

HLAVNÍ TÉMA

GENETIKA EPILEPSIÍ A SÚČASNÉ MOŽNOSTI ICH GENETICKEJ DIAGNOSTIKY

11. Habela CW, Schatz K, Kelley SA. Genetic Testing in Epilepsy: Improving Outcomes and Informing Gaps in Research. *Epilepsy currents/American Epilepsy Society* [Preprint]. 2024. <https://doi.org/10.1177/15357597241232881>.
12. Hebbar M, Mefford HC. Recent advances in epilepsy genomics and genetic testing. *F1000Research*. 9. 2020. <https://doi.org/10.12688/f1000research.21366.1>.
13. Helbig KL, Farwell Hagman KD, Shinde DN, et al. <http://paperpile.com/b/EgdPWZ/HepLo> Diagnostic exome sequencing provides a molecular diagnosis for a significant proportion of patients with epilepsy. *Genetics in medicine: official journal of the American College of Medical Genetics*. 2016;18(9):898-905.
14. International League Against Epilepsy Consortium on Complex Epilepsies. Genome-wide mega-analysis identifies 16 loci and highlights diverse biological mechanisms in the common epilepsies. *Nature communications*. 2018;9(1):5269.
15. Johannesen KM, Tumer Z, Weckhuysen S, et al. Solving the unsolved genetic epilepsies: Current and future perspectives. *Epilepsia*. 2023;64(12):3143-3154.
16. Krey I, Platzer K, Esterhuizen A, et al. Current practice in diagnostic genetic testing of the epilepsies. *Epileptic disorders: international epilepsy journal with videotape*. 2022;24(5):765-786.
17. Kurosawa R, Lida K, Ajiro M, et al. PDIVAS: Pathogenicity predictor for Deep-Intronic Variants causing Aberrant Splicing. *BMC genomics*. 2023;24(1):601.
18. Leu C, Stevelink R, Smith AW, et al. Polygenic burden in focal and generalized epilepsies. *Brain: a journal of neurology*. 2019;142(11):3473-3481.
19. Leu C, Richardson TG, Kaufmann T, et al. Pleiotropy of polygenic factors associated with focal and generalized epilepsy in the general population. *PLoS one*. 2020;15(4):e0232292.
20. Maillard PY, Baer S, Schaefer E, et al. Molecular and clinical descriptions of patients with GABA receptor gene variants (GABRA1, GABRB2, GABRB3, GABRG2): A cohort study, review of literature, and genotype-phenotype correlation. *Epilepsia*. 2022;63(10):2519-2533.
21. Mefford HC. Clinical Genetic Testing in Epilepsy. *Epilepsy currents / American Epilepsy Society*. 2015;15(4):197-201.
22. Mousavi N, Shleizer-Burko S, Yanicky R, et al. Profiling the genome-wide landscape of tandem repeat expansions. *Nucleic acids research*. 2019;47(15):e90.
23. Oliver KL, Scheffer IE, Bennett MF, et al. Genes4Epilepsy: An epilepsy gene resource. *Epilepsia*. 2023; 64(5):1368-1375.
24. Oyler J, Maljevic S, Scheffer IE, et al. Ion Channels in Genetic Epilepsy: From Genes and Mechanisms to Disease-Targeted Therapies. *Pharmacological reviews*. 2018;70(1):142-173.
25. Perucca P, Bahlo M, Berkovic SF. The Genetics of Epilepsy. *Annual review of genomics and human genetics*. 2020;21:205-230.
26. Rastin C, Schenkel LC, Sadikovic B. Complexity in Genetic Epilepsies: A Comprehensive Review. *International journal of molecular sciences*. 2023;24(19):14606.
27. Scheffer IE, Berkovic S, Capovilla G, et al. ILAE classification of the epilepsies: Position paper of the ILAE Commission for Classification and Terminology. *Epilepsia*. 2017;58(4):512-521.
28. Symonds JD, Zuberi SM, Johnson MR. Advances in epilepsy gene discovery and implications for epilepsy diagnosis and treatment. *Current opinion in neurology*. 2017;30(2):93-199.
29. Van Loo KMJ, Carvill GL, Beckej AJ, et al. Epigenetic genes and epilepsy – emerging mechanisms and clinical applications. *Nature reviews. Neurology*. 2022;18(9):530-543.
30. Ye Z, Chatterton Z, Pflueger J, et al. Cerebrospinal fluid liquid biopsy for detecting somatic mosaicism in brain. *Brain communications*. 2021;3(1):fcaa235.
31. Zhou J, Troyanskaya OG. Predicting effects of noncoding variants with deep learning-based sequence model. *Nature methods*. 2015;12(10):931-934.

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