFORMATION	
Last Name	_____
First Name	_____
Date of Birth	_____ Sex <input type="checkbox"/> M <input type="checkbox"/> F
	MM / DD / YYYY
Address	_____
	No Street
	_____
	City Province Postal Code
Telephone	_____

TEST REQUESTED	COST	LL TC	Mnemonic
<input type="checkbox"/> <b>Expanded Carrier Screen, Single</b> Carrier screening panel that performs sequencing of over 289 clinically significant conditions (Includes all genes in the French Canadian Carrier Screen)	\$625 CAD	4101	FP2
<input type="checkbox"/> <b>French Canadian Carrier Screen only</b> Carrier screening panel, including 8 conditions, specific to those with French Canadian Ancestry (Includes CFTR, FAH, GNPTAB, HEXA, LRPPRC, MPI, SACS, SLC12A6)	\$625 CAD	4101	FP2

PATIENT CONSENT - MANDATORY	
<p>I have read the Patient Information Form (on reverse). I understand that 1 blood sample (or saliva in some cases) will be taken by LifeLabs staff. I acknowledge that my sample and personal health information will be sent to Invitae for the purpose of carrier screening at their lab in the United States. I also understand that LifeLabs will contact me for a new blood sample if a test result cannot be provided from the original blood sample. I acknowledge that LifeLabs will receive the results from Invitae and it will disclose the results to the ordering physician. I also understand that I will be contacted by LifeLabs to obtain consent should LifeLabs be asked to disclose my information for another reason, other than as required or permitted by law. I acknowledge that I am responsible for the full cost of testing.</p>	
Patient Name	_____
Signature	_____
	MM / DD / YYYY

The minimum amount of patient information is collected for provision of the service requested. This information is considered confidential. Unauthorized use and disclosure are prohibited.

This reviews the benefits, risks, and limitations of DNA testing for the genetic disorders available through the Expanded Carrier Screen.

**PURPOSE OF THE TEST(S)**

The Expanded Carrier Screen (ECS), as performed by Invitae in California, analyzes specific changes in your DNA called mutations. Certain mutations can make it more likely that you could pass on a hereditary condition through a pregnancy. You may use this information to inform your decisions in preparing for a family. Information about diseases, such as description, course, and possible treatments, may be found in the Carrier Screen section of the LifeLabs Genetics website. For most of the conditions on the panel, both parents must carry a mutation in the same disease gene for their child to be at risk to be affected. This is called autosomal recessive inheritance. There are, however, a few diseases on the panel that can be transmitted when only one parent is a carrier. Fragile X is an example of a disease that requires only one parent to carry the mutation in order to be passed on. For some conditions on the panel, such as Gaucher's disease, it is possible to be diagnosed with a form of the condition that does not appear until adulthood (i.e., adult-onset). If you have a family history of one of the conditions on this panel, you should inform the LifeLabs Genetics team of the specific gene mutation(s) present in your family. Screening for the diseases on this panel may significantly reduce the likelihood that you are a carrier but does not guarantee that you are not a carrier.

**TEST RESULTS AND INTERPRETATION**

One tube of blood or saliva is required from each person consenting to testing. Your test results will be sent to the healthcare provider who ordered the test. The following describes the possible results outcomes:

- **Carrier (Positive):** A positive result indicates that a gene mutation has been identified and that you are a carrier of this disorder. You may be identified as a carrier for more than one disorder. Carriers usually do not experience symptoms of the disease.
- **No mutations detected (Negative):** A negative result indicates that no gene mutation was identified. This reduces but does not eliminate the possibility of being a carrier.
- **Indeterminate:** An "indeterminate" result indicates that we cannot confidently report a positive or negative result using stringent quality-control guidelines.
- **Homozygote or compound heterozygote:** This result indicates the presence of two disease-causing mutations in the same gene, which would typically indicate that you are affected now or may be affected in the future. However, some of the disorders in this panel may be mild or may vary in severity, so you may not experience clinically significant symptoms. In rare cases, a person may have two disease-causing mutations on the same chromosome, which may be revealed by further testing of either that person or their family.

There is a chance that the sample(s) submitted will not return results. In this case, your healthcare provider will be informed by LifeLabs Genetics and you may be asked to provide a second sample to repeat the test. There is no charge for a repeat. ECS testing is highly reliable with >99% accuracy for targeted mutations and regions. As with all medical screening tests, there is a chance of a false positive or false negative result. A "false positive" refers to identifying a gene mutation that is not present. A "false negative" is the failure to find a mutation that is present in the sample. Result interpretation is based on currently available information in the medical literature and scientific databases. Because literature and scientific knowledge are constantly being updated, new information may replace or add to the information that Invitae used to interpret your results. Invitae does not routinely re-analyze test results or issue new test reports, and has no obligation to do so.

**BENEFITS**

Your carrier screening results may help you and your partner make more informed decisions regarding your family, particularly if screening is performed prior to conceiving a pregnancy. Your results may also benefit family members. If you test positive, your biological relatives are more likely to test positive for the same mutation(s), thereby discovering previously unknown risks.

**RISKS**

Genetic testing may reveal sensitive information about your health or that of your relatives. If you and your partner are receiving simultaneous testing, each of your test results may be revealed to one another. Test results may reveal incidental, unsought information, such as discovering that a man is not the father of a child (non-paternity).

**TEST LIMITATIONS**

This test is designed to detect known DNA mutations associated with genetic disease. It cannot detect every mutation associated with each disease, nor does it look for all known genetic diseases. Because of this, the ECS test is risk-reducing, not risk-eliminating. Negative results do not guarantee that you or your offspring will be healthy. If you wish to further reduce your reproductive risks, your partner's carrier risk or the risk to potential pregnancies, additional testing may be available. Mutation scanning or sequence analysis for some disorders may not be available. Some biological factors, such as a history of bone marrow transplantation or recent blood transfusions, may limit the accuracy of results. Diagnostic errors may occur due to sample mix-up or contamination.

**CONFIDENTIAL REPORTING PRACTICES**

LifeLabs and Invitae have entered into a mutually binding distribution agreement whereby both organizations will comply with all applicable legislation. Invitae complies with HIPAA confidentiality laws; LifeLabs Genetics complies with Canadian privacy rules. LifeLabs will only report test results to the ordering healthcare provider(s) or genetic counsellor involved. You must contact your provider to obtain the results of the test. Additionally, the test results could be released to those who, by law, may have access to such data.

**FINANCIAL RESPONSIBILITY**

Some provincial and/or personal medical insurance plans may cover the cost of the test. Check with your insurance provider. Otherwise, you are responsible for the cost of the test and will provide payment to LifeLabs Genetics, who in turn will provide payment to Invitae. Payment can be made by credit card or debit.

**GENETIC COUNSELLING**

If you have remaining questions about carrier screening after talking with your healthcare provider, we recommend that you speak with a genetic counsellor who can give you more information about your testing options. You can find a genetic counsellor in your area by going to the Canadian Association of Genetic Counsellors website at <https://cagc-accg.ca/>. The cost of the test includes genetic counselling services provided by Invitae to discuss your Carrier Screen result. They can be reached at 1-800-436-3037 or by email at [gcservices@invitae.com](mailto:gcservices@invitae.com).

**DISPOSITION OR RETENTION OF SAMPLES**

Invitae may also keep your leftover de-identified samples for ongoing research and development. You and your heirs will not receive any payments, benefits, or rights to any resulting products or discoveries. Invitae may also contact you in the future for research opportunities.

Please contact Invitae at [clientservices@invitae.com](mailto:clientservices@invitae.com) or 1-800-436-3037 if you wish to opt out of such research or future contact.