Real People
Real Urgency

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

Child's diagnosis:
Infantile GM1 Gangliosidosis

Symptoms first noticed:
Eight to nine months

Diagnosis received:
Fifteen months

How was your child diagnosed?

Abnormal MRI, then an eye exam revealed cherry-red spots. Enzyme and genetic testing confirmed diagnosis.

How old was your child at the time symptoms began?

At eight to nine months of age. His pediatrician said he had an abnormally large head at 4 months, but he was also an overall big baby.

What were the signs that triggered concern?

Delayed development. Joey couldn't get into a sitting position by himself. He wasn't crawling or pulling himself up to stand. No vocalizations. Nystagmus (rapid and uncontrollable eye movement) and exaggerated startle reflex.

Did you share your concerns with your pediatrician?

Yes, at his nine-month checkup, but his doctor was not concerned.

How did your pediatrician respond?

His doctor thought Joey was a late bloomer and a big baby. I don't think he had the nystagmus at nine months and don't remember how exaggerated his startle was at that point.

What led to the diagnosis?

Early intervention physical therapy. The physical therapist noticed that Joey had clonus in his feet (involuntary and rhythmic muscle contractions) and nystagmus and said we should see a neurologist. I made appointments with another neurologist (for a second opinion) and an ophthalmologist.

Who made the diagnosis?

The second neurologist.

How long was your journey to diagnosis?

About 6 months.

Looking back, what advice would you give yourself?

I honestly think I did the best I could.

What difference would an earlier diagnosis have made?

We were desperate for answers. Having a diagnosis sooner would have given us more time to better understand what was happening and what we could expect.

Know the signs of
Infantile GM1 Gangliosidosis-1

You Could Make the Rare Dx

First signs

A baby with Classic Infantile GM1 gangliosidosis displays symptoms within the first six months. Symptoms may be apparent at birth. Early symptoms include poor appetite, weak suck, and failure to thrive. Only about 50% of cases display the cherry red spot in the back of the eye.

Gradual loss of skills

Infantile GM1 gangliosidosis children never learn to sit up or crawl, have generalized poor muscle strength, demonstrate progressive inability to swallow and have difficulty breathing. Some children also have an enlarged heart, known as cardiomegaly.

By age two and beyond

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

Diagnostic pathway

GM1 gangliosidosis is diagnosed through a blood test to check the level of beta-galactosidase (GLB1). A follow-up DNA test may be recommended. Any doctor can order the GM-1 GLB1 blood test. Often, diagnosis is made by a neurologist or geneticist.

Other forms of GM1

In addition to Infantile GM1 there are also juvenile and late onset forms of the disease. Children and adults affected by GM1 gangliosidosis disease do not exhibit the telltale cherry red spot in the eye. This can make the road to diagnosis long and challenging. Unfortunately, many healthcare providers are not aware of the rare juvenile and adult forms of this disease and dismiss the initial diagnosis due to age. Adults who display mental health symptoms before physical symptoms often experience the longest road to diagnosis.

Risk profile

GM1 gangliosidosis must be inherited from parents who are carriers of the disease. Anyone can be a carrier of GM1 gangliosidosis and not have any symptoms. When both parents are carriers, each child has a 25% chance of having the disease. The carrier rate for the general population is 1/250. Some evidence suggests people of Irish/British Isle descent have an increased risk over the general population with a carrier rate between 1/50 to 1/150. French Canadians, Louisiana Cajuns, and Ashkenazi Jews are all considered high risk with a carrier rate of 1/27.

NTSAD

National Tay-Sachs & Allied Diseases Association

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