

Determining the underlying genetic causes of Optic Nerve Hypoplasia at Children's Hospital Los Angeles

Dr. Laura Li and her colleagues at the Children's Hospital Los Angeles (CHLA) are working to determine the underlying genetic causes of Optic Nerve Hypoplasia (ONH), which is still unclear. ONH is the absence or under-development of the optic nerve and is currently the leading ocular cause of vision impairments and blindness in young children. ONH can also be combined with brain and endocrine abnormalities which can affect motor skills, intelligence, speech and social interactions and cause hormone deficiencies.

To further investigate the possible genetic factors of ONH, the team at CHLA is conducting whole exome trio analysis with 25 families. The team uses NexGen for alignment and variant calling and then imports their VCF files into Golden Helix's VarSeq software. Using VarSeq, Li quickly filters and annotates her data to produce a short list of genes and variants of interest. From this short list, Li then manually interprets the data, looking for genetic explanations to ONH.

Before implementing VarSeq, Li evaluated various other programs, including a couple of commercial software solutions and internal programs. Li found the other tools to be either cumbersome or very expensive to be feasible for a small research lab. "VarSeq is pretty user-friendly", said Li. "Also the support team is very helpful. They know the VarSeq software in and out and the tech support has helped me to quickly learn VarSeq."

Dr. Li received her PhD from Johns Hopkins School of Medicine where her thesis focused on Molecular Biology and Genetics. After completing her PhD, Li went to the California Institute of Technology (Caltech) in Pasadena, California where



Dr. Laura Li

she completed a fellowship studying Bloom syndrome, an inherited disorder characterized by short stature, a skin rash that develops after exposure to the sun, and a greatly increased risk of cancer. At Caltech, Li investigated the cellular and molecular mechanisms leading to Bloom Syndrome. After leaving Caltech, Li joined

Quest Diagnostics where she worked for a few years before being accepted into the American Board of Medical Genetics and Genomics training program at the University of California Los Angeles. She later completed a two-year fellowship with the renowned geneticist Dr. Wayne Grody. After her fellowship training, Li was board certified in 2013 when she began working at the Los Angeles Children's Hospital as the Associate Director for the Clinical Genomics Laboratory.

QUICK FACTS:

Name: Laura Li, PhD

Title: Associate Director

Institution: Children's Hospital Los Angeles

Location: Los Angeles, CA

Research Focus: Optic Nerve Hypoplasia

Golden Helix Software: VarSeq



Children's Hospital Los Angeles, California



Dr. Sergey Kornilov

About Golden Helix

Golden Helix has been delivering industry leading bioinformatics solutions for the advancement of life science research and translational medicine for over 17 years. Our innovative technologies and analytic services empower scientists and healthcare professionals at all levels to derive meaning from the rapidly increasing volumes of genomic data produced from microarrays and next-generation sequencing. With our solutions, hundreds of the world's top pharmaceutical, biotech, and academic research organizations are able to harness the full potential of genomics to identify the cause of disease, improve the efficacy and safety of drugs, develop genomic diagnostics, and advance the quest for personalized medicine. Golden Helix products and services have been cited in over 900 peer-reviewed publications.