How does Sema4 Elements™ ECS work?

1. To get started, talk to your healthcare provider about requesting a carrier screen.
2. Your healthcare provider will take a blood draw or saliva sample and send it to our lab.
3. We analyze your DNA sample using our state-of-the-art sequencing technologies. This usually takes about 2 weeks.
4. Once your carrier screen is complete, your healthcare provider 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 **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 Elements™ ECS, please visit **sema4.com/carrierscreen.** You can also watch our Pre-Test Video on this webpage, or by **texting ECS to 844-988-0992** or **scanning the QR code** with your smart phone camera.

Sema4 Elements™ offers a portfolio of information-driven genomic solutions, digital tools for patients and providers, and services that enable providers to treat patients holistically during their reproductive and generational health journey.

Learn more at **sema4.com/Elements**

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Sema4 Elements™

Expanded Carrier Screen

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 Elements™ Expanded Carrier Screen (ECS) tests for both autosomal recessive and X-linked diseases.

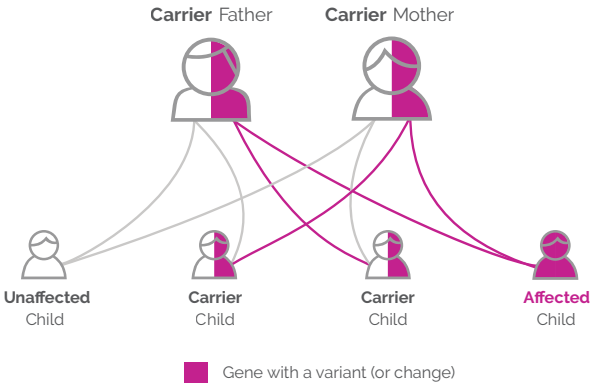
What are my options if I am a carrier?

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 healthcare provider or genetic counselor can provide information and support to help you understand your reproductive options and make important family planning decisions.

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.

If I receive a negative result, does that mean I have zero risk of being a carrier?

A negative result does not guarantee that you are not a carrier, due to the possibility of residual risk. Residual risk is your chance of being a carrier even though you received a negative result. It is based on your ancestry, as well as the ability of genetic testing technology to identify disease-causing variants in a particular gene.

Sema4 Elements™ ECS can identify ancestry information from your DNA, which is used to calculate the most accurate personalized residual risk. This ancestry information is not visible to your provider. Based on your specific ancestry, Sema4 can determine how likely we are to detect disease-causing variants in a particular gene, and provide you and your provider with your personalized residual risk information.

Why should I choose Sema4 Elements™ ECS?

Sema4 Elements™ ECS 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 Elements™ ECS is comprehensive, with various panel options that can screen for up to 502 genes, including cystic fibrosis, spinal muscular atrophy, fragile X syndrome, Duchenne muscular dystrophy, and congenital adrenal hypoplasia. For the full list of conditions that this test screens for, please visit **sema4.com.**

Accurate

When assessing your risk, precision matters. Sema4 Elements™ ECS uses advanced sequencing technologies that are >99% 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:

Expanded Carrier Screen
Our most comprehensive carrier screening panel, which tests for more than 500 genetic conditions to help guide your family planning.

Comprehensive Jewish carrier screen
This panel of 101 genes includes 47 genes relevant to the Ashkenazim, 37 genes relevant to the Sephardi and Mizrahi communities, and 17 genes common to all people of Jewish descent.

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.

Sema4 also offers other carrier screening panels, including a custom carrier screen that can test for one or more diseases. To learn more about which carrier screening panel may be right for you, please talk to your healthcare provider.

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