

Introduction

Tay-Sachs disease is a rare, inherited disorder. It causes too much of a fatty substance to build up in tissues and nerve cells of the brain.

As nerve cells become swollen with fatty material, mental and physical abilities get worse. This may cause a person to go blind and deaf over time. Muscles may waste away and cause paralysis. Children with Tay-Sachs disease usually die by age 4.

This reference summary explains what Tay-Sachs disease is and what causes it. It also covers the symptoms of Tay-Sachs, how it is diagnosed and treatment options.



What is Tay-Sachs Disease?

Tay-Sachs disease, or TSD, is a fatal genetic disorder. It results in the destruction of the nervous system over time. It happens most often in children. Rarely, it can happen in adults.

TSD is caused by the lack of an important enzyme in the body. An enzyme is a substance that helps start or speed up a chemical reaction in the body. Without the enzyme, a fatty substance known as a lipid collects in certain cells of the body.

The fatty substance collects in the nerve cells of the brain. This can cause damage. Over time, damaged cells may cause symptoms. Symptoms include the loss of mental and physical abilities.



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Infantile, or early onset, Tay-Sachs disease is caused by fatty substances that collect in nerve cells when a person is a fetus. Children born with this have badly affected nervous systems and death often happens by age four. A much rarer form of TSD is called late-onset Tay-Sachs disease. It affects adults. It also causes problems with moving and thinking.

Symptoms

Tay-Sachs disease may cause the following symptoms:

- Blindness.
- Deafness.
- Delayed mental skills.
- Delayed social skills.
- Dementia.
- Increased startle reaction.

It may also cause:

- Irritability.
- Listlessness.
- Loss of motor skills.
- Loss of muscle strength.
- Paralysis or loss of muscle function.
- Seizures.
- Slow growth.



Children born with TSD develop normally during the first few months of life. Symptoms usually appear at 3 to 6 months of age. They tend to get worse quickly.

Children born with TSD lose their ability to see, hear and move as they age. Children develop a red spot in the back of their eyes. Children with TSD will stop smiling, crawling, turning over and reaching out for things. By the age of 2, children may have seizures and become completely disabled. Death usually happens by the age of 4.

In rare forms of the disease, problems may not appear until the ages of 2 to 5. This form of TSD progresses more slowly. But death usually happens by age 15. In another, milder form of TSD, the disease causes muscle weakness and slurred speech. But sight, hearing and mental capabilities remain intact.

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Causes

TSD is rare in the general population. TSD is caused by a defect in the gene known as the HEXA gene. It is responsible for the production of an enzyme that prevents the build-up of fatty proteins. As fatty proteins build up in the brain, they hurt the baby's sight, hearing, movement and mental development.

A child can only get TSD by inheriting it. This means that the gene defect is passed down from a child's parents. Carriers of TSD are people who have one copy of the inactive gene along with one copy of the active gene. These people are healthy. They do not have TSD but they may pass on the faulty gene to their children.



Carriers have a 50 percent chance of passing on the defective gene to their children. A child who inherits one inactive gene is a TSD carrier like the parent. If both parents are carriers and their child inherits the defective Hex-A gene from each of them, the child will have TSD.

If both parents are carriers of the defective gene that causes TSD, their child has a:

- 25 percent chance of having TSD.
- 50 percent chance of being a carrier.

Diagnosis

To diagnose the disease, a health care provider will do a physical exam. He or she will also ask questions about your child and your personal and family health history. A health care provider can identify Tay-Sachs carriers using a blood test.



If both parents are carriers, they may want to talk with a genetic counselor. This can help them decide if they should have a baby or if they want to test the fetus for TSD. There are several prenatal tests that can be done to diagnose TSD. Talk with your healthcare provider about what tests are available for you.

Assisted reproductive therapy is an option for carrier couples who don't want to risk giving birth to a child with TSD. Embryos created in-vitro are tested for TSD genetic mutations before being implanted into the mother. This allows only healthy embryos to be selected.

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Treatment

There is no cure for Tay-Sachs disease. Instead, the goal of treatment is to control symptoms.

The following treatments may help control the symptoms of TSD:

- Medication.
- Respiratory care.
- Feeding tubes.
- Physical therapy.

Medications may be used to control the symptoms of TSD. Respiratory care is often needed to help prevent lung infections. Lung infections cause breathing problems when a large amount of mucus forms in the lungs. Respiratory care tries to remove the mucus to promote better breathing.



A health care provider may recommend the use of a feeding tube. A feeding tube can prevent problems related to inhaling food or liquid into the lungs while eating. During a tube feeding, nutrients pass through a tube that is inserted all the way to the stomach or intestines. The tube may be placed through the nose or directly into the stomach.

Physical therapy is exercise and activities that keep the joints flexible. This can help a person with TSD keep as much of their ability to move as possible. It can delay joint stiffness. It can also reduce the loss of function and pain in the muscles.

If your child has been diagnosed with TSD or both you and your partner are carriers of the gene, talk to your health care provider about ongoing research. The stress of raising a child with a terminal illness may be eased by joining support groups. Members can share common experiences and problems. You can also reach out to family members and friends to help you cope.

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Summary

Tay-Sachs disease, or TSD, is a fatal genetic disorder that results in the destruction of the nervous system over time. It happens most often in children. TSD is caused by the lack of an important enzyme in the body. Without the enzyme, a fatty substance known as lipid collects in certain cells of the body.

TSD is caused by a defect in a gene. The gene is responsible for the production of the enzyme HEXA. Most people have two copies of this gene. If either or both HEXA genes are active, the body produces enough of the enzyme to prevent the build-up of the fatty proteins.

A health care provider can identify Tay-Sachs carriers using a blood test. There is no cure for Tay-Sachs disease. Instead, the goal of treatment is to control symptoms.

The stress of raising a child with a terminal illness may be eased by joining support groups. Members can share common experiences and problems. You can also reach out to family members and friends to help you cope.



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