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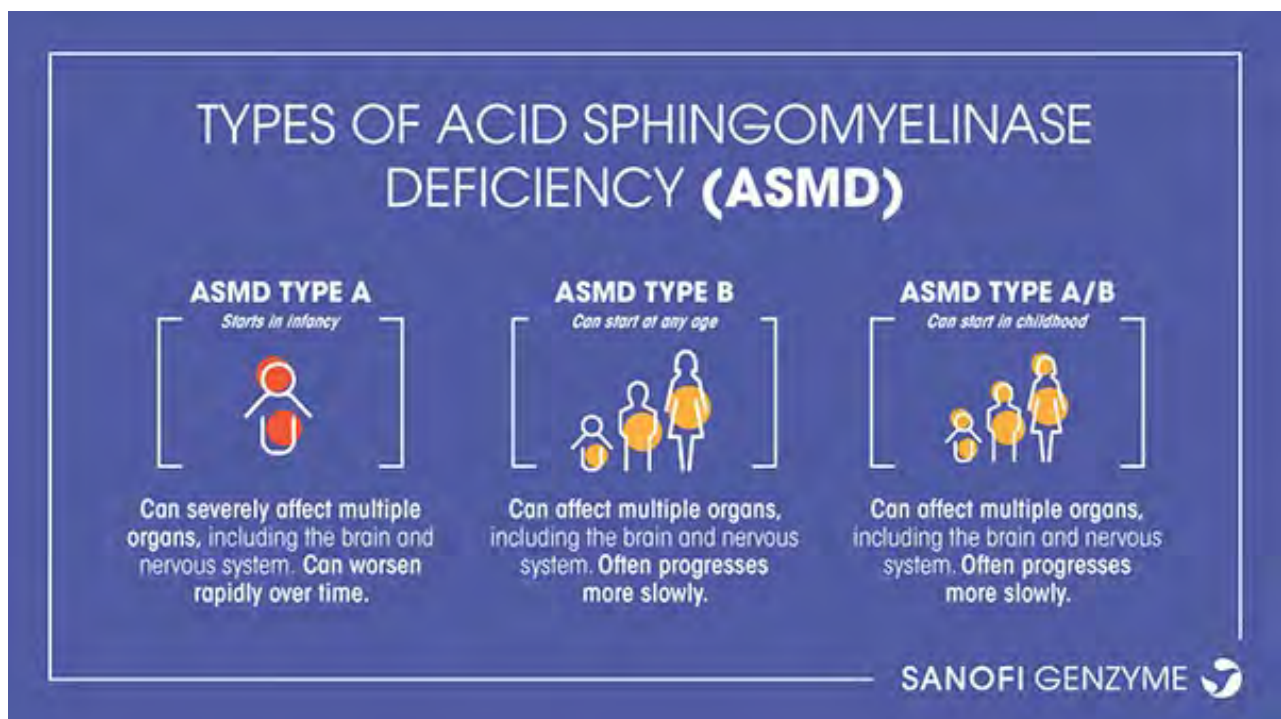
Invisible Threads: Uncovering the Purpose of Community Connections

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There are certain watershed moments in every person's life that propel them to find a community. From a neighborhood or school community to a network with a shared bond of life-changing experiences like a medical diagnosis, being part of a community can foster feelings of connection. For Isabelle and Garrett's families, connecting to others living with a rare disease offered answers, courage and hope.


Isabelle and Garrett were each diagnosed at a young age with acid sphingomyelinase deficiency (ASMD). Historically known as Niemann-Pick Disease, ASMD is a rare genetic disease that impacts multiple organs in the body, sometimes including the brain. Symptoms can include an enlarged spleen or liver, difficulty breathing, lung infections, heart problems, chronic fatigue and unusual bruising or bleeding.

There are different types of ASMD. Type A starts in infancy and can affect multiple organs, including the brain and nervous system. Type A/B can start from infancy to childhood with a varied progression rate and varied severity. Type B can start at any age and progresses more slowly than other forms of the disease and can impact multiple organs, with little to no involvement on the brain.



TYPES OF ACID SPHINGOMYELINASE DEFICIENCY (ASMD)

ASMD TYPE A	ASMD TYPE B	ASMD TYPE A/B
<i>Starts in infancy</i>	<i>Can start at any age</i>	<i>Can start in childhood</i>
		
Can severely affect multiple organs, including the brain and nervous system. Can worsen rapidly over time.	Can affect multiple organs, including the brain and nervous system. Often progresses more slowly.	Can affect multiple organs, including the brain and nervous system. Often progresses more slowly.

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For the families of Isabelle and Garrett, the diagnosis of ASMD initially brought more questions than answers. Neither family knew at the time of diagnosis that ASMD would lead them to connect with other families from across the country who would become a deep well of insight and support. These families found each other and formed a strong connection – a close-knit community to rely on while coping with a difficult disease.

Fighting for Answers: Isabelle's Story



Around five years ago when Isabelle was only one year old, her doctor voiced concern about her lack of growth and distended stomach – all of which weighed heavily on Isabelle's mom, Amy. Isabelle's doctors first ruled out food allergies. By 15 months old, Isabelle was still unable to walk without assistance, so she started physical therapy. The family endured countless visits to children's hospitals, seeing cardiologists, pulmonologists and metabolic experts, but kept receiving negative result after negative result. They had no confirmed diagnosis and no clear answers.

Isabelle's parents were steadfast in their resolve to leave no stone – or possible diagnosis – unturned. Amy started to research. Day and night over the course of several months, she continued her research. During a deep dive into metabolic disorders, Amy came across information about ASMD and saw several pictures of children with distended stomachs who resembled Isabelle. She made the connection and decided to get a second opinion from a well-known pediatrician. She pushed for genetic testing, even though the pediatrician stressed ASMD was near the bottom of the list of possibilities. Eventually, a genetic test was conducted, and exactly six weeks later (two days after Isabelle's second birthday), she was diagnosed with ASMD type B.

"I encourage any parent who has a strong feeling about their child's health to listen to their gut. Do your own research, push for answers and never stop advocating for your child," advises Amy.

Following the diagnosis, Isabelle's family connected with others in the ASMD community. Becoming a part of an ASMD community helped Isabelle's family feel less alone by hearing the personal stories of the other families and exchanging information on social media. The family now shares its story, helping to provide hope to others living with ASMD.

"Our path fortunately led us to a small, yet mighty community and support system for people with ASMD. Finding other families with children who had ASMD helped our family cope with Belle's diagnosis," said Amy. "Staying connected to those who have been with us throughout our ASMD journey has proved extremely valuable, and we are grateful for the hope and constant support."

Finding Strength in Friendships: Garrett's Story



When Garrett was born, he was healthy and thriving. Around six months old, his parents began to notice he wasn't gaining weight like his brother and sister did at the same age. When he was one year old, his inability to gain weight, poor sleep and poor motor skills led the family back to his pediatrician who found Garrett had an enlarged spleen. After a series of blood tests, Garrett was diagnosed with ASMD type A.

When Garrett's parents learned of his diagnosis, their first reaction was to retreat.

"We're private people, so we kept the diagnosis to ourselves initially – only sharing our news with close friends and family," said Garrett's father, Justin. "When you're told your one-year-old son will likely only live a few years, enjoying him while he's still here becomes priority above all else."

But, once Garrett began hitting some developmental milestones, his family's instinct to reach out and connect with others kicked in. Soon, Garrett and his family began attending conferences and becoming actively involved across multiple ASMD organizations and advocacy groups.

During one such conference, Justin connected with an ASMD expert, who noticed Garrett swinging a golf club in the back of the room during a presentation. The physician pointed it out to the family and told them his ability to do that was a clear indicator that he had ASMD type B as opposed to ASMD type A. This fortunate, unexpected encounter dramatically altered the disease management approach recommended by Garrett's care team. While the journey has been intense and emotional, the ASMD community has been a constant source of support for Garrett and his family.

"Being connected with our ASMD network has given us a sense of positivity, access to education and loving families who know exactly what we're dealing with on a day-to-day basis," said Justin. "The shared experience has been a great help in managing our process."

Connecting with a Community

Living on opposite sides of the country, the likelihood that these two very similar families of five would cross paths was very low. But a community bound by a shared rare disease diagnosis did just that. Both families are inspired to share their stories and bring others into this supportive community.

Isabelle's mom Amy has become a blogger for the National Niemann-Pick Disease Foundation (NNPDF), and Garrett's dad Justin has taken on a leadership role as NNPDF Board Chair. Today, Isabelle is an energetic, independent 6 year old; and Garrett is a joyful, sports-loving 10 year old.

There is power in finding a community. A community provides access to deep, personal connections to others who truly understand the shared struggles and celebrations. A community also brings valuable connections to resources – which can be critical in informing the best course of action for people living with a rare disease. For Garrett and Isabelle's families, the connection to the ASMD community represents friendship and knowledge – and in both, there is hope.