Quick Facts

- If you are found to be a carrier of a condition, your relatives may also be carriers of the same condition
- Complimentary post-test counseling is available
- This test is covered by most insurance plans
- The Genetic Information Nondiscrimination Act (GINA) protects individuals from discrimination based on genetic testing results and genetic information

Benefits of Carrier Screening

Provides information to make informed decisions regarding:

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

Inherited Diseases Covered by ASPIRA GENETIX Screening

ASPIRA's Targeted panel includes:

- Cystic Fibrosis
- Fragile X Syndrome
- Sickle Cell Anemia
- Spinal Muscular Atrophy
- Tay-Sachs
- Thalassemia (Alpha & Beta)
- Canavan Disease
- Bloom Syndrome
- Wilson Disease

ASPIRA Labs® offers Genetic Counseling

Personalized genetic expertise for your patients

RESULTS CONSULTATION

Review test results with a genetic counselor or medical geneticist. During this phone or video session, you are provided an opportunity to discuss the results of the test and their implications.

aspirawh.com/patient-resources/

Payment Options

ASPIRA Labs® offers payment options to make our testing both affordable and accessible. We offer financial assistance and/or payment plans for costs associated with testing.

Call ASPIRA Labs® directly to discuss your options at 866.927.7472 or billing@aspirawh.com

ASPIRA WOMEN'S HEALTH™

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Carrier Screening Information for Patients

ASPIRA GENETIX BY ASPIRA LABS | ASPIRA WOMEN'S HEALTH™

Know Your Risk

Genetic conditions are caused by a change in either the chromosomes or the genes found on those chromosomes. These changes are referred to as "variants." Genetic conditions are heritable, meaning they can be passed down within a family.

Genetic conditions detected by carrier screening typically follow one of two different inheritance patterns - **Autosomal recessive or X-linked.** Children inherit two copies of every gene, one from each parent. Parents with one variant are called carriers because they "carry" the gene change but do not have the disease. Most carriers are unaware of their status because they do not have any symptoms. However, being a carrier can significantly increase the risk for your child to inherit a genetic condition.

Genetic conditions can include:

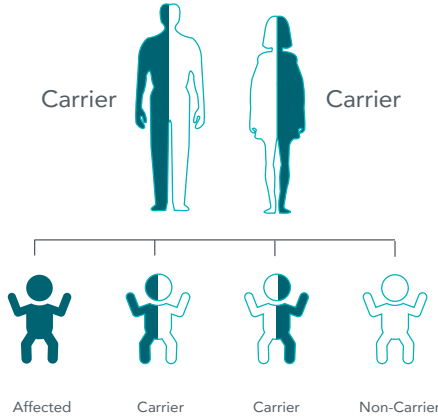
- Neuromuscular disorders
- Bleeding blood disorders
- Intellectual disabilities
- Respiratory disorders
- Metabolic conditions

The American College of Obstetricians and Gynecologists (ACOG) recommends carrier screening for all women who are planning to become pregnant. If a woman is found to be a carrier of an autosomal recessive genetic condition, testing for her partner is recommended to determine the risk that their child could have the condition.

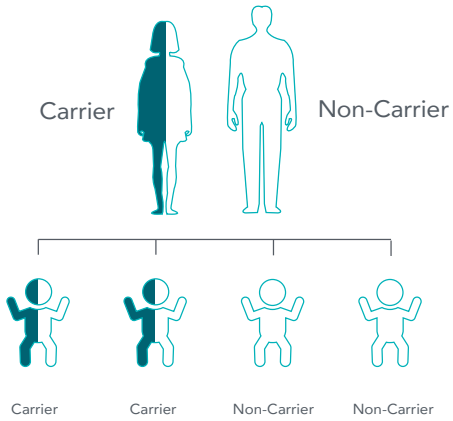
Many carriers of genetic conditions are not aware of their status because they do not have symptoms or a family history of the disease.

Autosomal Recessive Conditions

We inherit **two copies of each gene** from our parents — one from our fathers and one from our mothers. Autosomal recessive conditions occur when both parents pass down a disease-causing variant in the same gene to their child.



If both parents carry a disease-causing variant in the same gene, each time they become pregnant together, there is a 1 in 4 (25%) chance to have an affected child.

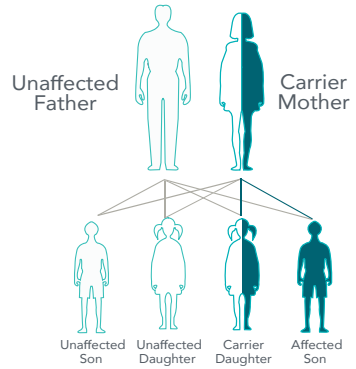


If only one parent carries a disease-causing variant, but the other parent does not, their children will not be affected. Each child they have together has a 50% chance of being a carrier of that condition.

X-Linked Conditions

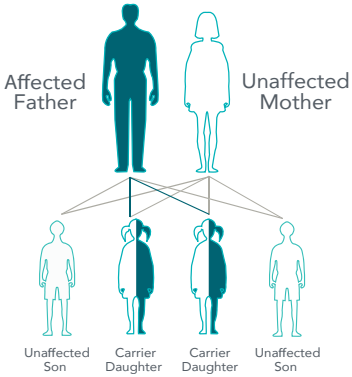
Females have two X chromosomes while males have only one. Some disease-causing variants are located on the X chromosome. Because males only have one X chromosome, when they inherit a disease-causing variant, they will be affected with the genetic condition. Females that inherit a disease-causing variant are carriers, and may experience more minor symptoms of the condition or none at all.

X-Linked Recessive Inheritance, Carrier Mother



If a woman carries a disease-causing variant on the X-chromosome, each of her children has a 1 in 2 (50%) chance to inherit the variant. Sons that inherit the variant will be affected and daughters that inherit the variant will be carriers.

X-Linked Recessive Inheritance, Affected Father



If a man is affected by an X-linked condition, all his daughters will be carriers. His sons will not be affected as they will inherit his Y chromosome, not the affected X chromosome.