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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		Good		
Techn. Quality	Excellent			
Clarity	Excellent			
Significance	Excellent			

Reviewer's ID: Dr. Sumathi

Detailed Reviewer's Report

1. **Dravet syndrome is a rare, severe, and medication-resistant form of developmental and epileptic encephalopathy (DEE) that typically begins during the first year of life.**
2. **Dravet syndrome (formerly known as Severe Myoclonic Epilepsy of Infancy) is a rare, severe, and lifelong genetic epilepsy classified as a developmental and epileptic encephalopathy (DEE). It typically manifests in the first year of life in otherwise typically developing infants, causing frequent, prolonged, and drug-resistant seizures that significantly alter cognitive and physical development.**
3. **Childhood epilepsy is a neurological condition defined by two or more recurring, unprovoked seizures caused by sudden bursts of abnormal electrical activity in the brain. It affects roughly 1% to 2% of children worldwide.**
4. **Childhood epilepsy is a neurological brain condition characterized by repeated, unprovoked seizures caused by sudden bursts of irregular electrical activity in the brain. It is the most common childhood brain disorder in the United States, affecting roughly 450,000 to 470,000 children and youth. A child is typically diagnosed**

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with epilepsy if they experience two or more unprovoked seizures occurring more than 24 hours apart.

5. **The SCN1A gene provides instructions for making the Nav1.1 protein, a voltage-gated sodium channel alpha subunit. These channels are found on the surface of brain cells and allow sodium ions to flow into neurons, which is essential for creating and passing electrical signals.**
6. **The SCN1A gene (Sodium voltage-gated channel alpha subunit 1) is a critical gene located on the long (q) arm of human Chromosome 2 (at position 2q24.3). Famously referred to in medical literature as a "super culprit gene," it is one of the most frequently mutated genes associated with human epilepsy.**
7. **Dravet syndrome is a rare, severe developmental and epileptic encephalopathy that typically begins in the first year of life in a previously normal infant.**
8. **While typical Dravet Syndrome (DS) is a severe, early-onset genetic encephalopathy primarily driven by de novo loss-of-function mutations in the SCN1A gene, clinical literature highlights several "atypical" or "borderland" presentations. These atypical cases challenge standard diagnostic pathways through variations in genetic causes, imaging results, and clinical progressions.**
9. **Whole exome sequencing (WES) plays a critical role in diagnosing atypical or non-classical presentations of Dravet syndrome and related developmental and epileptic encephalopathies (DEEs) when standard epilepsy panels fail or yield ambiguous results.**
10. **Whole exome sequencing (WES) has become a critical tool for revealing atypical presentations of Dravet syndrome by identifying novel variants, unexpected gene interactions, or entirely different genes that mimic its phenotype.**
11. **Key words can be included more words.**
12. **Pictures with significant points are awesome.**
13. **Result part can be made graphs and tables for values.**
14. **Summary points also be included.**
15. **References should be with alphabetical order.**
16. **After those changes good to publish in your journal.**