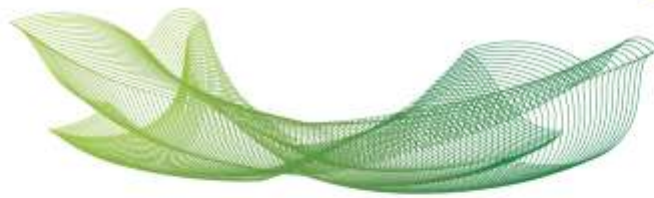




Tipo	Periódico
Título	Genetics in epilepsy, Dravet and SUDEP: A systematic review
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Resumo	<p>Introduction: Epilepsy is a neurologic condition and its patients have higher mortality rates than healthy individuals. One of the most frequent death causes in epilepsy is SUDEP. Moreover, another cause in epilepsy, although rare, is Dravet Syndrome and it's generally diagnosed in children in their first year of life. In around 70% of the patients there is a genetic inheritance involved and several genes have been described as related to SUDEP and Dravet Syndrome which the most prevalent were DNA and belonged to the voltage-gated channel coding.</p> <p>Objectives: Identify genes related do Dravet Syndrome, SUDEP and Epilepsy in literature, analyze the convergence of results, and discuss the differences that were found, contributing to the study of genetic mapping.</p> <p>Methodology: Meta-analysis realized through a bibliographical research in the electronic data banks such as PUBMED, NCBI, Scholar Google and Scielo with the descriptors Epilepsy; Epilepsies, Myoclonic; Polymorphism, Single Nucleotide; MicroRNAs; Death, Sudden. The languages used to filter the articles were English, Spanish and Portuguese. The year was a filtering factor, selecting only articles published in the last 20 years.</p> <p>Results: About forty (40) genes were found to be SUDEP related and the most relevant was KCNH2, appearing more frequently than other genes. Additionally, in Epilepsy without risk for SUDEP twenty-eight (28) DNA genes were found, which the most frequently mentioned was HCN2, appearing six (6) times. Among Dravet syndrome patients, only two (2) genes were described: SCN1A and SCN9A, both of them were SUDEP</p>



risk related. In epilepsy nineteen (19) microRNA genes were found in five (5) different articles.

Project: It was identified genes related to Dravet Syndrome, SUDEP and Epilepsy in literature and was noted that some genes appeared more frequently than others. Among the DNA related genes, the most prevalent were around those belonging to the voltage-gated channel coding, being them sodium, hydrogen and potassium. The results demonstrate that 25% of all articles mentioned SCN1A; 17,5% mentioned HCN2 and 15% mentioned KCNH2. Together these findings provide a basis for further investigation around the role of ionic channels in SUDEP and DS. On the other hand, regarding to MicroRNA linked genes data were scarce. Only five articles described changes on SUDEP and neither described changes compatible with DS. There was no most prevalent mRNA gene.

Conclusion: A range of studies associated DNA mutations with Epilepsy, DS and SUDEP. However, MicroRNA research could not provide concrete evidence on its influence in the development of the epilepsy and epilepsy related conditions. The identification of possible etiological factors can allow a correct and early diagnosis, providing better prognosis and therapeutic outcome, besides it can evaluate the recurrence of the disease in the family. Still, further prospective studies in larger cohorts are required to further define the genetic predisposition to SUDEP, DS and Epilepsy.

Fomento