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Writing assignment #4

The article that is being used would be considered a primary article, this is because it follows three of the major characteristics of a primary article. To be a primary article an article has to be published in a peer-reviewed journal, discuss a research topic, and contain a section called “methods” and “results” where the performed experiments are discussed. This article was published in journal *Nature*, which indicates that the article has been peer-reviewed. The article also has a consistent topic that the authors go on to discuss. Lastly, there are sections containing the original methods and the results from the experiment that the authors performed. This article touches on the topic of genetic variations, in particular a variant of NPRL2.

In this article the authors describe an individual with multiple seizure types that are harboring a splicing variation in the NPRL2 (nitrogen permease regulator like 2). They were able to identify a variant of NPRL2 in a family with familial focal epilepsy with a variable foci, which they were able to confirm through the use of minigene assays. They confirmed that the NPRL2 variant leads to exon 3 skipping and an increase in the frequency of seizures in the left frontal and central regions of the brain. The experiment that the authors performed provided them with evidence for pathogenicity of the splicing variation in the GATOR1 complex and expanded the phenotype and genotype spectrum of FFEVF.

Zhang, J. et al. A splicing variation in *NPRL2* causing familial focal epilepsy with variable foci: additional cases and literature review. *J Hum Genet* **67**, 79-85 (2022).