



## What is Tay-Sachs Disease?

**Tay-Sachs is...** an inherited progressive neurological genetic disorder that appears in three forms: Classic Infantile, Juvenile and Late Onset or Chronic Tay-Sachs. Like most storage disorders, Tay-Sachs disease occurs along a continuum. From Infantile, the most severe form in which there is complete absence of activity of the Hex-A enzyme, to Juvenile, which affects older children, to Late Onset Tay-Sachs (LOTS), impacting even older children, teens and adults who have very low or moderately low levels of Hex-A.

### What are the symptoms?

#### *Classic Infantile*

A baby with Classic Infantile Tay-Sachs appears to develop normally for the first few months. Then, as the nerve cells begin to be affected, development slows and the baby gradually regresses, losing skills one by one and is eventually unable to turn over, sit or reach out. Other symptoms include increasing loss of motor control, progressive inability to swallow and breathing difficulties. Eventually the child becomes unresponsive to his or her environment. To date, there is no cure or effective treatment for Tay-Sachs Disease. The sad reality is, that even with the best of care, all children with Classic Infantile Tay-Sachs die in early childhood; the average lifespan is 5 years.

#### *Juvenile*

Children with Juvenile Hex-A deficiency develop symptoms as young as 2 years old or later — even in their early teens. Symptoms of Juvenile Tay-Sachs vary greatly from child to child. Generally, the age of onset indicates severity and course. An affected child with symptom onset at age 3 will experience a more severe and aggressive disease progression similar to Infantile Tay-Sachs, than an affected child with symptom onset at age 13, which can closely resemble Late Onset Tay-Sachs. Symptoms include progressive loss of motor function, which can appear as unsteady gait and clumsiness, swallowing and respiratory issues and, in rare cases, symptoms of manic depression or psychotic episodes. Older affected children who are aware of the

changes occurring in their bodies experience a wide range of emotions and are often socially ostracized.

### **Late Onset**

The symptoms typically present in adolescence, with slurred speech, muscle weakness, tremor and loss of coordination. Muscle cramps, especially in the legs at night, and twitching are common. Not all symptoms are present in every individual affected by the disease; muscle weakness, however, is a symptom common to all. Symptoms of chronic Hex-A deficiency also may begin as early as age 5, but are far milder than those that characterize the infantile and juvenile forms. Mental abilities, vision and hearing remain intact, but affected individuals develop slurred speech, muscle weakness, muscle cramps, tremors, unsteady gait and sometimes mental illness. Symptoms of manic-depression or psychotic episodes may be present in about 30% of affected persons.

As a recently recognized rare disorder, the course of LOTS is not completely known. Life expectancy is probably not reduced. Affected individuals understandably report feelings of isolation and uncertainty as to what to expect as the disease progresses. Social and academic difficulties, resulting from the physical and psychiatric manifestations of the disease, are also a concern.

## **Am I at risk of being a Tay-Sachs carrier?**

- 1 in 27 individuals of Ashkenazi Eastern European, Jewish, French-Canadian and Cajun descent;
- 1 in 36 non-Jewish individuals from the British Isles;\*
- 1 in 250 of the general population is a carrier of the Tay-Sachs gene.

\* Branda, K., Tomczak, J. and Natowicz, M. (2004) Heterozygosity for Tay-Sachs and Sandhoff Diseases in Non-Jewish Americans with Ancestry from Ireland, Great Britain, or Italy. *Genetic Testing*, 8, 174-180.



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