# Tay-Sachs Disease

## Does this test have other names?

Tay-Sachs carrier screening, Tay-Sachs prenatal testing, Tay-Sachs disease DNA analysis, Hexosaminidase A disease testing,

## What is this test?

This test looks for specific gene changes in a sample of your blood. If you are already pregnant, the test looks at a sample of the fetus' blood. These gene changes (mutations) cause most cases of Tay-Sachs disease.

Tay-Sachs disease is caused by a lack of a vital enzyme, Hex A (hexosaminidase-A). This can cause a fatty substance called GM2 ganglioside to build up in cells, especially in the brain.

Symptoms of the disease start in babies. Babies who have Tay-Sachs disease have trouble moving, develop seizures, and become blind. Children with the disease typically die before age 5. There is no cure or effective treatment for Tay-Sachs disease.

This DNA test can find out whether parents or potential parents are carriers of the disease. It can also be used to see if a fetus has the condition.

Anyone can be a carrier, but the disease is more common in people of Eastern European Jewish (Ashkenazi) descent. This means they have gene mutations that can cause the disease in a child. About 1 in 27 American Jews is a carrier. If both parents are carriers, a child has a 1 in 4 chance of getting the disease. If unaffected, the child has a 2 in 3 chance of being a carrier. Carriers do not have symptoms of the disease. Carriers can pass the defective gene to their own children.

This DNA test checks for 3 or more specific gene changes. Testing can identify nearly all carriers.

Before having this test, discuss your family background with your healthcare provider or a genetic counselor. This will help find out which type of testing you and your partner should have.

## Why do I need this test?

You may need this test if your healthcare provider wants to check whether you are a Tay-Sachs carrier. If you are pregnant, both you and your partner can be tested. If both you and your partner are known carriers, DNA testing can diagnose Tay-Sachs disease in the fetus.

## What other tests might I have along with this test?

Your healthcare provider may also order blood tests to measure Hex A and Hex B enzymes. These tests find out whether you are a carrier.

Your provider may also order screening for cystic fibrosis, Canavan disease, and familial dysautonomia. This is especially true if you are of Ashkenazi Jewish descent.

If you are not of Ashkenazi Jewish descent, your healthcare provider may also order a screening test for an additional DNA mutation that has been linked to Tay-Sachs disease in other groups.

## What do my test results mean?

Test results may vary depending on your age, gender, health history, and other things. Your test results may be different depending on the lab used. They may not mean you have a problem. Ask your healthcare provider what your test results mean for you.

Normal results are negative. This means no gene mutations were found. It means you aren't a carrier for Tay-Sachs disease. A positive result means that a gene mutation was found and that you are a carrier.

Normal results for prenatal testing are negative. This means no gene mutations were found. It means your fetus does not have Tay-Sachs disease. A positive result means that the gene mutations have been found and that your fetus does have the disease. A genetic counselor or your healthcare provider can discuss what these results mean for you and your child.

## How is this test done?

The test is done with a blood sample. A needle is used to draw blood from a vein in your arm or hand.

Testing the fetus during pregnancy needs one of these procedures:

- **Amniocentesis.** The healthcare provider takes a sample of amniotic fluid from around the baby.
- **Chorionic villus sampling.** The healthcare provider takes a sample of cells from the placenta.

## Does this test pose any risks?

Having a blood test with a needle carries some risks. These include bleeding, infection, bruising, and feeling lightheaded. When the needle pricks your arm or hand, you may feel a slight sting or pain. Afterward, the site may be sore.

Risks from amniocentesis include:

- Cramping and vaginal bleeding
- Fetal injuries
- Amniotic fluid leakage
- Infections, such as hepatitis or HIV, spreading from you to your fetus
- Slight risk of miscarriage

Risks from chorionic villus sampling include:

- Miscarriage
- Vaginal bleeding
- Leaking of fetal blood into your bloodstream. This may cause problems in later pregnancies.

## What might affect my test results?

The test is not 100% accurate, so you may have a false-negative result. This means you may be a carrier even though the test results came back negative.

## How do I get ready for this test?

You don't need to prepare for the blood test. If you are having an amniocentesis or chorionic villus sampling procedure, you may need to avoid sex or exercise on the day of the test. Be sure your healthcare provider knows about all medicines, herbs, vitamins, and supplements you take. This includes medicines that don't need a prescription and any illegal drugs you may use.