

Expanded Carrier Screening



Knowing whether you are a carrier of genetic conditions provides valuable health information when planning a family.



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Expanded Carrier Screening

Next-generation sequencing of the entire gene of interest provides the most comprehensive analysis for detection of genetic mutations.

289
Genes tested

FULL
Gene sequencing

~3
Week turnaround

The LifeLabs Expanded Carrier Screen covers many types of diseases:

Disease examples that

impact life expectancy and quality of life

▶ **Cystic fibrosis**
40 years

▶ **Tay-Sachs disease**
3-5 years

▶ **Spinal muscular atrophy**
less than 2 years

Disease examples that

have treatment options available

▶ **MCADD**
normal life quality expected with treatment

▶ **Wilson Disease**
most symptoms prevented with early treatment

▶ **Beta thalassemia**
can extend beyond 40 years with treatment

Disease examples that

carry a significant risk for intellectual disability

▶ **Fragile X syndrome**

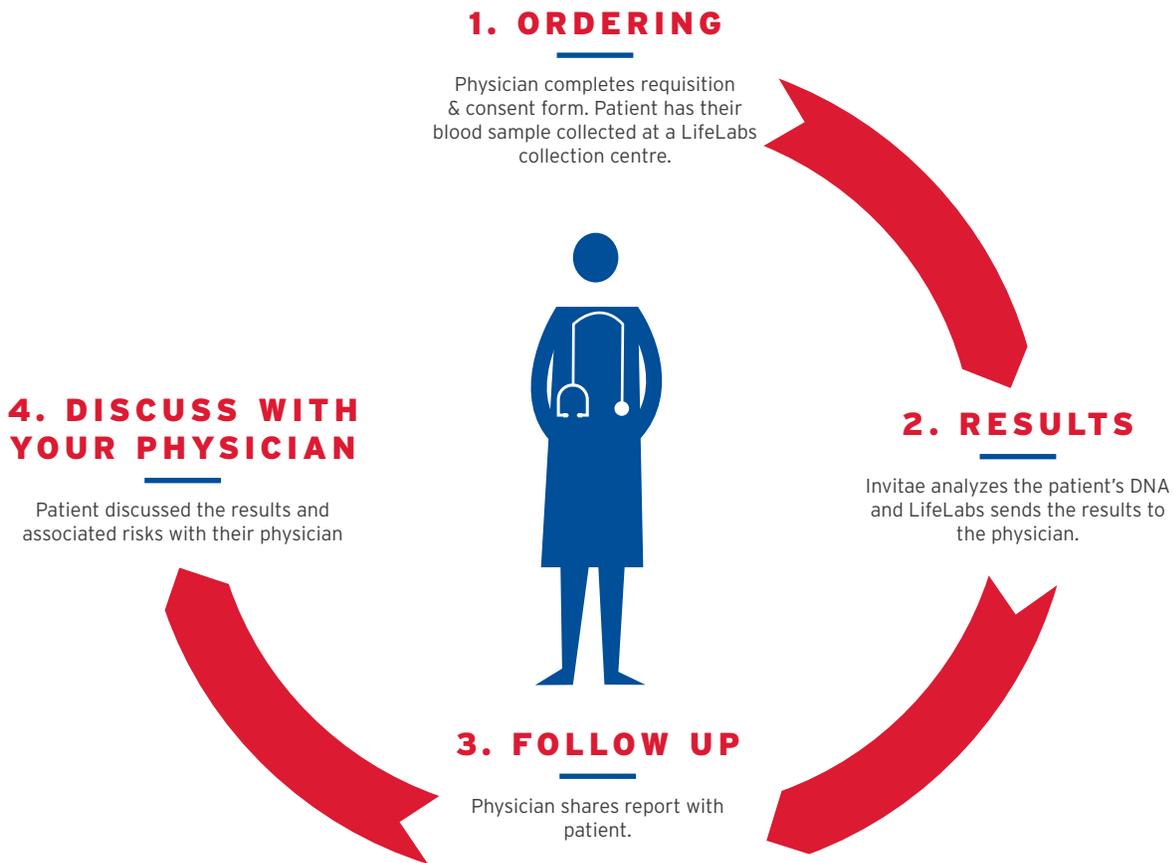
▶ **Smith-Lemli-Opitz syndrome**

▶ **Costeff optic atrophy syndrome**

1/550

pregnancies are affected by a condition on this Expanded Carrier Screen.

Patient Flow



Patients to Consider

"All individuals, regardless of race or ethnicity, are offered screening for the same set of conditions." - ACOG, ACMG, NSGC, PQF, SMFM Joint Statement ¹

The percentage of affected pregnancies missed by the current, ethnicity-based guidelines include:²

94% EAST ASIAN

79% HISPANIC

65% NORTHERN EUROPEAN

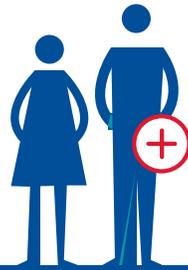
55% ASHKENAZI JEWISH

"The option of (Expanded Carrier Screening) should be part of the informed consent process." - CCMG and SOGC Joint Opinion ³

1. Edwards JG et al. (2015) Expanded carrier screening in reproductive medicine-points to consider: a joint statement of the American College of Medical Genetics and Genomics, American College of Obstetricians and Gynecologists, National Society of Genetic Counselors, Perinatal Quality Foundation, and Society for Maternal-Fetal Medicine. *Obstet Gynecol.* 125(3):653-662
2. Haque IS et al. (2016) Modeled fetal risk of genetic diseases identified by expanded carrier screening. *JAMA.* 316(7): 734-742.
3. Wilson RD et al. (2016) Joint SOGC-CCMG opinion for reproductive genetic carrier screening: An update for all Canadian providers of maternity and reproductive healthcare in the era of direct-to-consumer testing. *J Obstet Gynaecol Can.* 38(8): 742-762.

The MOH evaluates each case on an individual basis. MOH funding has been observed for, but not limited to, individuals whose partner is a carrier of a condition on the Invitae panel, for individuals with limited family history (adopted), and for Ashkenazi Jewish, French Canadian, and consanguineous couples.

Patients to Consider



Patients are encouraged to share their results with close relatives, especially those who are planning on having children in the near future

“I am the type of person that likes to have as much information as possible to prepare.”

Heather - *negative for all diseases screened*

WHAT IS CARRIER SCREENING?

A carrier screen analyzes a person's genes to determine if they carry a recessive genetic condition. A screen is able to detect if you are a carrier of many, but not all, conditions.

WHAT IS A RECESSIVE DISEASE AND WHAT IS A CARRIER?

Recessive conditions are caused by changes, called mutations, in a person's genes. Each person has 2 copies of any given gene, one copy inherited from each parent. A recessive condition occurs when both copies of the same gene have a mutation. A carrier is someone who has only one gene with a mutation and one gene that is unaffected. Carriers are typically symptom-free and do not know they carry a mutation.

When two parents are carriers of the same genetic mutation, each child has a 1 in 4 (or 25%) chance of being affected by the associated condition. For certain conditions, such as Fragile X syndrome, only the mother needs to be carrier for the child to be at an increased risk of being affected by the condition.

WHAT IF AN INDIVIDUAL IS NOT A CARRIER?

Generally, no follow-up testing is suggested for the conditions screened. It is important to understand that no screen is able to identify every carrier of every condition. You should also know that while the Expanded Carrier Screen covers a lot of information, we cannot screen for all possible birth defects and genetic condition. Family history or other factors should also be considered.

“Having all that knowledge will lead to better decisions for myself, for my wife, for my future family.”

Rajeev - *partner tested positive*

SOGC-Recommended Conditions

Alpha Thalassemia (HBA1/HBA2) ACOG ACMG	Glycogen Storage Disease, Type Ia (G6PC)
Andermann Syndrome (SLC12A6)	GNPTAB-Related Disorders (GNPTAB)
ARSACS (SACS)	Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (HBB) ACOG
Bardet-Biedl Syndrome, BBS1-Related (BBS1)	Hexosaminidase A Deficiency (including Tay-Sachs Disease) (HEXA) ACOG ACMG
Bardet-Biedl Syndrome, BBS10-Related (BBS10)	Joubert Syndrome 2 (TMEM216)
Bardet-Biedl Syndrome, BBS12-Related (BBS12)	KCNJ11-Related Familial Hyperinsulinism (KCNJ11)
Bardet-Biedl Syndrome, BBS2-Related (BBS2)	Leigh Syndrome, French-Canadian Type (LRPPRC)
Bloom Syndrome (BLM) ACMG	Lipoamide Dehydrogenase Deficiency (DLD)
Canavan Disease (ASPA) ACOG ACMG	Maple Syrup Urine Disease, Type IB (BCKDHB)
CLN3-Related Neuronal Ceroid Lipofuscinosis (CLN3)	Mucopolidosis IV (MCOLN1) ACMG
CLN5-Related Neuronal Ceroid Lipofuscinosis (CLN5)	NEB-Related Nemaline Myopathy (NEB)
CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 (CLN6)	Niemann-Pick Disease, Type C2 (NPC2)
Congenital Disorder of Glycosylation, Type Ib (MPI)	Spinal Muscular Atrophy (SMN1) ACOG ACMG
Cystic Fibrosis (CFTR) ACOG ACMG	Tyrosinemia, Type I (FAH)
ERCC6-Related Disorders (ERCC6)	Usher Syndrome, Type 3 (CLRN1)
Familial Dysautonomia (IKBKAP) ACOG ACMG	
Fanconi Anemia, Type C (FANCC) ACOG	
FKTN-Related Disorders (including Walker-Warburg Syndrome) (FKTN)	
Fragile X Syndrome (FMR1) X-linked	
Gaucher Disease (GBA) ACMG	

The SOGC-recommended conditions listed above are included in the 288 that are tested on the Expanded Carrier Screen.

ACOG Indicates testing also recommended by ACOG
ACMG Indicates testing also recommended by ACMG
X-linked Indicates X-linked disorders

Disease Categories

Many of the diseases included in our screen are vital to know about. The categories below provide an overview of the types of conditions included in our Expanded Carrier Screen.



EARLY INTERVENTION: Some of the conditions on the Expanded Carrier Screen can be treated early in life, like Wilson disease and PKU.



INTELLECTUAL DISABILITY: Some result in intellectual disabilities, as with fragile X syndrome and Niemann-Pick disease.



SHORTENED LIFE EXPECTANCY: Others are chronic and require lifelong management, like cystic fibrosis and Bloom syndrome.



LIMITED OR NO TREATMENT: Finally, some of the conditions have no treatments available, like spinal muscular atrophy and Canavan disease.

- You can take the Expanded Carrier Screen before or during pregnancy
- It's normal to be a carrier - what you really want to know is if both partners are carriers of the same disease
- Most carriers have no history of the condition within their family



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LifeLabs Genetics is at the forefront of clinical genetic testing and personalized medicine in Canada, working with the world's leading laboratories, healthcare providers, and government partners to find the most cost-effective and meaningful, clinically-relevant way to bring genetic advancements to Canadians.

Our partners include:

