REVIEW ARTICLES

DOI: 10.5281/zenodo.4018958
UDC: 616.853:575.113

Open Access

Applicability of next generation genetic testing in epilepsy through whole exome sequencing

*[1,3]Daniela Catereniuc, [1,2]Viorica Chelban, [3]Stanislav Groppa

[1]Laboratory of Neurobiology and Medical Genetics, [3]Department of Neurology No 2
Nicolae Testemitanu State University of Medicine and Pharmacy, Chisinau, the Republic of Moldova
[2]Department of Neuromuscular Diseases, Queen Square Institute of Neurology, University College London, Great Britain

Authors' ORCID iDs, academic degrees and contributions are available at the end of the article

*Corresponding author: daniela.catereniuc@usmf.md
Manuscript received August 10, 2020; revised manuscript September 14, 2020; published online October 02, 2020

Abstract

Background: Epilepsy affects around 1% of the general population. With already acknowledged strong genetic contributions, >50% of epilepsy cases still remain undiagnosed. This is primordially due to the multifactorial condition of epilepsy that makes it a challenge to select the optimal genetic test for each specific case. Recently, next-generation sequencing (NGS) led to massive gene discovery, including epilepsy that also imposed serious financial burdens on healthcare systems. This study review highlights the progress in the field of epilepsy genetics and argues on how the genetic architecture of common epilepsies is progressively being unraveled. Since the 1995 finding of *CHRNA4* mutation, more than 500 genes were estimated to play a significant role in epilepsy. To date, the majority of diagnostic genetic testing is conducted in the pediatric population, while the utility of such testing is less well understood in adults with epilepsy. A broad range in the diagnostic rate of NGS, especially of the Whole Exome Sequencing (WES), in epilepsy has been described. However, NGS introduces new challenges, yet to be resolved.
Conclusions: Epilepsy's genetic background is nowadays undeniable; however, the complexity of this condition makes it difficult to be solved. WES has increasingly been used to uncover the role of the coding genetic material in the human genome and is nowadays considered one of the most cost-effective genetic tests for epilepsy, being a prerequisite for personalized treatment approaches and for reducing the epilepsy patient's "diagnostic odyssey".
Key words: epilepsy genetics, next-generation sequencing, whole exome sequencing.

Cite this article
Catereniuc D, Chelban V, Groppa S. Applicability of next generation genetic testing in epilepsy through whole exome sequencing. *Mold Med J.* 2020;63(5):54-61. doi: 10.5281/zenodo.4018958.