About Hope in Focus

Hope in Focus is a 501(c)(3) nonprofit organization dedicated to providing information and support to individuals and families affected by Leber congenital amaurosis and other rare inherited retinal diseases. We also raise funds for genetic testing and research to support more effective diagnosis and treatment, with the hope of an eventual cure for LCA.

The organization was co-founded in 2014 by Laura Manfre and Charles Priebe, whose daughter Sofia has LCA. At the time, it was called Sofia Sees Hope. The name was changed to Hope in Focus in 2021, to better reflect the transformation from a small NPO into a robust, international organization.

Hope in Focus offers opportunities for individuals and families to connect, share information and provide mutual support.

About Leber congenital amaurosis

Leber congenital amaurosis (LCA) is a rare inherited retinal disease (IRD) that causes the degeneration of the cone and rod cells in the retina. It is characterized by severe vision loss at birth and results in complete blindness.

LCA can be difficult to diagnose. In many cases, diagnosis involves sophisticated clinical tests and genetic testing to identify the associated genetic abnormality that causes LCA. Many experienced eye doctors are not familiar with LCA, and in many cases patient families do not know where to get information or appropriate support. LCA is estimated to occur in about 1 in 33,000 people. There are nearly

30 genes that are believed to be associated with this rare disease. A mutation in just one of these genes can result in blindness.

It is essential for all people at risk of LCA to receive genetic testing to avoid the possibility of misdiagnosis and inappropriate forms of treatment. Testing is based on a saliva or blood sample. Testing must be completed by an experienced laboratory and patients should have access to genetic counselors who can explain the results and any appropriate next steps.

Living with LCA

As a rare disease that is not widely known or understood, LCA can often create feelings of isolation, frustration and helplessness. Access to medical services for accurate diagnosis of LCA is a critical step in supporting patients and their families. Hope in Focus advocates for all patients to participate in an LCA patient registry to build broader awareness of the incidence and impact of LCA. Registries will also play a critical role in guiding future research, and establishing more effective treatment and patient management guidelines.

All families affected by LCA can benefit from opportunities to connect and share information with other families, medical specialists and advocacy groups. A vital element of the Hope In Focus mission is to provide more opportunities for all people affected by LCA to connect and provide mutual support and information.

Hope in Focus' advisors and partners include Dr. Jean Bennett from the University of Philadelphia, senior geneticist Emily Place from Mass Eye and Ear, and industry and advocacy partners including Spark Therapeutics, Editas Medicine, AGTC and Foundation Fighting Blindness, among others.

Looking to the future

In recent years there have been many exciting developments in efforts to treat IRDs. These include important advances in gene therapy, CRISPR and other cutting-edge treatments involving genetic engineering. At the end of 2017, the U.S. Food and Drug Administration approved LUXTURNA ®, the first gene therapy for a genetic disease in the United States.

Hope in Focus continues to support many innovative early-stage proof of concept research programs and calls on both government and academic research centers to continue to support this promising research in the years ahead.

To date, Hope in Focus has provided financial support for research underway at the Foundation Fighting Blindness related to the genes CEP290 and IQCB1/NPHP5. These efforts have been led by renowned researchers, including Dr. Rob Collin, Radboud University Medical Center in Nijmegen, The Netherlands, Dr. Anand Swaroop, National Eye Institute and Dr. Wolfgang Baher, The University of Utah.

Hope in Focus also provides funding to ensure that families who need genetic testing will have access to it, regardless of ability to pay.