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REVIEWER'S REPORT

Manuscript No.: IJAR-55179

Title: CACH syndrome: A case report

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 **Date:** 11.12.25

Detailed Reviewer's Report

Strengths of the Paper

- **Comprehensive Case Description**: The paper provides a detailed account of the clinical presentation, neurological examination, laboratory findings, and imaging results of the patient, which enhances understanding of the disease presentation.
- **Integration of Diagnostic and Genetic Data**: The inclusion of MRI findings and genetic confirmation through EIF2B mutation enhances the diagnostic rigor and demonstrates the importance of combining imaging with molecular diagnostics.
- **Clear Illustrations of Imaging**: The descriptions of MRI findings are well-articulated, and the inclusion of figure references aids in visual understanding of the characteristic white matter changes.
- **Contribution to Rare Disease Literature**: As CACH syndrome is rare, this case adds valuable information to the limited existing literature, especially in a specific geographical context.

Weaknesses of the Paper

- **Limited Discussion on Management and Follow-Up**: The paper briefly mentions corticosteroid therapy but lacks an in-depth discussion on long-term management, prognosis, or potential therapeutic strategies.
- **Insufficient Literature Context**: The literature review is somewhat limited; extending it to include recent studies or broader epidemiological data could strengthen the background context.
- **Lack of Standardized Assessment Tools**: The report does not specify the use of standardized scales or assessments to quantify neurological or cognitive deficits.
- **Inadequate Details on Genetic Testing Methods**: Specifics about the genetic testing process, such as the technique used or mutation type, are not provided.

Reviewer Comments

- **Ethical Clearance Status**: The manuscript does not mention whether ethical approval or informed consent was obtained for publication. Ethical approval is typically required for case reports involving patient data, and this should be clearly stated.
- **Methodology Issues**: As a case report, methodology primarily involves clinical evaluation and genetic testing. However, details about the genetic testing methodology, including laboratory procedures, should be included to enhance reproducibility.
- **Typographical Mistakes**: Several minor typographical errors are present, including missing spaces (e.g., "white matterlesionspredominantly") and inconsistent formatting.

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- **Grammar and English Language Quality**: Overall, the language is clear, but there are some grammatical issues, such as awkward phrasing and punctuation problems, that need correction.
- **Formatting Issues**: The manuscript would benefit from consistent formatting, especially regarding headings, subheadings, and figure references, to improve readability.
- **Clarity of Objectives, Results, and Conclusion**: The objectives are implied but not explicitly stated. The results are clearly described, but the conclusion could be more explicitly linked to the case findings and broader implications.
- **Adequacy of References**: The references cited seem appropriate and relevant. However, incorporating more recent studies may enhance the literature review.
- **Missing or Incomplete Information**: Additional details about the genetic mutation specifics, management plan, and follow-up outcomes would augment the case report's completeness. Also, mentioning whether the patient's parents received genetic counseling would be informative.