

BE RARE AWARE



What is Tay-Sachs Disease?

Tay-Sachs is a rare, genetic disease that is passed onto a child if both parents carry the “faulty” gene. (There is a 25% chance with each pregnancy when both parents are carriers.) The enzyme needed to breakdown the waste brain cells produce is missing, or is very low, leading to an accumulation of waste which leads to brain cell death.

What happens?

The infantile and juvenile forms of Tay-Sachs are cruelly progressive, leading to death. Adults with the late onset form progressively lose their independence, ability to walk without assistance or at all, and some struggle with their ability to talk. Diagnosis often takes up to 10 years as adults are often first misdiagnosed with ALS, MS, or Parkinson’s disease.

What are the symptoms?

Children with the infantile form lose the ability to crawl, play with toys, and lift their head. They become sensitive to sound and are startled at the slightest noise. As the disease progresses, children suffer from seizures and lose the ability to safely swallow leading to an early death.

Children with the juvenile form of Tay-Sachs lose their fine motor skills and experience a gradual loss of speech and the ability to walk and run between two and five years of age. Life expectancy is typically limited to their early teens due to complications following the loss of the ability to swallow.

Adults with the Late Onset form begin to show symptoms in early adolescence, including clumsiness and heightened anxiety. Some people experience mood disorders such as schizophrenia or bipolar disease in their late teens/early adulthood. The disease causes increased weakness in the triceps and quadricep muscles, making it difficult to get up from a chair or off the floor. Walking independently becomes difficult leading to dependence on a walker or for some a wheelchair. For some adults, speech is affected due to the disease’s impact on the muscles controlling speech. In more severe cases, adults also struggle with the ability to swallow.

How can you help?

Support families affected with Tay-Sachs by making a gift to NTSAD at www.ntsad.org. Share this post and be a voice for the voiceless.

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TAY-SACHS DISEASE VISIT
WWW.NTSAD.ORG

