

Gaucher Disease

Positive Carrier Screening Result

You have recently learned that you are a Gaucher disease (GD) carrier. Here is important information about being a GD carrier.

Being a carrier does not affect your health. Everyone carries genes that do not work. Being a GD carrier means that one copy of the Gaucher gene does not work properly. Carriers have a second copy of the Gaucher gene that works normally. GD carriers do not develop Gaucher disease. In fact, one of your parents is probably a GD carrier just like you. If you have brothers or sisters, they could also be carriers.

If you are pregnant, the next step is to test your partner. Both parents must be GD carriers to be at risk for a baby with GD. If one parent has a negative (normal) test result, the chance that a baby will have Gaucher disease is very small. Testing can be done at any Kaiser Permanente laboratory. A genetic counselor will let you know how to arrange testing.

Why test your partner?

- You want to know the chance that your baby could have GD.
- You want to be prepared if your baby is found to be at risk for GD.
- You might consider testing the baby for GD during pregnancy.

Why would you not test your partner?

- You do not want to learn about a risk for GD during pregnancy.
- The result would not change anything you do during pregnancy.

What happens if my partner is a GD carrier?

When both parents are GD carriers, there is a 1 in 4 (25%) chance for a baby to develop Gaucher disease. There is also a 3 in 4 (75%) chance that a baby will not have GD. If your partner is a GD carrier, you will be offered prenatal testing to find out if your baby could have Gaucher disease. A procedure such as amniocentesis or CVS is able to diagnose GD during pregnancy. This test allows parents to decide whether or not to continue the pregnancy. You could also choose to wait until after birth to test your baby.

What is Gaucher disease?



Gaucher disease is an inherited disease that affects many areas of the body. A person with GD does not make enough of an enzyme called glucocerebrosidase. This enzyme helps break down worn out cells in the body. When there is not enough enzyme, the cells are not broken down properly. This leads to a build up of substances in the spleen, liver, bone marrow, and other organs.



The most common form of GD is called type 1. Type 1 GD can range from mild to severe and a person may show symptoms anytime from childhood to adulthood. Symptoms often include a large liver and spleen, a low number of red blood cells (anemia), bleeding problems, and bone disease. Other types of GD (type 2 and type 3) have similar symptoms, but also include changes in the brain that get worse over time. These types of GD are rare.



There is no cure for Gaucher disease at this time. Treatment can help with some symptoms. Research is being done to find new ways to treat GD.

Gaucher Carrier Screening

Anyone can be a GD carrier, even if there is no history of GD in the family. The chance to be a GD carrier is highest for a person with Ashkenazi Jewish ancestry. For this reason, carrier screening for GD is routinely offered only when someone has Jewish ancestry. Carrier screening detects 98% of carriers in this population.



Approximate carrier rates by ancestry or ethnic background:

- Jewish: 1 in 15 (6%)
- Non-Jewish: unknown

The chance to be a carrier could be higher if there is any history of GD in the family. Before having carrier screening, it is important to let your provider know if someone in the family had GD or is a GD carrier. Special testing may be offered.

Limits of carrier screening:

- A small number of carriers have changes in the GD gene that cannot be found by routine testing. This can lead to an apparently normal result in someone who really is a carrier.

Newborn Testing for Gaucher



Testing can be requested soon after birth for babies who are at risk to develop GD. An early diagnosis can allow children to be monitored more carefully. Gaucher disease is not part of routine newborn screening in California at this time.

This information is not intended to diagnose health problems or to take the place of medical advice or care you receive from your physician or other health care professional. If you have persistent health problems, or if you have additional questions, please consult your doctor.