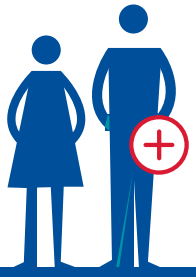
Expanded Carrier Screening

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

LifeLabs GENETICS®

Ask.Genetics@LifeLabs.com
www.lifelabsgenetics.com
1-844-363-4357

Patients to Consider



IT'S GOOD TO KNOW

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 copy of a gene with a mutation and one copy of a gene that is unaffected. Carriers are typically symptom-free and do not know they carry a mutation.

When two parents are carriers of a mutation in the same gene, 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 conditions. 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*

Expanded Carrier Screening

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

569	FULL	~3
Genes tested	**Gene sequencing**	**Week turnaround**

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.

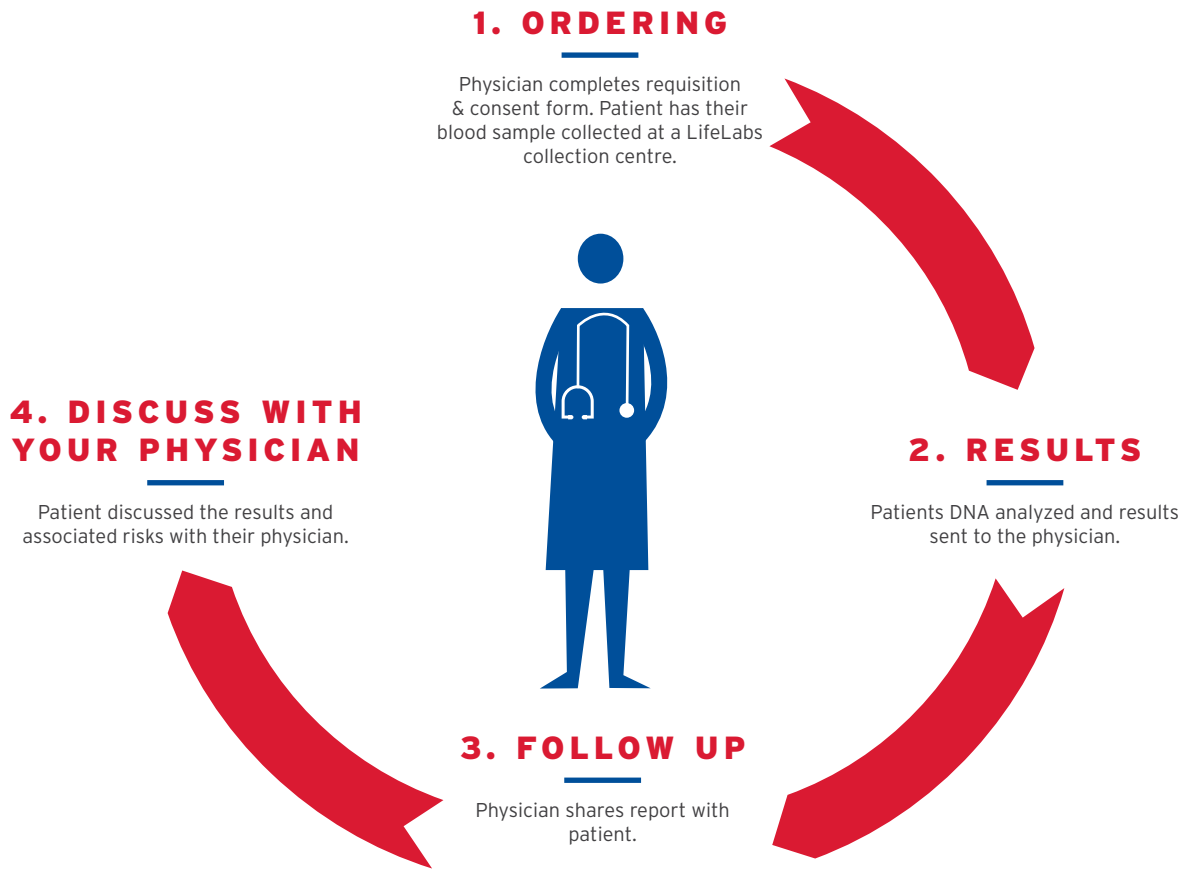
SHORTENED LIFE EXPECTANCY: Others are chronic and require lifelong management, like cystic fibrosis and Bloom syndrome.

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

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 screening 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 fo 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 panel, for individuals with limited family history (adopted), and for Ashkenazi Jewish, French Canadian, and consanguineous couples.

SOGC-Recommended Conditions

Alpha Thalassemia (HBA1/HBA2) ACOG ACMG

Andermann Syndrome (SLC12A6)

ARSACS (SACS)

Bardet-Biedl Syndrome, BBS1-Related (BBS1)

Bardet-Biedl Syndrome, BBS10-Related (BBS10)

Bardet-Biedl Syndrome, BBS12-Related (BBS12)

Bardet-Biedl Syndrome, BBS2-Related (BBS2)

Bloom Syndrome (BLM) ACMG

Canavan Disease (ASPA) ACOG ACMG

CLN3-Related Neuronal Ceroid Lipofuscinosis (CLN3)

CLN5-Related Neuronal Ceroid Lipofuscinosis (CLN5)

CLN6-Neuronal Ceroid Lipofuscinosis, Type 6 (CLN6)

Congenital Disorder of Glycosylation, Type Ib (MPI)

Cystic Fibrosis (CFTR) ACOG ACMG

ERCC6-Related Disorders (ERCC6)

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

Glycogen Storage Disease, Type Ia (G6PC)

GNPTAB-Related Disorders (GNPTAB)

Hemoglobinopathy (including Beta Thalassemia and Sickle Cell Disease) (HBB) ACOG

Hexosaminidase A Deficiency (including Tay-Sachs Disease) (HEXA) ACOG ACMG

Joubert Syndrome 2 (TMEM216)

KCNJ11-Related Familial Hyperinsulinism (KCNJ11)

Leigh Syndrome, French-Canadian Type (LRPPRC)

Lipoamide Dehydrogenase Deficiency (DLD)

Maple Syrup Urine Disease, Type IB (BCKDHB)

Mucolipidosis IV (MCOLN1) ACMG

NEB-Related Nemaline Myopathy (NEB)

Niemann-Pick Disease, Type C2 (NPC2)

Spinal Muscular Atrophy (SMN1) ACOG ACMG

Tyrosinemia, Type I (FAH)

Usher Syndrome, Type 3 (CLRN1)

The SOGC-recommended conditions listed above are included in the 569 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

Please visit LifeLabsGenetics.com for the full list of conditions covered on our Expanded Carrier Screen.

Genetic Counselling Support

Post-test genetic counselling with a summary letter is included with the price of the test and can be accessed by calling **1-800-436-3037.**

Also, certified Canadian genetic counsellors at LifeLabs are available to healthcare providers and patients to answer any questions about the test or results at:

Ask.Genetics@LifeLabs.com **1-844-363-4357.**

Services are available from 8am - 7pm EST in both English and French.

LifeLabs GENETICS®

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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.

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