New study validates usefulness of genomic medicine in children with neurologic disorders

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Results from more than 100 families with children affected by a broad range of neurologic and developmental disorders who underwent genomic testing to end their quest for a diagnosis, were published today in *Science Translational Medicine*. This is the first study to show that a genome-based diagnostic approach directly impacts patient care of both infants and older children with neurologic disorders. Forty-five percent of families received a diagnosis by exome or genome sequencing, fifty percent of those diagnosed had a change in clinical impression or management and, in older children, genome-based diagnosis could have cut the wait for a diagnosis by more than six years. The study was led by Drs. Sarah Soden and Stephen Kingsmore of the Center for Pediatric Genomic Medicine at Children's Mercy Kansas City.

"We are striving to realize the benefits of genomic medicine in clinical care," said Dr. Soden. "These are exciting results that validate the promise of these technologies in children with a broad range of neurologic and developmental disorders. We've shown that genomic sequencing can end the diagnostic odyssey and have an immediate impact on patient care. Now we want to determine what kind of long-term impact that can have on a child's health and development; and how it impacts the family."

Eighty-five of the families studied had been seeking a diagnosis for their children for an average of six and a half years. They had spent an average $19,100 on prior tests that did not give a diagnosis. After exome or whole genome sequencing, forty percent of these children had a definitive diagnosis. Fifteen families in the study had infants with an acute medical condition at birth likely caused by a genetic disease; testing by rapid whole genome sequencing (STAT-Seq) enabled a diagnosis for seventy-three percent.

STAT-Seq test was one of TIME magazine's Top 10 Medical Breakthroughs of 2012 and is being developed in collaboration with Illumina, Inc. Still under research protocol, STAT-Seq is the fastest whole-genome test in the world, taking less than 50 hours from test order to delivery of an initial report.

In this study, half of the diagnoses affected the physician's impression of the patient's disorder or the clinical treatment plan. Physicians started new medications, discontinued treatments or supplements, and tested patients for complications associated with the genetic finding. Sequencing technologies not only identified known genetic diseases, but also new genetic diseases and previously unreported manifestations of known genetic disorders.

More information: "Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders," *Science Translational Medicine*, stm.sciencemag.org/lookup/doi/ … scitranslmed.3010076

Provided by Children's Mercy Hospital

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