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Utilization Of NGS data in Neurogenetic Disorders

Neurogenetic disorders are a frequent reason for medical consultation in pediatric neurology service. Clinical variability and genetic heterogeneity are the whole mark of these diseases. Despite extensive metabolic and radiologic workup, the diagnostic yield has been disappointingly low. Whole exome sequencing (WES) has substantially accelerated the pace of discovery of disease associated genes and ending the diagnostic odyssey for many disorders. The diagnostic yield of WES is approximately around 30% in neurogenetic disorders. The diagnostic yield of 220 patients with various neurogenetic disease from highly consanguineous families evaluated in our center revealed a 25% pathogenic variants and 29% variant of unknown significance. The analysis of WES remains challenging. Determining whether this variant is disease causing or not is not an easy task and it requires cumulative level of evidence to ascertain pathogenicity. The ACMG/AMP guidelines attempts to ensure evidence-based interpretation of variants by considering multiple categories of data including allele frequency, computational prediction tools, functional studies that include cell lines or animal models, segregation studies and gene specific information (Hoskinson, Dubuc et al. 2017).

Here we will shed some light on the challenges of variant interpretation with some clinical cases illustration, we will discuss the limitation of NGS testing as a stand alone test, the clinician role in the rapidly expanding genomic medicine, the importance of deep clinical phenotyping and we will share our experience in WES in our neurogenetic clinic

Speaker Biography

Aqeela Al-Hashim,obtained her Bachelor of Medicine and Surgery (MBBS) from King Faisal University in 2002, then she obtained her Saudi Board and Arab Board of Pediatrics last 2008 after finishing her pediatric residency program in Riyadh, Saudi Arabia. She obtained her certificate of Royal College of Physician and Surgeon of Canada in child neurology (FRCPC) last 2015. She did Neurogenetics fellowship in clinical and molecular genetics research at the Hospital for Sick Children , Toronto, Canada. She is currently working as a consultant in pediatric neurology / neurogenetics at King Fahad medical city in Riyadh , Saudi Arabia. She was awarded the very prestigious John Prichard Prize for her research in Toronto. This prize resulted from her work in Corpus callosum abnormalities. Her main research interest is translational research mainly in genomic medicine, with utilization of functional molecular genetic studies to validate novel genetic variations in neurogenetic disorders.

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