The Yield of Genetic and Metabolic Testing in Epileptic Spasms



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INTRODUCTION

Epileptic spasm (ES) are a challenging type with huge of seizures implications. Identifying the etiology in a cost-effective manner helps in management, utilization of personalized therapeutics and prognostication

OBJECTIVES

genetic and To evaluate the yield of metabolic testing in patients with ES.

MATERIALS AND METHODS

This is a retrospective review the patients with ES who were treated at King Fahad Dammam, Specialist Hospital Saudi 2009 2020. Arabia, between and Pediatric patients less than 16-year-old presenting with ES and diagnosed by video-EEG were included. Our centers approach in ES workup was to start with clinical assessment and neuroimaging. If the etiology was not identified, we ordered metabolic and/or genetic work up.



RESULTS

We identified 126 patients with ES during the study period. Among these patients,

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Genetic Test	Contributed	Did not contribute	Marginal Row Totals	d
WES	15 (11.41) [1.13]	12 (15.59) [0.83]	27	<u>REF</u> 1-
Epilepsy panel	15 (18.59) [0.69]	29 (25.41) [0.51]	44	- W
Marginal Column Totals	30	41	71 (Grand Total)	n





an etiology was identified in 58 patients by clinical and imaging studies, while the remaining (n=68, 53.9%) patients required further workup. Metabolic testing helped identify the etiology in 8.3% of patients tested, while genetic testing identified the etiology in 46% of patients tested, with varying yield of each test. WES provided the highest testing yield, as 55.6% of WES tests provided a final diagnosis compared with comprehensive epilepsy panel (34%), and targeted gene testing (5%).

CONCLUSIONS

yield in Metabolic workup has a low identifying ES etiology. Genetic workup, on the other hand, had a high yield in identifying the etiology of ES when initial clinical assessment and neuroimaging were non-contributory. WES, in our cohort, was the most cost-effective genetic test in letermining the etiology of ES.

FERENCES

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