Quick Facts

- Results are available in 2-3 weeks
- Genetic counselling services are strongly recommended for screens that come back with a positive result
- Genetic counselling services are offered to anyone with questions or concerns
- If you are found to be a carrier of one of the screened conditions, your relatives may also be carriers of the same condition
- Carrier screening is an optional tool to assist in reproductive planning
- Carrier screening is not covered by OHIP but may be covered by your private insurance.
- Please refer to www.mssscreening.com for current pricing.

Need additional help?

Receiving your result can be confusing especially if there are decisions that need to be made. Genetic counsellors are experienced in providing support and information that will help you and your partner make an informed decision. The only right decision is the one that is best for you and your family. Genetic counselling services are offered by calling us at **1 866 330-8797** or e-mailing **info@mountsinaiservices.com**

GETTING STARTED IS EASY

1. Pick up your kit

Pick up your carrier screening sample collection kit from your healthcare provider.

2. Informed Consent

By providing consent you understand the nature and purpose of the screen and any questions or concerns have been answered to your satisfaction.

3. Provide a sample

Collect a cheek (buccal) swab and ship with our prepaid pre-addressed FedEx package.

4. Receive your results

Your results will be sent to your doctor. Your doctor will contact you to discuss your options.

5. Get additional support

Speak to an experienced genetic counsellor that will go over your results, discuss your options and help you make informed decisions.

Learn more

Talk to Mount Sinai Services staff about carrier screening and learn more about your options. For more information contact us by:

1 866 330-8797

info@mountsinaiservices.com

www.mssscreening.com

MSS is associated with Sinai Health System. Net proceeds from the program will be reinvested to support patient care and research at Sinai Health. Sinai Health has been involved in the validation of the medical content of the document and other program support. MSS takes responsibility for its overall content.

Compensation may be provided to physicians or associated parties for test kits ordered as a result of their consultation, subject to applicable law and standards of practice.

Mount Sinai SERVICES

Powered by fulgent

Mount Sinai SERVICES

CARRIER SCREENING

INFORMATION FOR PATIENTS

Powered by fulgent

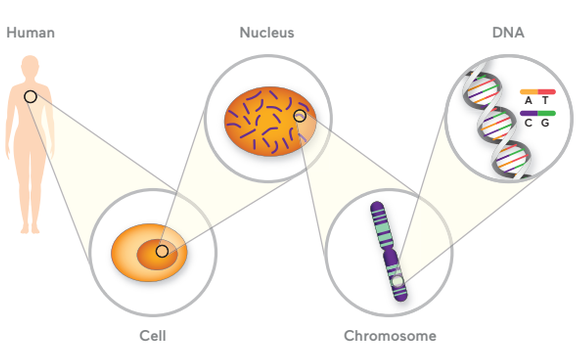
Most people do not know their genetic carrier status because they do not have symptoms or a family history of disease.

If you are pregnant or planning on becoming pregnant, carrier screening can provide information about your risk of having a child with a genetic condition. Carrier screening is being used as part of pregnancy planning, and may allow you to make a more informed decision about your reproductive options (eg IVF) and prenatal care.

What is a carrier?

A carrier is a person who has a genetic change, also known as a variant, in their DNA. Most of us are carriers for many genetic variants, but we do not know the gene variants we carry. This is because carriers are typically healthy and do not show signs or symptoms of the condition.

However, if both parents are carriers for the same condition, the couple can have an increased risk of having a child that is affected.



What is carrier screening?

Carrier screening is a genetic screen that looks at the variants in a person's DNA to determine the risk of having a child that may develop a genetic condition.

What is the expanded carrier screen?

The expanded carrier screen analyzes more than 400 genes (out of 25,000) to identify if you are a carrier of a genetic condition that could, otherwise unknowingly, be passed to your child. Some of the screened conditions are:

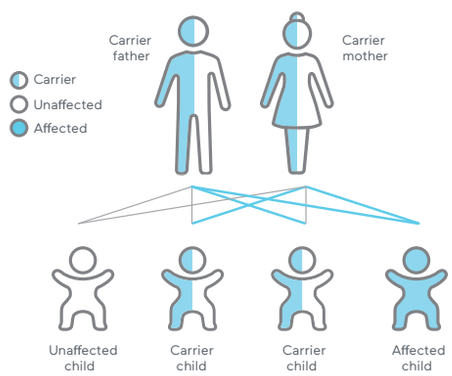
- Cystic Fibrosis
- Fragile X
- Sickle Cell Anemia
- Spinal Muscular Atrophy
- Tay-Sachs
- Thalassemia
- Canavan Disease
- Bloom Syndrome
- Wilson Disease

These conditions can be passed on in a family in two different ways: by autosomal recessive and X-linked recessive inheritance.

Autosomal recessive conditions

For all genes we have two copies — ones we inherit from our fathers and ones we inherit from our mothers. Autosomal recessive conditions will only develop if both copies of the gene change.

Autosomal recessive inheritance

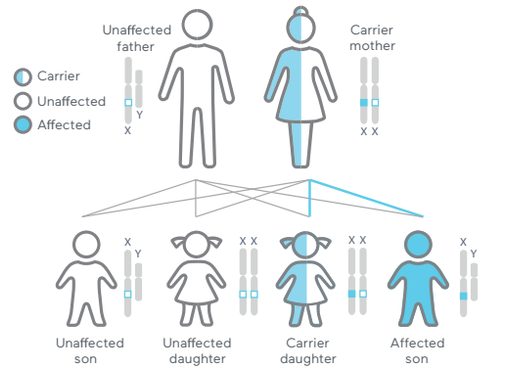


If both parents carry a disease-causing variant in the same gene, there is a 1 in 4 (25%) chance of having an affected child or a 3 in 4 (75%) chance of having an unaffected child each time they are pregnant.

X-linked conditions

Some disease-causing variants are located on the X chromosome. Females have two X chromosomes while males have one X chromosome and one Y chromosome. Males that inherit disease-causing variants on the X chromosome are always affected, while females are often unaffected carriers.

X-Linked recessive inheritance



In every pregnancy, there is a 1 in 2 (50%) chance that each child will inherit the variant from the mother. Sons who inherit the disease-causing variant will be affected and daughters will be carriers.

Why screen for these conditions?

In the past, the only clue that a healthy person was a carrier was the diagnosis of a genetic condition in their child or family member. That has now changed. We can test genes of an individual or a couple to see if they are carriers and at increased risk of having a child with a disorder before becoming pregnant (preconception) or in early pregnancy.

Benefits of carrier screening

- Reproductive planning
- Prenatal testing options
- Preparation for the birth of a child with a genetic disorder
- Healthcare options that are more appropriate for you and your family