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REVIEWER'S REPORT

Manuscript No.: IJAR-59640

Title: Dravet Syndrome: An Atypical Case Revealed by Whole Exome Sequencing.

Recommendation:

Accept as it is .....
Accept after minor revision.....
✓ Accept after major revision .....
Do not accept (*Reasons below*) .....

Rating	Excel.	Good	Fair	Poor
Originality		✓		
Techn. Quality		✓		
Clarity		✓		
Significance	✓			

Reviewer Name: Dr S. K. Nath

Detailed Reviewer's Report

Strength of the study:

- Presents an uncommon clinical presentation of Dravet syndrome.
- Describes the patient's seizure history and family history.
- Includes EEG and brain MRI findings.
- Uses whole exome sequencing to identify an SCN1A variant.
- Reports the specific heterozygous variant c.2839G>A (p.Val947Met).
- Describes the patient's treatment and six-month clinical progress.
- Includes a family pedigree and EEG image.
- Highlights the value of genetic testing in atypical epilepsy.

Weakness of the study:

- The age and timing of initial seizure presentation need clarification.
- Detailed evidence supporting the pathogenic classification is not provided.
- Confirmation of the variant's inheritance is still pending.
- The family history requires better genetic correlation.
- Long-term neurological and developmental outcomes are unavailable.
- The rationale for the four-drug treatment combination needs clarification.
- Written consent and ethics approval or exemption are not clearly reported.
- Grammar, spacing and reference formatting require substantial correction.

Reviewers Comments:

The manuscript describes a clinically interesting case of Dravet syndrome identified through whole exome sequencing. The EEG findings, family pedigree and identification of the SCN1A variant provide useful diagnostic information. The six-month improvement following combination treatment is also worth reporting. However, the authors should clarify the timeline of seizure onset, as the abstract and case presentation do not clearly establish the age at initial presentation. The pathogenic classification of the variant should be supported with detailed ACMG evidence, and the pending parental testing should be updated if available. Longer follow-up is needed to assess cognitive and developmental outcomes. Written informed consent from the child's legal guardian and the applicable ethics approval or exemption should be clearly stated. The manuscript contains frequent spacing, grammatical and reference-formatting errors that require correction. Overall, the case is relevant, but the clinical and genetic details need strengthening before publication.

Previously Published anywhere/Plagiarism check:

A closely matching conference abstract with the same title, authors and SCN1A variant (c.2839G>A, p.Val947Met) appeared in the 2025 European Journal of Human Genetics conference abstracts. The reported patient also appears to be the same case. The authors must disclose this prior presentation and clarify what new information the full case report adds. This overlap requires editorial assessment and a formal similarity check.