

Tay-Sachs Disease

Positive Carrier Screening Result

You have recently learned that you are a Tay-Sachs (TS) carrier. Here is important information about being a TS carrier.

Being a carrier does not affect your health. Everyone carries genes that do not work. Being a TS carrier means that one copy of the Tay-Sachs gene does not work properly. Carriers have a second copy of the Tay-Sachs gene that works normally. TS carriers do not develop Tay-Sachs disease. In fact, one of your parents is probably a TS 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 TS carriers to be at risk for a baby with TS. If one parent has a negative (normal) test result, the chance that a baby will have Tay-Sachs 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 TS.
- You want to be prepared if your baby is found to be at risk for TS.
- You might consider testing the baby for TS during pregnancy.

Why would you not test your partner?

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

What happens if my partner is a TS carrier?

When both parents are TS carriers, there is a 1 in 4 (25%) chance for a baby to develop Tay-Sachs disease. There is also a 3 in 4 (75%) chance that a baby will not have TS. If your partner is a TS carrier, you will be offered prenatal testing to find out if your baby could have Tay-Sachs. A procedure such as amniocentesis or CVS is able to diagnose TS 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 Tay-Sachs disease?



Tay-Sachs is an inherited disease that affects the brain. A person with TS does not make enough of an enzyme called hexosaminidase A (HexA). This enzyme normally breaks down fatty waste proteins in the brain. When there is not enough HexA, those waste proteins build up and damage the brain's nerve cells, affecting vision, movement, hearing, and learning.



Babies with TS appear normal at birth, but development starts to slow by six months. By two years of age most children with TS have seizures and begin to lose motor skills and mental functions. TS usually leads to death before age five.

There is no cure for Tay-Sachs at this time. Treatment can help with some symptoms but does not slow the disease. Research is being done to find new ways to treat TS.

Tay-Sachs Carrier Screening

Anyone can be a TS carrier, even if there is no history of TS in the family. The chance to be a TS carrier is highest for a person with Ashkenazi Jewish ancestry. For this reason, carrier screening for TS 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 25 (4%)
- Non-Jewish: 1 in 250

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

Limits of carrier screening:

- A small number of carriers have changes in the TS 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 Tay-Sachs



Testing can be requested soon after birth for babies who are at risk to develop TS. An early diagnosis can allow medical care to start as soon as possible. Tay-Sachs 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.