

CARRIER SCREENING

WHAT IS CARRIER SCREENING?

Carrier screening, as prescribed by your Women's Care Florida provider, can detect health conditions that can be passed unknowingly from parent to child. This screen includes:

- ▶ Cystic fibrosis — affecting the lungs and pancreas, requiring lifelong treatment or lung transplantation
- ▶ Fragile X syndrome — the leading inherited cause of intellectual disabilities and autism
- ▶ Spinal muscular atrophy — the most common inherited cause of infant death

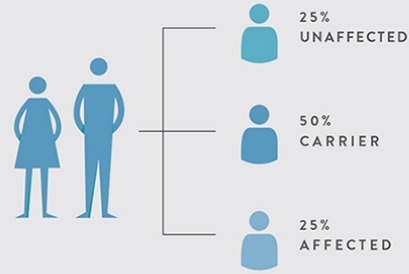
In addition to the above, patients who are of Ashkenazi Jewish descent will be screened for an additional panel of diseases, such as Tay-Sachs disease, that are more prevalent among Ashkenazi Jews.

WHO SHOULD BE SCREENED?

Physician societies recommend that pregnant women or anyone planning a pregnancy should be offered carrier screening.

I DON'T HAVE A FAMILY HISTORY OF DISEASES — SHOULD I STILL BE SCREENED?

Even without a family history of diseases, you can still be a carrier. When two people are carriers of the same disease, they can unknowingly have a child with life-long health issues. In fact, 4 out of 5 children with recessive genetic diseases are born to couples with no known family history of that disease.



Autosomal Recessive Inheritance

WHAT IS A RECESSIVE DISEASE AND WHAT IS A CARRIER?

Recessive diseases are caused by changes (called mutations) in a person's genes. Every person has two copies of each gene, one inherited from each parent. A recessive disease occurs when both copies of the same gene have a mutation.

A carrier is someone who has only one gene with a mutation and one gene that is unaffected. Carriers are typically symptom-free and do not know that they carry a mutation.

Some of these diseases are inherited differently — only the female needs to be a carrier to have a baby at risk. Fragile X syndrome is a significant example of this.

WHAT IF I FIND OUT I AM A CARRIER?

It is important for you to know that you have a 1 in 4 (or 25%) chance that your children will have a disease. When two parents are carriers of the same disease, their children have a 1 in 4 (or 25%) chance of having the disease. For certain diseases, such as Fragile X syndrome, only the mother needs to be a carrier for the child to be at high risk. Your medical professional is available to help you through the various options to find out what your risk is. Knowing your carrier status before or early in pregnancy gives you time to learn about the disorder and make decisions about your pregnancy.

WHAT IF I AM NOT A CARRIER?

Generally, no follow-up testing is suggested if you are not a carrier. It is important to understand that you are not able to identify every carrier of every disease. You should also know that while your carrier screening results provide information, we cannot screen for all genetic and genetic diseases.

Speak to your healthcare provider if you have any concerns due to family history or other factors.

HOW CAN I GET SCREENED?

Carrier screening is a simple blood test prescribed by your provider. The turnaround time for results is typically 1-2 weeks.

If you have additional questions or concerns, please discuss them with your Women's Care Florida provider.

CARRIER SCREENING for genetic health conditions

