SOFIA SEES HOPE
Supporting the LCA and rare retinal disease community.
We see a cure for blindness.

SEEING HOPE | Newsletter

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From the Founder

Twelve years ago, my daughter Sofia was clinically diagnosed with Leber congenital amaurosis (LCA). Seven years after that, she received her genetic diagnosis, IQCB1/NPHP5. At that time, there were 20 genes associated with LCA. Today there are nearly 30.

Sofia Priebe & Laura Manfre

Of course I have reacted to all of this as a mother. I want a cure for my daughter. I want her to realize her dream of seeing the stars in the night sky. I want her to see the leaves on the trees and the shells on the beach.

But I also was struck by the need to do more. As I navigated my way through the world of inherited rare diseases, and more specifically LCA, I discovered there are others like me feeling alone, with many questions and much confusion. I knew I needed to help more than my daughter.

And so in 2014, I founded Sofia Sees Hope with my husband, Chuck, and the help of friends and family. Over the last four years it has evolved from its infancy to what I now consider its adolescence. Through the support of sponsors and individuals, networking and education, we have experienced a breakthrough year in 2017.

Last year, Sofia Sees Hope launched a new website designed to be a hub

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Living with LCA: Enzo's Story

By Rosanne Smyle

Enzo was born in 2014 in Lausanne, Switzerland, with a clubfoot. He immediately received physical therapy, then a plaster cast for three months and one minor surgery.

His parents — Laura Steinbusch, a post-doctoral fellow researching the neuroscience of diabetes, and Merlijn Meens, a scientist investigating cardiovascular function — felt confident as they were reassured through medical literature and by doctors that in time their son's left foot would be fine.

But there was more to come. At 3 months, as Enzo's therapy for his foot progressed, something seemed wrong with his vision and his ability to focus. Doctors diagnosed him with nystagmus, a condition in which Enzo's eyes involuntarily moved side to side.

An MRI showed normal brain development and then doctors did an electroretinogram (ERG), placing electrodes on his eyes to measure the electric response of their light-sensitive cells. Enzo also underwent Visual Evoked Potential (VEP) testing, a non-invasive exam that measures his entire vision system.

Doctors fitted Enzo with glasses at 6 months and said the ERG and VEP results needed further study.

Enzo began rubbing his eyes, prompting people to ask Laura whether her son was tired or shy. A few months later, doctors diagnosed 10-month-old Enzo with Leber congenital amaurosis.

"While walking home from this last doctor's appointment, we were in shock, but after a while we realized that Enzo had not changed. He was still our cheerful son that likes to sing and cuddle," Laura wrote in her blog for the Eye Association of the Netherlands.

"We wondered how we could raise Enzo as normal as possible and how we could help him discover the world. The solution turned out to be simple: We will not despair and (will) come up

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