Real People **Real Urgency**

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

Child's diagnosis:	**Symptoms first noticed:**	**Diagnosis received:**
Canavan Disease	Four months	Ten months

How was your child diagnosed?

MRI confirmed weeks later with DNA and urinalysis.

How old was your child at the time symptoms began?

At four months of age.

What were the signs that triggered concern?

Vayle was not meeting milestones. She was not pushing up on her hands in tummy time. She was not cooing or laughing a lot–lots of crying. Vayle had difficulty eating, significant growth in head size from four to six months of age, and fisted palms.

Did you share your concerns with your pediatrician?

Sort of. I was trying not to be too alarmist, so I didn't overly voice concerns.

How did your pediatrician respond?

Was told my child was okay and if she was laughing and smiling and drooling a lot at four months of age then we were developmentally on target.

What led to the diagnosis?

She was not doing what other babies at daycare could do. Vayle was not rolling or pushing up, her hands were fisted, and it was difficult to put her clothes on.

Who made the diagnosis?

Emergency department neurologist.

How long was your journey to diagnosis?

About 10 months.

Looking back, what advice would you give yourself?

Make doctors spend more time with your child at well-visits. They're not just for routine shots. Don't be afraid to speak up.

What difference would an earlier diagnosis have made?

Today it could make all the difference in the world. With a gene therapy ready to start for young babies, every day matters in treatment. A two-month delay in getting a diagnosis and missing important signs and symptoms could mean the difference between life and death.

Know the signs of **Canavan Disease**

You Could Make the Rare Dx

First signs

A baby with Canavan may appear normal at birth and typically develops normally for several months. Sometimes symptoms are noticeable at birth. As development slows, parents may notice a reduction in vision and tracking.

Gradual loss of skills

Children with Canavan 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 three and beyond

By age three, children progressively lose muscle tone and function. Some children experience recurrent seizures. Despite their inability to communicate by traditional means, many parents believe their child understands their surroundings and can speak volumes with their eyes.

Diagnostic pathway

Canavan disease is diagnosed through a combination of biochemical and DNA tests. Increased amounts of N-acetylaspartic acid (NAA) are found in the urine. Profound deficiency of aspartoacylase is found in cultured skin fibroblasts. Known disease-causing mutations are frequently found through DNA testing. Occasionally, known mutations are not found, suggesting novel (or unknown) mutations. Often an MRI or CT scan showing signs of decreased myelination leads to the Canavan diagnosis.

Risk profile

Anyone can be a carrier of Canavan. When both parents are carriers, each child has a 25% chance of having the disease. The carrier rate for the general population is 1/300. Ashkenazi Jews are at high risk with a carrier rate of 1/40.

NTSAD

National Tay-Sachs &
Allied Diseases Association

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