

# TAY-SACHS DISEASE

Everything you  
need  
to know about  
being a carrier

## What Does It Mean To Be A Carrier of Tay-Sachs Disease?

You've recently learned that you are a carrier for Tay-Sachs disease. You should know that being a carrier is not an illness and does not affect your health. In fact, you would probably not have found out that you are a carrier without having a special blood test. Being a carrier simply means that one copy of the gene that can cause Tay-Sachs disease has a change that keeps it from working properly. You have a second copy of the Tay-Sachs gene that is working normally.

What is a gene and what does it do? Genes are our body's "instructions" that determine our physical characteristics, such as blood type and hair color. They are located on chromosomes, which are found in almost every cell of our bodies. We inherit one set of chromosomes from each of our parents; therefore, our chromosomes, and the thousands of genes located on each of them, come in pairs.

The Tay-Sachs gene pair tells our bodies how to make a specific enzyme, called hexosaminidase A (or hex A). This enzyme breaks down a certain type of fat normally found in our brain cells. If a person has one copy of the Tay-Sachs gene that does not work properly, he or she is called a **carrier** of Tay-Sachs disease. As long as a person has one working copy of this gene, the body has enough hex A to do its job, which is why he/she will never have Tay-Sachs disease.

What is Tay-Sachs disease?

When babies inherit two nonworking copies of the Tay-Sachs gene, they are unable to make hex A. Without this enzyme the fat, which is usually broken down by hex A, begins to build up and gradually destroys brain and nerve cells until the entire central nervous system stops working. Babies appear normal until about six months old. However, once the disease begins, there is rapid deterioration. Death occurs between 3 to 5 years of life.

How does one become a carrier?

Since all of our genes are inherited from our parents on their chromosomes, individuals who are Tay-Sachs disease carriers have inherited their non-working gene from one of their parents. This means that brothers and sisters of a carrier have a 50% chance to be a carrier.

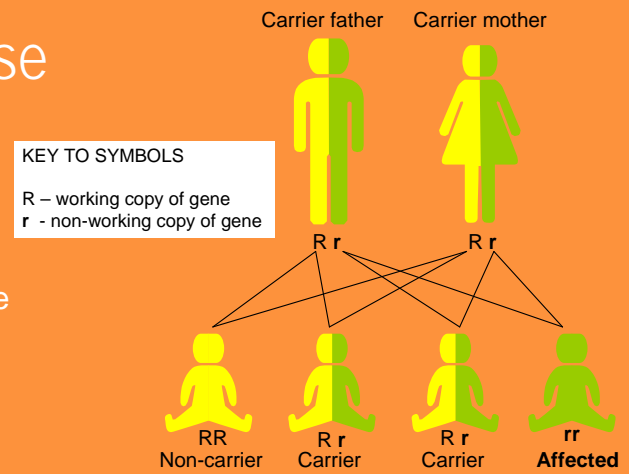
Can anyone be a carrier of Tay-Sachs disease?

Yes. Tay-Sachs carriers can be found in any nationality and ethnic group but are much more commonly found in Jewish individuals whose ancestors came from Eastern Europe (Ashkenazi Jews). In the Eastern European Jewish population, about one in every 30 individuals carries a change in the Tay-Sachs gene.

# Inheritance of Tay-Sachs Disease

When both parents are Tay-Sachs carriers there is:

- ❖ A 25% chance that the baby is not a Tay-Sachs carrier and does not have Tay-Sachs disease.
- ❖ A 50% chance that the baby is a Tay-Sachs carrier just like his or her parents.
- ❖ A 25% chance that the baby has Tay-Sachs disease.



Is there a cure for Tay-Sachs disease?

No. Although research is being conducted, there is nothing that can be done to change the course of this disease at this time. The treatment of Tay-Sachs disease is aimed at easing the problems associated with the disease.

Can being a carrier lead to having Tay-Sachs disease?

No. Carriers will never develop the disease because they have one working copy of the hex A gene. Their bodies make enough of the enzyme to be healthy.

Can my children have Tay-Sachs disease?

Since children receive half of their genetic information, like eye and hair color, from their mother and half from their father, the answer to this question depends on whether or not your partner is a carrier.

If your partner is not a Tay-Sachs carrier, then your children will not have Tay-Sachs disease. With each pregnancy, you will have a 50% chance of having a child who is a Tay-Sachs carrier just like you and a 50% chance of having a child with two working copies of the hex A gene. None of these children will have Tay-Sachs disease.

If your partner also is a Tay-Sachs carrier, then there are three possible pregnancy outcomes, including the possibility of a child with Tay-Sachs disease (see diagram above).

To find out the chance of having a baby with Tay-Sachs disease, your partner should be tested.

What choices do I have if my partner is also a carrier of Tay-Sachs disease?

There are several options available if both you and your partner are carriers. In order to find out before birth if a baby has Tay-Sachs disease, prenatal testing can be performed as early as the 10<sup>th</sup> week of pregnancy. If the results are normal, the parents can be reassured. If the results show that the baby will be affected, the parents can be better prepared, and they can make informed decisions regarding continuation or termination of the pregnancy.

Will my child be tested for Tay-Sachs disease at birth?

Although babies born in California have newborn screening for many different genetic disorders, babies are not routinely tested for Tay-Sachs disease. Carrier testing in the parents allows families to know if their baby may be at risk for this condition.

## Contact information for KP GENETICS DEPARTMENTS

Fresno: (559) 324-5330  
Oakland: (510) 752-6298  
Sacramento: (916) 614-4075  
San Francisco: (415) 833-2998  
San Jose: (408) 972-3300

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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.

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