Pediatrician Navigates Rare Genetic Diagnosis Using Peer-to-Peer Genetics Support Service

Unusual Clinical Observation Prompts Referral

With both a 13-year-old girl and her mother having an unusual ophthalmologic finding associated with a rare inherited genetic disease, the girl's pediatrician wanted to investigate further.

Genetic Testing and Counseling Confirm a Rare Disease

The patient was referred to Genome Medical for evaluation, genetic counseling, education and testing which confirmed a diagnosis of Fabry Disease.

A Complex Disease

Fabry disease affects multiple organ systems and can be life-threatening. It is difficult to diagnose and can often take more than 10 years after the first symptoms appear to receive a proper diagnosis. Early treatment is essential to manage symptoms and to limit irreversible, life-long damage.

Geneticist Expertise Needed to Develop Care Plan

Having no experience with Fabry disease, the Pediatrician turned to Genome Medical's Peer-to-Peer Genetics Support service. Within a few days, the Pediatrician met with a geneticist to discuss the case and determine next steps for the patient's care.

After discussing Fabry symptoms and therapy options, together they mapped out the evaluations and testing that would be needed and developed a schedule for ongoing monitoring.

"Time is an issue with this disease. The longer it's left untreated, the more damage will be done, which eventually leads to neuropathy that can be extremely painful, cardiovascular disease and kidney failure. There are, however, condition-specific treatment options, including enzyme replacement therapy, which will make a difference if Fabry disease is diagnosed and treated early."

GENETICIST, GENOME MEDICAL

Early Treatment for Improved Quality of Life

With guidance from Genome Medical, the pediatrician assembled a team of specialists including nephrology, cardiology, ophthalmology, and neurology and immediately began evaluations to ensure the patient received the best care to manage her disease.

Within just months, the girl had a diagnosis, a path forward and a full care team ready to support her treatment.

Medical Geneticist Peer-to-Peer Genetics Support Service

During a 30-60 minute phone call with one of our Medical Geneticists, any provider managing a patient's care can access our genetics expertise to help make appropriate and timely genetic testing decisions, confidently interpret genetic test results and determine the next steps for their patients.

Family Variant Testing Reveals Diagnosis for Mother As Well

After the girl's genetic diagnosis was confirmed by molecular analysis, it was recommended that her mother pursue genetic counseling and genetic testing for herself. Genome Medical facilitated family variant testing which confirmed a diagnosis of Fabry Disease for her as well.

Thanks to the pediatrician and rapid genetic services through Genome Medical, both the mother and her daughter can receive optimal care from their local providers to manage their disease.

Learn more about Genome Medical Peer-to-Peer Genetics Support.
Visit GenomeMedical.com/services/peer-to-peer/.