BE RARE AWARE

What is GMI Gangliosidosis?

GMI Gangliosidosis 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 produced by brain cells is missing, or the amount of enzyme is extremely low, leading to an accumulation of waste which leads to brain cell death.

What happens?

The infantile and juvenile forms of GMI are cruelly progressive and life-limiting. Adults with the late onset form progressively become dependent on others, losing their ability to walk independently, to talk, and to take care of themselves. It is extremely rare with only a few cases in the world.

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 eventually lose the ability between the ages of 2-5 years to walk, run, and they lose their fine motor skills along with their speech. Life expectancy varies with some people living well into their twenties.

Adults with Late Onset begin to show symptoms in early adolescence, including clumsiness, heightened anxiety, and weakening of their bones. Walking independently becomes extremely difficult leading to the need for a wheelchair.

How can you help?

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

FOR MORE INFORMATION ABOUT
GMI GANGLIOSIDOSIS VISIT
WWW.NTSAD.ORG

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