Real People **Real Urgency**

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

Child's diagnosis:	**Symptoms first noticed:**	**Diagnosis received:**
Infantile Tay-Sachs	Three months	Ten months

How was your child diagnosed?

Cherry red spot in the eye, then genetic testing to confirm.

How old was your child at the time symptoms began?

I knew from the moment she was born that something was wrong, but within the first few months that something was atypical. By three months, I began to seek answers.

What were the signs that triggered concern?

She would not hold her bottle, track us with her eyes, or imitate our speech. As time went on, she was slow to reach more of her milestones.

Did you share your concerns with your pediatrician?

Yes, over and over.

How did your pediatrician respond?

I don't feel they (my concerns) were anything disturbing enough individually or even collectively that caused them to be concerned enough to dig deeper and search for any real medical answer. I was told to keep watching and waiting. Every three months I would go in for the next well-child check hoping for confirmation of some issue/explanation or enough information to finally make my fears subside.

What led to the diagnosis?

I thought she needed intervention to help her get over the hump of her issues/hindrances. We were seeking out physical therapists and other professionals as resources to help her achieve her milestones. I just didn't stop looking for answers as to why she wasn't progressing.

Who made the diagnosis?

Ten doctors (and several misdiagnoses) later: the ophthalmologist.

How long was your journey to diagnosis?

About seven months.

Looking back, what advice would you give yourself?

I would tell myself to keep pushing. I was right that something was wrong. Much to my horror, and it was worse than I ever imagined, but my baby wasn't fine, and I wasn't just an overly concerned mother (two notes someone actually made on a form). I just never knew it could have been a neurological issue. I thought I could help her get better. I wish I wouldn't have been made to feel like my concerns were being dismissed, or at least not taken as seriously as they should have been. I wish I hadn't been made to sit back and wait so long for answers.

What difference would an earlier diagnosis have made?

The long road to diagnosis was both mentally and emotionally debilitating to me as a mother. I was living in constant fear and isolation. Earlier diagnosis would have confirmed that I wasn't crazy–as I was being made to feel, or "overly concerned" as one evaluator wrote, but it would have provided answers that would have steered us in a clear direction much sooner. It would have given us community and support when we needed it most.

Know the signs of
Classic Infantile Tay-Sachs

You Could Make the Rare Dx

First signs

A baby with classic Infantile Tay-Sachs appears normal at birth and typically continues to develop normally for the first six months of age. Around six months of age, development slows. Parents may notice a reduction in vision and tracking and the baby does not outgrow normal startle response.

Gradual loss of skills

Infantile Tay-Sachs children gradually regress, losing skills one by one. Over time they 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

Tay-Sachs disease is diagnosed through a blood test to check the level of Hexosaminidase A (HexA). 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 Tay-Sachs are frequently diagnosed by a 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 Tay-Sachs or similar devastating disease is made. Diagnosis also can be made by a neurologist or geneticists and the completion of a metabolic evaluation.

Other forms of Tay-Sachs

There are three forms of Tay-Sachs. The form is determined by the age of the individual when symptoms first appear. Only one form of Tay-Sachs occurs in a family. If a child has Infantile, older siblings are not at risk to develop Juvenile or Late-Onset Tay-Sachs later in life.

Risk profile

Anyone can be a carrier of Tay-Sachs. 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.