Review of: "Identification of Novel ADGRV1 and KCNC2 Variants Using Whole-Exome Sequencing in Two Colombian Patients with Usher and Encephalopathy Syndromes"

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Potential competing interests: The author(s) declared that no potential competing interests exist.

This is a segregation analysis in a Colombian family involving 2 rare missense variants in ADGRV1gene showing compound heterozygous inheritance associated with Usher syndrome type 2C in the mother and a second heterozygous de novo variant in KCNC2 gene in a child with neurodevelopment disorder with epilepsy.

The variants should be assessed according to CADD scores and ACMG guidelines and this reported in the results.

A limitation of the study is that there is no information about structural variants, including copy number variants which are very relevant in the diagnosis of neurodevelopment disorders.