

How does Sema4 Expanded Carrier Screen work?



To get started, talk to your doctor about requesting a carrier screen.



Your doctor will take a blood draw or saliva sample and send it to our lab.



We analyze your DNA sample using our state-of-the-art sequencing technologies. This usually takes about 2 weeks.



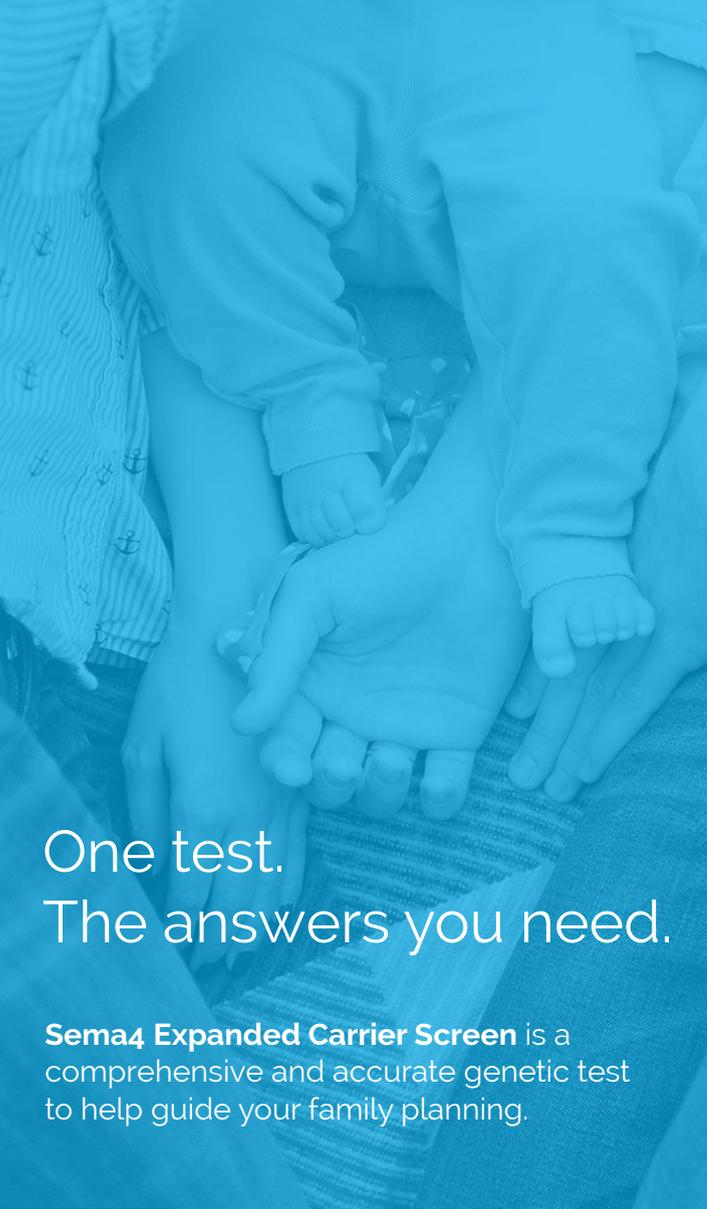
Once your carrier screen is complete, your doctor or a genetic counselor from Sema4 may contact you to explain your results and answer any questions you have.

Billing and insurance

Carrier screening is covered by most insurance plans, however, copays, co-insurance, and/or deductibles may vary by health plan. To determine what your out-of-pocket expenses may be, please contact our billing specialists at **1-800-298-6470** or your insurance provider by calling the number on the back of your insurance card.

If you need help covering the cost of carrier screening, please contact our billing specialists to learn more about our financial assistance options and payment plans.

To learn more about Sema4 Expanded Carrier Screen, please visit sema4genomics.com.



One test.
The answers you need.

Sema4 Expanded Carrier Screen is a comprehensive and accurate genetic test to help guide your family planning.



What is carrier screening?

If you are pregnant or planning for pregnancy, carrier screening can help you understand your risk of passing on a genetic condition to your child.

Carrier screening is a test that analyzes your DNA to see if you are a carrier of a genetic variant (or change) that could cause an inherited disease in your children. Medical experts, such as the American College of Obstetricians and Gynecologists, recommend that carrier screening be offered to all women who are pregnant or planning for pregnancy.

Carrier screening plays an important role in family planning, and is ideally performed before you get pregnant. This gives you the most reproductive options if you test positive as a carrier for a genetic condition. If you or your partner are already pregnant, carrier screening is still important to help you better understand your reproductive risk and prepare for a healthier future for your family.

What does it mean to be a carrier of a genetic condition?

Every person has two copies of each gene. Carriers have one normal copy of a gene and one copy with a disease-causing variant (or change). Most carriers are healthy and do not have a family history of genetic conditions.

There are two ways that a carrier may pass on a genetic condition to their child:

- **Autosomal recessive inheritance**, when a child inherits a gene with a disease-causing variant (or change) from both the mother and the father
- **X-linked inheritance**, which typically occurs when a male inherits an X-chromosome with a disease-causing variant from the mother

Sema4 Expanded Carrier Screen tests for both autosomal recessive and X-linked diseases.

Why should I choose Sema4 Expanded Carrier Screen?

Sema4 Expanded Carrier Screen is designed to give you a comprehensive and accurate understanding of your risk of passing on a genetic condition to your child.



Simple

With a simple blood or saliva test, find out if you and your partner are carriers of genetic variants (or changes) that could affect your children.



Complete

Sema4 Expanded Carrier Screen is a comprehensive carrier screen for **more than 280 inherited diseases**, including cystic fibrosis, spinal muscular atrophy, and Fragile X syndrome. For the full list of conditions that this test screens for, please visit sema4genomics.com.



Accurate

When assessing your risk, precision matters. Our Expanded Carrier Screen uses advanced sequencing technologies for results that are >95% accurate.

What options are available for carrier screening?

While a comprehensive carrier screen is recommended if you are pregnant or planning for pregnancy, carrier screening can also be customized for your unique family planning needs. We offer several carrier screening options, including:

Standard Carrier Screen

This panel screens for four genetic conditions that are more common — spinal muscular atrophy, cystic fibrosis, Smith-Lemli-Opitz syndrome, and Fragile X syndrome. The American College of Obstetricians and Gynecologists recommends that all women should be offered carrier screening for spinal muscular atrophy and cystic fibrosis, as well as screening for Fragile X syndrome in certain cases of personal or family health history.

High-Frequency Carrier Screen

This panel include all of the conditions in our standard carrier screen panel, plus six other diseases that are known to occur more frequently in all ethnic groups.

Comprehensive Jewish Carrier Screen

This panel of 96 diseases includes 48 diseases relevant to the Ashkenazim, 38 diseases relevant to the Sephardi and Mizrahi communities, and 10 diseases common to all people of Jewish descent.

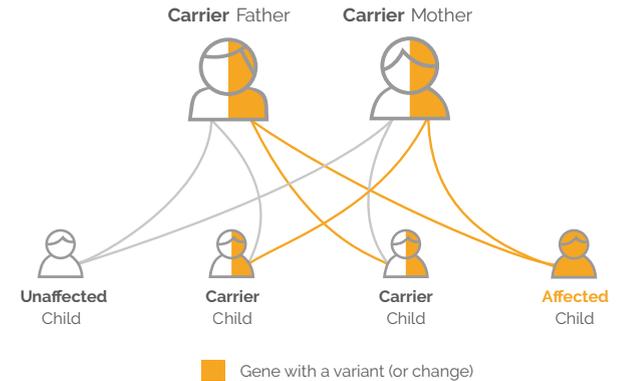
Expanded Carrier Screen

A comprehensive carrier screen that tests for more than 280 genetic diseases.

If I am a carrier, what is the risk that my children will inherit the condition?

If both you and your reproductive partner are carriers of the same autosomal recessive condition, with each pregnancy you have a

- **1 in 4 (25%)** chance of having a child affected with that disease.
- **1 in 4 (25%)** chance of having a child who is not affected by the disease and not a carrier
- **2 in 4 (50%)** chance of having a child who is a carrier of the disease



If a woman is identified as a carrier of an X-linked condition, she has a **1 in 2 (50%)** chance with each pregnancy to pass on the variant to her child. If a male child inherits the disease-causing variant, he will be affected with the X-linked condition.

What are my options if I am a carrier?

If you are a carrier of a genetic condition, carrier screening may also be recommended for your reproductive partner. Our genetic counselors can help coordinate partner testing. If you and your partner are identified as a high-risk couple, your doctor or genetic counselor can provide information and support to help you understand your reproductive options and make important family planning decisions.