

Clinical utility of rapid whole genome sequencing in neonates with seizures

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A new study aims to determine the underlying etiology of seizures and help to target therapy, improve control of seizures, and potentially reduce morbidities in children. Findings from the study will be presented during the Pediatric Academic Societies (PAS) 2019 Meeting, taking place on April 24—May 1 in Baltimore.

"As a neonatologist working with the team at the Genomics Institute, I've seen first-hand that rapid whole genome sequencing (rWGS) can be effective in identifying etiology of unexplained seizures in neonates and subsequently optimizing their care," said Jeanne Carroll, MD, one of the authors of the study. "Early rWGS can give answers to distressed families, help physicians provide a prognosis, and most importantly may help guide therapy with the potential to impact outcomes. A [retrospective analysis](#) of 19 patients who were admitted to the [neonatal intensive care unit](#) (NICU), with unclear etiology of seizures received WGS resulting in a [molecular diagnosis](#) for six infants. Of those six patients, four received a change in [medical management](#) as a result of the genetic diagnosis."

This study retrospectively identified a cohort of patients admitted in the first 30 days of life with presenting symptom of seizures who also underwent rapid whole genome sequencing during the admission. These cases were reviewed to assess for etiology of seizure, results of rWGS, and changes in management based on rWGS results.

Nineteen patients were identified with average age at admission of four

days and average hospital day at which sequencing was sent of 3.3. There were six diagnoses made by rWGS (31.6%). Four patients were later found on neuroimaging to have a stroke and three had changes of hypoxic ischemic encephalopathy (HIE) on MRI. Four of the six diagnoses led to a change in management including three with targeted [seizure](#) medications, and one with a referral to neurometabolic specialist and addition of dietary supplements. Two [patients](#) were found to have KCNQ2 mutations, both had significant side effects from antiepileptic medications. In each case, the medication regimen was optimized based on genetic findings leading to control of seizures and reduction in side effects from non-targeted therapies. In this cohort there was also a pyridoxine dependent epilepsy, two syndromic causes of seizures and one metabolic condition identified.

The study concluded that rWGS can identify etiology and direct therapy in the neonate with unexplained seizures.

More information: Dr. Carroll will present findings from "Clinical Utility of rWGS in the Evaluation of Neonatal Seizures" on Saturday, April 27 at 9:15 a.m. EDT.

Provided by American Pediatric Society

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