# Tay-Sachs Disease

Tay-Sachs Disease (TSD) is a rare disease that causes an abnormal build up of fatty material (*lipid*) in the body, especially in the nerves of the brain. Because this abnormal lipid build up increases over time, TSD is known as a *storage disease*. There are three kinds of TSD:

- **Infantile.**
- **Juvenile.**
- **Late-onset.**

## CAUSES

TSD is caused by lack of an important substance that helps body cells to work (*enzyme*). This enzyme is called Hexosaminidase-A (Hex-A). When Hex-A does not work properly, lipid builds up inside nerve cells. This build up damages nerve cells and prevents them from functioning normally.

### Inheritance

TSD is a genetic disease. This means that the disease is caused by a change in a gene, or instruction, that is inherited from both parents. The type of TSD, when it starts and how severe it is, is based on how much Hex-A is working inside the nerve cells.

## SYMPTOMS

### Infantile

Babies with infantile TSD usually seem normal at birth and for the first 6 months. After that symptoms slowly begin to show up, including:

- Slowing of development.
- Low muscle tone, inability to hold up the head.
- Seizures (*convulsions*).
- Loss of previous abilities.
- Decrease in mental function.
- Blindness.
- Breathing and swallowing problems.
- Weakness, decreased movement and paralysis.
- Lack of response to the environment.
- Hearing problems.

Infantile TSD is usually fatal by age 5 years.

### Juvenile

Juvenile TSD that shows up between age 2 and 5 has the same symptoms as infantile TSD. Juvenile TSD develops more slowly. Juvenile TSD is usually fatal by the late teens or early 20's.

Juvenile TSD that shows up after age 5 tends to be milder. Symptoms include:

- Coordination problems.
- Slurred speech.
- Muscle weakness.
- Tremors.
- Walking problems.
- Muscle cramps.
- Mental illness.

### Late-onset

Late-onset Tay-Sachs (LOTS) usually shows up in adolescents. The symptoms of LOTS are similar to Juvenile TSD. The muscle weakness is often in the muscles closest to the trunk (upper leg muscles and upper arm muscles). Muscle twitching is common. Little is known about this rare and newly recognized form of TSD. Life

expectancy is probably normal.

## DIAGNOSIS

Diagnosis of TSD is made by a blood test that either measures the amount of Hex-A in the blood or that finds the problem in the parent's genes. An eye exam can also help diagnose TSD. Parents, brothers, sisters and other relatives of those with TSD can be tested to determine if they are carriers. Carriers do not develop Tay-Sachs disease. Carrier testing is recommended before pregnancy or in the early stages of pregnancy. Carrier testing is often offered to pregnant women, especially to couples of Jewish, French Canadian or Cajun heritage, because these groups have more TSD carriers.

## TREATMENT

There is no treatment for TSD. Research is being done. Management of TSD depends on the symptoms. Treatment is best done by a team of specialists experienced in TSD and related diseases.

## HOME CARE INSTRUCTIONS

Medical care is focused on keeping the patient with infantile TSD comfortable and well nourished in the least restrictive environment possible. In infantile TSD, nutrition may be poor. Even with feeding tubes, weight gain is slow. Pneumonia and other infections are common. Over time, seizures become more frequent and more difficult to control. Affected individuals eventually become unresponsive and do not move.

A written management plan should be agreed upon between the parents and medical staff to deliver care that is right for the child including if and when to seek hospitalization. As this is a lethal disorder, mechanical ventilation and cardio-pulmonary resuscitation are not recommended. Antibiotics may be used for infections. Medicine can be used to treat seizures and reduce pain to improve the quality of life.

In infantile TSD, medical care may be arranged in the home with the support of a coordinated group of skilled medical professionals including a:

- Pediatrician.
- Dietician.
- Nurse.
- Other therapists (respiratory, physical) as needed.

A hospice team should also be available. This medical team should work in a coordinated way to support the family and make the child comfortable as muscle and nerve functions decline. In the early stages of infantile TSD, parents can be taught tube feeding and suctioning of oral secretions. As the disorder progresses, skilled nursing assistance is needed around the clock to give medications, respond to seizures and control oral secretions. In some cases, children are cared for in skilled nursing facilities rather than at home in the later stages of the disorder.

## SEEK MEDICAL CARE IF:

- A change in mental status.
- Signs of upper respiratory infection such as cough, runny or stuffy nose, rapid breathing.
- A change in bowel or bladder habits, such as vomiting or diarrhea.
- Fever over 100° F that lasts longer than 24 hours and is unresponsive to routine therapy.
- An increase in seizure frequency or duration.
- Pain or tenderness in the abdomen or elsewhere.

## SEEK IMMEDIATE MEDICAL CARE IF:

- Sudden change in consciousness, lack of responsiveness.
- Seizures lasting more than 15 minutes that cannot be controlled by routine medications.
- Fever over 101° F (38.3 ° C).
- Respiratory distress including fast or labored respirations.

- Blue color around the mouth or finger tips.
- Uncontrolled pain.

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