



Genetic susceptibility in Juvenile Myoclonic Epilepsy: Systematic review of genetic association studies

Daniel Leite Góes Gitai

Department of Cellular and Molecular Biology, Institute of Biological Sciences and Health, Federal University of Alagoas, Maceio, Alagoas, Brazil

Abstract:

Background: Several genetic association investigations have been performed over the last three decades to identify variants underlying Juvenile Myoclonic Epilepsy (JME). Here, we evaluate the accumulating findings and provide an updated perspective of these studies.

Methodology: A systematic literature search was conducted using the PubMed, Embase, Scopus, Lilacs, epiGAD, Google Scholar and Sigle up to February 12, 2016. The quality of the included studies was assessed by a score and classified as low and high quality. Beyond outcome measures, information was extracted on the setting for each study, characteristics of population samples and polymorphisms.

Results: Fifty studies met eligibility criteria and were used for data extraction. With a single exception, all studies used a candidate gene approach, providing data on 229 polymorphisms in or near 55 different genes. Of variants investigating in independent data sets, only rs2029461 SNP in GRM4, rs3743123 in CX36 and rs3918149 in BRD2 showed a significant association with JME in at least two different background populations. The lack of consistent associations might be due to variations in experimental design and/or limitations of the approach.

Conclusions: Thus, despite intense research evidence established, specific genetic variants in JME susceptibility remain inconclusive. We discussed several issues that may compromise the quality of the results, including methodological bias, endophenotype and potential involvement of epigenetic factors.



Biography:

Daniel Leite Góes Gitai, Brazilian biology professor, researcher. Certified biologist Federal University Alagoas, Maceió, Brazil.

Publication of speakers:

1. Genton P, Gelisse P. The history of juvenile myoclonic epilepsy. *Epilepsy Behav.* 2013;28 Suppl 1: S2–7. pmid:23756475.
2. Proposal for revised classification of epilepsies and epileptic syndromes. Commission on Classification and Terminology of the International League Against Epilepsy. *Epilepsia.* 30: 389–99. Available: <http://www.ncbi.nlm.nih.gov/pubmed/2502382> pmid:2502382.
3. Gitai, Daniel & Santos, Ygor & Upadhy, Raghavendra & Maheedhar, Kodali & LN, Madhu & Shetty, Ashok. (2019). Extracellular Vesicles in the Forebrain Display Reduced miR-346 and miR-331-3p in a Rat Model of Chronic Temporal Lobe Epilepsy. *Molecular Neurobiology.* 57. 10.1007/s12035-019-01797-1.
4. Upadhy, Raghavendra & Maheedhar, Kodali & Gitai, Daniel & Castro, Olagide & Hattiangady, Bharathi & Dinesh, Upadhy. (2019). A Model of Chronic Temporal Lobe Epilepsy Presenting Constantly Rhythmic and Robust Spontaneous Seizures, Co-morbidities and Hippocampal Neuropathology. *Aging and Disease.* 10. 10.14336/AD.2019.0720.

Webinar on Gene Therapy | October 6th, 2020 | London, UK

Citation: Daniel Leite Góes Gitai, Genetic susceptibility in Juvenile Myoclonic Epilepsy: Systematic review of genetic association studies; *Gene Therapy* 2020; October 6th, 2020; London, UK