

Nora

THIS IS A PERSONAL STORY SHARED BY A FAMILY WHO LOST A CHILD TO
LONG-CHAIN HYDROXYACYL-COA DEHYDROGENASE DEFICIENCY (LCHAD).

Our four-month-old daughter died suddenly and unexpectedly. This did NOT have to happen. A simple newborn screening test, which most parents are not aware of, would have saved her life. Please take the time to read our story. We want to tell it in hopes of helping others.



My name is Sirpa Waananen. On March 29th I gave birth to a beautiful full-term, healthy girl. My pregnancy and delivery were normal. Nora was a very content and happy baby, developing normally. On August 9th I left her for the first time to attend work related training. My husband was also working and my sister was watching our children (we also have a two-year-old boy, Tommy). After training, I called home and told my sister that I was on my way. My sister told me that Nora appeared to be a little bit sleepy and was not her happy self. We were both concerned and so she called the doctor. My commute home seemed like an eternity. When I reached my car, my husband called and said that he was in the hospital with Nora. In the hospital, Nora quickly slipped into a coma and by the time I got there, she had already died. We were devastated!

Luckily we had a connection to the Mayo Clinic, and shortly after Nora's death we found out that she had died of a "Fatty Acid Oxidation Deficiency" (FOD). We had never even heard of such a thing. Adding to the devastation, we found that a simple newborn screening test not only would have saved Nora's life, but her prognosis would have been excellent!

Most parents' trust that with the mandatory PKU screening test, our babies are being taken care of. What most parents and doctors do not know is that through many private laboratories our babies can be tested for an additional 30+ brain damaging and/or life threatening metabolic disorders. All these disorders are treatable, but if untreated they will cause brain damage and/or death.

- Almost 3,000 US-born babies, each year, die unnecessarily, have brain damage, or come near death before diagnosis can be made clinically, due to a lack of "Comprehensive newborn screening" (CNBS).
- About one in 1,300 babies are born with some type of screenable metabolic disorder.
- Some cases of SIDS, Reye's Syndrome, Cerebral Palsy and Autism simply do not have to happen, and any

children in the past have been misdiagnosed as such.

- At least 7 or the 30+ disorders are "silent" and give no symptoms until it is too late. Usually there is no family history or warning signs. Children can be screened at any age.

To obtain a screening packet, call:

1-866-463-6436 PerkinElmer Genetics

1-800-422-9567 Baylor Medical Center

You can also contact me at the email address below for more information.

Written March 2002 by Sirpa Waananen

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Mom to Nora - LCHAD (March 29, 2001 - August 9, 2001)

Chico, California