Boston Children's Hospital to tap IBM Watson to tackle rare pediatric diseases

November 11 2015, by Christine Douglass

Dr. Christopher Walsh, Chief of Genetics at Boston Children's Hospital explores the IBM Watson for Genomics Dashboard. IBM and Boston Children's Hospital today announced a new collaboration that aims to help clinicians identify potential diagnosis and treatment options for children with rare diseases using genomic data and cognitive technology. Credit: Boston Children's Hospital

In an initial project focused on kidney disease, Watson will analyze the massive volumes of scientific literature and clinical databases on the Watson Health Cloud to match genetic mutations to diseases and help uncover insights that could help clinicians identify treatment options.

Today about 1 in 10 Americans suffer from a rare disease, and half of these patients are children, according to the Global Genes Project. There are some 7,000 known rare disorders ranging from benign abnormalities to life-threatening disease. Many affected children pass away before a diagnosis can be made, leaving families to grieve without knowing what might have been done to help them, or how to avoid additional brothers or sisters being affected.

At Boston Children's, investigators at The Manton Center for Orphan Disease Research are focused on diagnosing children with a wide variety of rare genetic conditions. While strides have been made, the interpretation of sequencing results can be a labor-intensive process, presenting an overload of information whose analysis may not always yield a definitive causative variant.

In the new collaboration, Watson will be trained in nephrology by reading related medical literature and aggregating information on causative mutations for steroid-resistant nephrotic syndrome (SRNS), a rare genetic form of kidney disease. Then, experts at Boston Children's Hospital intend to feed genomic sequencing data from retrospective patients into Watson to further train the system. The goal is to create a cognitive system that can help clinicians interpret a child's genome sequencing data, compare this with medical literature and quickly identify anomalies that may be responsible for the unexplained symptoms.

"Coping with an undiagnosed illness is a tremendous challenge for many of the children and families we see," said Christopher Walsh, MD, PhD,

director of the Division of Genetics and Genomics at Boston Children's Hospital. "Watson can help us ensure we've left no stone unturned in our search to diagnose and cure these rare diseases so we can uncover all relevant insights from the patient's clinical history, DNA data, supporting evidence and population health data."

Even with a diagnosis, effective treatment for rare conditions can be elusive. For example, SRNS are usually unresponsive to immunosuppressive therapy, and often must go on chronic dialysis or wait for a kidney transplant—only to have their disease frequently recur in the new organ.

"One of Watson's talents is quickly finding hidden insights and connecting patterns in massive volumes of data," said Deborah DiSanzo, general manager, IBM Watson Health. "Rare disease diagnosis is a fitting application for cognitive technology that can assimilate different types and sources of data to help doctors solve medical mysteries. For the kids and their families suffering without a diagnosis, our goal is to team with the world's leading experts to create a cognitive tool that will make it easier for doctors to find the needle in the haystack, uncovering all relevant medical advances to support effective care for the child."

The kidney project will be done in collaboration with Friedhelm Hildebrandt, MD, chief of the Division of Nephrology at Boston Children's and Claritas Genomics. Following its successful completion, Boston Children's plans to expand the effort into undiagnosed neurologic disorders and other disease areas studied by The Manton Center, improving diagnostic and treatment services for patients nationwide.

Boston Children's Hospital is part of the Undiagnosed Diseases Network, a NIH program that aims to solve medical mysteries by integrating genetics, genomics, and rare disease expertise. Boston Children's was also the incubator behind Claritas Genomics, a genetic diagnostic

laboratory that offers genetic testing and develops new diagnostic tests and solutions, and organizer of the CLARITY Undiagnosed Challenge, a crowd sourcing competition seeking best practices in clinical genomics. The results and winner of the Challenge will be announced at the Boston Children's Hospital Global Pediatric Innovation Summit on November 10.

IBM has been developing Watson's ability to analyze genomic data in collaboration with leading cancer centers around the world. The system is currently being used at 16 cancer institutes to analyze and translate genomic data to help oncologists uncover personalized treatment options. The new project with Boston Children's represents the first time this technology will be applied to help clinicians efficiently identify possible options for rare disease diagnosis and treatment.

IBM and Boston Children's are also working together to build OPENPediatrics, an online platform designed to bring life-saving medical knowledge to pediatric caregivers worldwide. In September, the two organizations announced they will integrate Watson's deep and iterative question and answer capability to enhance and scale the OPENPediatrics initiative.

Provided by IBM