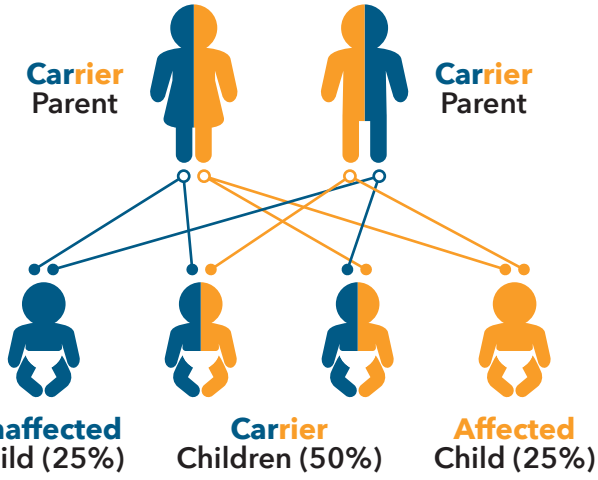
KAISER PERMANENTE®

Genetic Carrier Screening

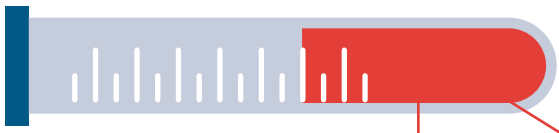
Kaiser Permanente offers testing to see if you are a carrier for certain genetic diseases.

What is a carrier?

We get thousands of genes from our parents. They come in pairs—one from each parent. Genes control traits like eye color, but they can also cause diseases if they don't work right. To have certain diseases, a baby must get a nonworking gene for the same disease from both parents.



Each parent has only one nonworking gene and does not have the disease. These parents are called "carriers." With each pregnancy, two carrier parents have a 25% chance that the baby will have the disease.

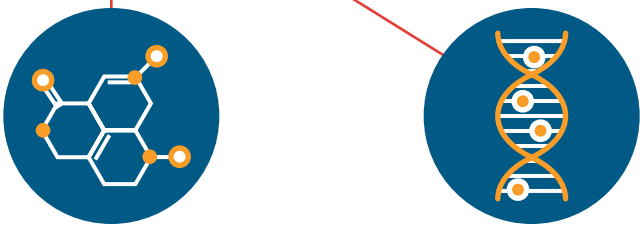


How do I know if I am a carrier?

You can find out if you're a carrier by having a blood sample taken and studied.

How accurate is the test?

For some of these tests, the accuracy of the results varies based on your ethnic background.



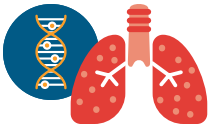
There is no test for all genetic diseases, and some are so rare that they are not usually tested for.

Who should be tested?

You will be offered carrier screening for **cystic fibrosis**, **spinal muscular atrophy**, and **inherited anemias** like **sickle cell** and **thalassemia**. If you or the father of the pregnancy are Ashkenazi (eastern European) Jewish, you will be offered specific screening such as Tay Sachs. If you are both Ashkenazi Jewish, ask your doctor about further options.

IMPORTANT TO REMEMBER

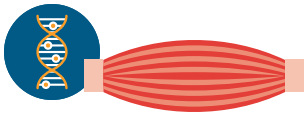
- If you are a carrier for one or more of these diseases, your partner should also be tested. If your partner is a carrier for the same disease, you should be referred for genetic counseling to talk about options for prenatal diagnostic testing.
- Talk to your doctor if you have a family history of genetic disease, if you and your partner are related to each other, or if you have more questions, as other tests may be offered.



CF (cystic fibrosis)

1 in 3,000 births[1]

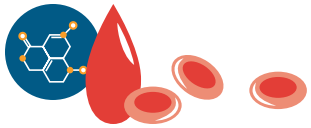
Cystic fibrosis is a disease that causes the lungs and intestines to not work right. This can lead to chronic illness, but it does not generally cause intellectual disability. The symptoms vary in severity and lifespan. CF is more common in the Caucasian population, including those of Ashkenazi Jewish background.



SMA (spinal muscular atrophy)

1 in 6,000 to 1 in 10,000 live births

Spinal muscular atrophy causes muscle weakness and is thought to be the leading genetic cause of infant death. There are different types of SMA based on the age when the symptoms show and on the severity of the symptoms. Type 1 is the most severe, and symptoms show at less than 6 months old.

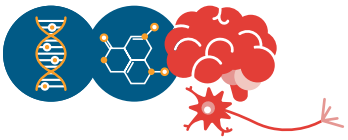


Inherited anemias

Varies

Inherited anemias (sickle cell and the thalassemias) affect red blood cells and can cause pain and organ damage. They can affect life expectancy but do not generally cause intellectual disability. Inherited anemias are more common in African-American/black, Asian, Mediterranean, and some Hispanic populations. For example, sickle cell disease occurs in around 1 in 400 births in the African-American/black population.

When both you and your partner are of Ashkenazi Jewish background, talk to your doctor or genetic counselor, as several other testing options are recommended.



Tay-Sachs disease

1 in 3,000 births[2]

Tay-Sachs disease is not obvious at birth but starts to affect the baby within the first year of life. It causes intellectual disability, the loss of other abilities, and early childhood death.

Tay-Sachs disease is also more common in the French-Canadian population. If you are from this background, discuss screening with your doctor.

Canavan disease

1 in 6,400 births[2]

Canavan disease is not obvious at birth but starts to affect the baby within the first year of life. It causes intellectual disability, the loss of other abilities, and early childhood death.

Familial dysautonomia

1 in 3,600 births[2]

Familial dysautonomia affects the part of the nervous system that controls breathing, swallowing, heart rate, blood pressure, temperature, and feeling pain. It does not cause intellectual disability.

[1]In the Caucasian population
[2]In the Ashkenazi Jewish population