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Priya Monrad
Pediatric Epileptologist, USA

Whole genome/exome sequencing in pediatric epilepsy: Are there real clinical benefits?

Whole Exome Sequencing (WES) is becoming increasingly recognized as a valuable diagnostic tool in pediatric neurological disorders and pediatric epilepsy, but insurance or access to specialist testing remain major barriers in its widespread implementation. Currently it is primarily performed after standard workups have failed to provide a unifying diagnosis. Studies have demonstrated the cost-effectiveness and sensitivity of WES as a diagnostic modality, but a major remaining question for clinicians is whether WES results affect daily clinical decision-making about patient care in pediatric epilepsy. We will review the existing evidence for the use of WES in pediatric epilepsy, discuss case studies where WES is used in pediatric epilepsy patients in our personal practice and will discuss whether WES could become a useful clinical tool for changing management of pediatric epilepsy patients.

Biography

Priya Monrad is a practicing Pediatric Epileptologist and the program director for the Child and Adolescent Neurology program in Milwaukee, Wisconsin, USA. Her child neurology training was completed at Mayo Clinic, Rochester, MN and her clinical interests are prenatal/neonatal neurology and genetic/metabolic disorders.

pmonrad@mcw.edu

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