Genetics of Epilepsy – Literature Review

Laura Micevičiūtė[1], Rūta Mameniškienė[2]

[1] Faculty of Medicine, Vilnius University, Vilnius, Lithuania
[2] Clinic of Neurology and Neurosurgery, Institute of Clinical Medicine, Faculty of Medicine, Vilnius University, Vilnius, Lithuania

Introduction. Epilepsy is a common neurological disorder with significant etiological heterogeneity. Genetic factors play a major role, particularly in early-onset epilepsies, developmental and epileptic encephalopathies, and drug-resistant cases. Advances in next-generation sequencing have improved the identification of genetic causes and understanding of molecular mechanisms. This study aimed to review the spectrum of genetic epilepsies, assess genetic testing methods and their diagnostic yield, and evaluate the clinical significance of genetic diagnosis, including perspectives of precision medicine and gene therapy.

Methods. A narrative literature review of peer-reviewed publications on genetic epilepsies was conducted, focusing on genetic mechanisms, diagnostic methods, and clinical relevance.

Results. Genetic etiology was addressed in 40 studies, mainly focusing on specific genes and associated phenotypes. The diagnostic value of genetic testing was evaluated in 6 publications, particularly regarding next-generation sequencing methods. Treatment aspects were discussed in 4 studies, with 2 highlighting the impact of genetic findings on treatment decisions. Approximately 5 studies emphasized the role of genetic diagnosis in prognosis and genetic counseling. Overall, genetic etiology was the predominant focus, while treatment-related aspects were less frequently addressed.

Conclusions. Genetic epilepsies constitute a significant proportion of cases and should be considered in selected patients. Modern genomic technologies improve diagnostic accuracy and support personalized treatment strategies, potentially leading to better clinical outcomes.