

REVIEWER'S REPORT

Manuscript No.: IJAR-55220

Title: A CASE OF REFRACTORY SEIZURES IN JUVENILE CANAVANS DISEASE

Recommendation:

Accept as it is

Accept after minor revision.....x.....

Accept after major revision

Do not accept (*Reasons below*)

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

Reviewer Name: Dr. Mohammad Nadeem Khan

Detailed Reviewer's Report

General Assessment

This manuscript presents a rare and clinically relevant case of juvenile-onset Canavan disease manifesting as refractory seizures in early adulthood. The topic is important due to the rarity of juvenile Canavan disease and the diagnostic challenges it poses in patients presenting primarily with seizures. The manuscript is concise, clinically focused, and contributes valuable insight to the existing literature.

Title and Abstract

- The title is clear, precise, and accurately reflects the content of the manuscript.
- It appropriately highlights the rarity and clinical focus of the case.
- Suggestion (minor): The inclusion of "case report" in the title may improve indexing and clarity for readers.

Introduction

- The introduction provides adequate background on Canavan disease, including genetics, pathophysiology, clinical forms, and diagnostic markers.
- The distinction between infantile and juvenile forms is clearly explained.
- The rationale for reporting this case is justified due to the uncommon presentation and age group.

Case Report / Clinical Description

- The case is well-documented, with a clear chronological progression of symptoms.
- Neurological findings are described in sufficient clinical detail.

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- MRI and MR spectroscopy findings are relevant and appropriately interpreted, particularly the elevated NAA peak.
- The association of consanguinity strengthens the genetic plausibility.
- Suggestion (minor): Adding results of genetic confirmation (if available) or clarifying whether genetic testing could not be performed would further strengthen the case.

Diagnostic Workup and Interpretation

- The diagnostic reasoning is logical and supported by clinical, radiological, and spectroscopic findings.
- The emphasis on refractory seizures as a presenting feature is clinically important.
- The manuscript effectively highlights the need to consider genetic leukodystrophies in atypical seizure disorders.

Discussion and Clinical Relevance

- Although brief, the discussion appropriately emphasizes the key learning point.
- The conclusion reinforces an important clinical message regarding early genetic evaluation.
- Suggestion (optional): A short comparison with previously reported juvenile Canavan disease cases may enhance academic depth.

Ethical Considerations

- The case report appears to comply with ethical standards.
- It is assumed that informed consent was obtained; explicit mention of patient consent is recommended for completeness.

Language, Structure, and Presentation

- The manuscript is clearly written, concise, and easy to follow.
- Minor grammatical polishing may be done during copyediting, but no major language revisions are required.
- Figures and imaging descriptions are appropriate and relevant.

Strengths

- Rare disease presentation in an uncommon age group
- Clear clinical narrative and diagnostic reasoning
- Strong educational value for clinicians and neurologists
- Emphasis on genetic evaluation in refractory seizures
- Lack of genetic confirmation details (if unavailable, this should be stated)
- Brief discussion section (acceptable for a case report)