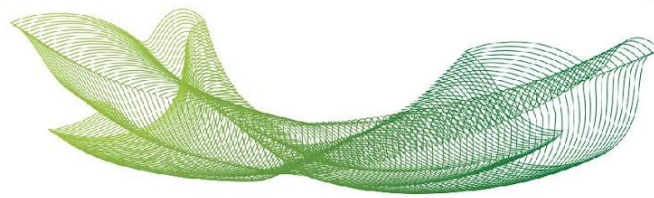


Tipo	Periódico
Título	Single-Nucleotide Variants in microRNAs Sequences or in their Target Genes Might Influence the Risk of Epilepsy: A Review
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Resumo	Single-nucleotide variant (SNV) is a single base mutation at a specific location in the genome and may play an import role in epilepsy pathophysiology. The aim of this study was to review case–control studies that have investigated the relationship between SNVs within microRNAs (miRs) sequences or in their target genes and epilepsy susceptibility from January 1, 2010 to October 31, 2020. Nine case–control studies were included in the present review. The mainly observed SNVs associated with drug-resistant epilepsy (DRE) risk were SNVs n.60G > C (rs2910164) and n.-411A > G (rs57095329), both located at miR-146a mature sequence and promoter region, respectively. In addition, the CC haplotype (rs987195-rs969885) and the AA genotype at rs4817027 in the MIR155HG/miR-155 tagSNV were also genetic susceptibility markers for early-onset epilepsy. MiR-146a has been observed as upregulated in human astrocytes in epileptogenesis and it regulates inflammatory process through NF-κB signaling by targeting tumor necrosis factor-associated factor 6 (TRAF6) gene. The SNVs rs2910164 and rs57095329 may modify the expression level of mature miR-146a and the risk for epilepsy and SNVs located at rs987195-rs969885 haplotype and at rs4817027 in the MIR155HG/miR-155 tagSNV could interfere in the miR-155 expression modulating inflammatory pathway genes involved in the development of early-onset epilepsy. In addition,



SNVs rs662702, rs3208684, and rs35163679 at 3'untranslated region impairs the ability of miR-328, let-7b, and miR-200c binding affinity with paired box protein PAX-6 (PAX6), BCL2 like 1 (BCL2L1), and DNA methyltransferase 3 alpha (DNMT3A) target genes. The SNV rs57095329 might be correlated with DRE when a larger number of patients are evaluated. Thus, we concluded that the main drawback of most of studies is the small number of individuals enrolled, which lacks sample power.

Fomento