Real People
Real Urgency

Making an Early Diagnosis in Rare Diseases Can Make a Meaningful Difference

Child's diagnosis:	**Symptoms first noticed:**	**Diagnosis received:**
Sandhoff Disease	Seven to eight months	One year

How was your child diagnosed?

Genetic testing (unofficial diagnosis was made in May 2018, based on clinical presentation).

How old was your child at the time symptoms began?

Between seven to eight months of age, Embree lost her ability to sit up and was getting more "floppy" in her trunk and arms. She also was never able to push herself up on her arms.

What were the signs that triggered concern?

Muscle weakness, difficulty eating, and vision problems.

Did you share your concerns with your pediatrician?

Yes.

How did your pediatrician respond?

The doctor was concerned that Embree could have spinal muscular atrophy. She sent us to a neurologist and referred us to a geneticist. Unfortunately the neurologist didn't provide any insight into what could be wrong, and the wait was six months for an appointment with the geneticist.

What led to the diagnosis?

One day I put her favorite giraffe toy in front of her face and she looked past it. She used to light up and smile at it. I knew she was going blind. So we took her to the children's hospital emergency department to seek answers.

Who made the diagnosis?

A geneticist at a children's medical center.

How long was your journey to diagnosis?

Ten months.

Looking back, what advice would you give yourself?

Losing milestones is a huge red flag. Embree wasn't gaining as much strength from physical therapy, and something was very wrong. She also was having trouble eating pureed foods, and this was also a big warning sign. I wish we would have taken her to the hospital sooner, because we weren't getting answers fast enough via outpatient care.

What difference would an earlier diagnosis have made?

I feel like an earlier diagnosis would have helped us (at least) gain access to more resources earlier. We never got in-home nursing or additional healthcare coverage like Medicaid because the process was so long. Of course, gene therapy wasn't available at the time, but if it was, an earlier diagnosis would have made a difference in our decision making. However, keeping our daughter as comfortable as possible was also very important to us.

Know the signs of
Infantile Sandhoff Disease

You Could Make the Rare Dx

First signs

A baby with Classic Infantile Sandhoff appears normal at birth and typically develops normally for the first six months of age. As development slows, parents may notice a reduction in vision and tracking. The baby does not outgrow normal startle response.

Gradual loss of skills

Infantile Sandhoff children gradually regress, losing skills one by one and eventually are unable to crawl, turn over, sit, or reach out. Other symptoms include loss of coordination, progressive inability to swallow and difficulty breathing.

By age two and beyond

Most children experience recurrent seizures by age two and eventually lose muscle function, mental function, and sight, becoming mostly non-responsive to their environment.

Diagnostic pathway

Sandhoff disease is diagnosed through a blood test to check the levels of Hexosaminidase A (HexA) and Hexosaminidase B (HexB). A follow-up DNA test may be recommended. Any doctor can order the Tay-Sachs HexA blood test. Often, diagnosis is made by a neurologist or geneticist. Babies affected by the infantile form of Sandhoff are frequently diagnosed by the cherry red spot on the retina of the eye. Initially many parents notice developmental delays, but pediatricians often dismiss these concerns by stating "every baby develops differently" and "the baby will catch up." Often at about 10-14 months of age, children may start to exhibit trouble tracking and/or focusing with their eyes, so parents schedule an appointment for an eye exam. The cherry red spot is quickly seen, and an initial diagnosis of Sandhoff disease is made. Diagnosis can also be made by a neurologist or geneticists and the completion of a metabolic evaluation.

Other forms of Sandhoff

In addition to Infantile Sandhoff there are also Juvenile and Late Onset forms of the disease. Symptoms of Juvenile Sandhoff can appear after the first year of life (usually between two and five years of age). With the late onset form of the disease, symptoms can appear in adolescence or early adulthood (but also can occur later).

Risk profile

Anyone can be a carrier of Sandhoff. When both parents are carriers, each child has a 25% chance of having the disease. The carrier rate for the general population is low, approximately 1/600, and it is not yet clear whether Sandhoff disease is more common in any population, but it may have a higher carrier rate in several somewhat isolated populations.

NTSAD
National Tay-Sachs &
Allied Diseases Association

For more information, please visit NTSAD.org or call 617.277.4463.