Here is the statement sent to lawmakers from Connecticut-based Sofia Sees Hope on Rare Disease Day. Recipients included Connecticut Gov. Ned Lamont, members of the Connecticut General Assembly's Public Health Committee, and U.S. Senators Richard Blumenthal and Chris Murphy, and Rep. Joe Courtney.

Feb. 26, 2021

Leber congenital amaurosis (LCA) is characterized by severe vision loss at birth. While some children are born with little or no vision, others may have significant vision loss in the first few years of life, stable vision for a period, and then eventually complete vision loss as the retina deteriorates into total blindness.

The optimal window for reversing vision loss is during the early phase of the disease. Creating avenues to affordable treatments and accessibility to resources is imperative and often can be inhibited by insurance regulations and other rules limiting access to help and support patients.

More than 25 genes are associated with LCA and a mutation in just one of these can result in blindness. The rare disease occurs in 1 in 33,000 to 1 in 88,000 people and makes up 5 percent of all retinal dystrophies. Twenty percent of children with visual impairment and attending special schools have LCA; it is the second most common inherited retinal dystrophy after retinitis pigmentosa.

A patient needs a confirmed genetic diagnosis to proceed with appropriate treatment avenues. Sofia Sees Hope has given more than $100,000 to provide families, including those in Connecticut, free access to genetic testing and has directed $275,000 to genetic retinal research. Patients also need support from their lawmakers to ensure they receive the quality of life to which they are deserving.

After decades of research and dedicated investment in studies, scientists created a breakthrough genetic therapy that helps restore vision in patients with one of the genetic mutations causing LCA. The U.S. Food and Drug

Administration in 2017 approved this treatment – developed by Spark Therapeutics and called LUXTURNA® – which also is the first genetic therapy ever in the United States to treat ANY rare inherited disease.

LCA patients treated with LUXTURNA experienced dramatic changes in their lives with greatly improved or restored vision. Children who are 5, 6, 7 years old and have been treated with LUXTURNA view life in a new light in big and little ways. They now can see rainbows in the sky and stars shining at night.

Our lawmakers need to know that we fully support the principle that all FDA-approved treatments should be made available to all those who will benefit from such treatment, and to reject any proposed requirements restricting access to medications.

Sofia Sees Hope also encourages the Connecticut General Assembly to establish a Rare Disease Advisory Council comprised of patients, patient advocates, doctors, researchers, and community members to address the emerging public health priority of rare diseases, including LCA.

Thank you for your time and attention.

Sincerely,

Rosanne Smyle

Sofia Sees Hope