BE RARE AWARE

What is Sandhoff Disease?

Sandhoff is a genetic disease 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 Sandhoff are cruelly progressive and life-limiting. Adults with the late onset form experience symptoms that can affect their day-to-day life. Diagnosis often takes up to 10 years and often adults are initially misdiagnosed with ALS, MS, or Parkinson’s disease.

What are the symptoms?

Children with the infantile form often lose the ability to crawl, play with toys, and lift their head. They become sensitive to sound and are startled at the slightest noise. Symptoms progressively get worse including seizures and losing the ability to swallow safely leading to eventual death.

Children with the juvenile form of Sandhoff 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 the early teens due to complications following the loss of the ability to swallow.

Adults with Late Onset begin to show symptoms in early adolescence – clumsiness being the most common. The disease causes increased muscle weakness, making it difficult to walk any long distance and negotiating uneven surfaces leading to tripping and falling. Some adults report tingling in their hands and feet, along with some loss in cognitive functioning.

How can you help?

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

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SANDHOFF DISEASE VISIT
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