

Review Form 3

Journal Name:	Asian Journal of Pediatric Research
Manuscript Number:	Ms_AJPR_124214
Title of the Manuscript:	Neuronal ceroid lipofuscinosis a very early infantile form Infantile neuronal ceroid lipofuscinosis: about a new case
Type of the Article	Care report

General guidelines for the Peer Review process:

This journal's peer review policy states that **NO** manuscript should be rejected only on the basis of '**lack of Novelty**', provided the manuscript is scientifically robust and technically sound. To know the complete guidelines for the Peer Review process, reviewers are requested to visit this link:

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PART 1: Review Comments

Compulsory REVISION comments	Reviewer's comment	Author's Feedback <i>(Please correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)</i>
<p>Please write a few sentences regarding the importance of this manuscript for the scientific community. Why do you like (or dislike) this manuscript? A minimum of 3-4 sentences may be required for this part.</p>	<p>Reporting a rare genetic diseases will give clinical idea for treating physician.</p>	
<p>Is the title of the article suitable? (If not please suggest an alternative title)</p>	<p>The title clearly indicates the disease and case report nature. It is concise and appropriate for its clinical focus.</p> <p>keywords should be streamlined and linked to more searchable terms. "Santavuori-Haltia" should be consistently used as a key term, and "myoclonus" and "epilepsy" can be refined with better descriptors, like "myoclonic seizures in neurodegenerative disorders."</p>	
<p>Is the abstract of the article comprehensive? Do you suggest the addition (or deletion) of some points in this section? Please write your suggestions here.</p>	<p>The abstract provides a summary of the case, mentioning key clinical symptoms (seizures, nystagmus, microcephaly) and the diagnostic process (brain MRI, biopsy).</p> <p>The abstract could be more structured, including the "Introduction" and "Conclusion" separately. Additionally, "Santavuori-Haltia disease" should be defined at least briefly for clarity. The abstract doesn't explicitly mention interventions or follow-up, which is crucial in understanding patient care. It also lacks the final genetic diagnosis, which would strengthen the summary.</p>	
<p>Are subsections and structure of the manuscript appropriate?</p>	<p>Strength: The introduction effectively outlines neuronal ceroid lipofuscinoses (NCLs), focusing on the autosomal recessive inheritance and lysosomal storage mechanisms. It also introduces the clinical manifestations.</p> <p>The description of the child's early symptoms and medical history (consanguinity, normal birth, spasms) is well-documented. The physical examination details (microcephaly, cranial measurements, neurological findings) are comprehensive.</p> <p>The case effectively integrates EEG, MRI, metabolic assessments, and skin biopsy to confirm the diagnosis. The brain imaging and electrophysiological findings are consistent with the disease, providing strong diagnostic evidence.</p> <p>The patient was treated with anticonvulsants (Depakine, Rivotril), which is appropriate for seizure management in Santavuori-Haltia disease.</p> <p>The discussion provides a good summary of the disease progression and diagnostic methods. It contextualizes the findings within the broader knowledge of NCL and references important literature.</p> <p>Suggestions for improvement:</p> <p>The introduction could be expanded to highlight the importance of early diagnosis and typical treatment strategies. Additionally, current epidemiological data or recent advancements could improve the relevance of the introduction. A clearer distinction between Santavuori-Haltia disease and other forms of NCL would strengthen the context.</p> <p>Including a timeline of symptom onset would aid in understanding disease progression better. There is a lack of discussion on family medical history or other social factors that may influence outcomes. Furthermore, while consanguinity is mentioned, details about genetic counseling or family implications are missing.</p> <p>The report could have provided more detail on the molecular biology and genetic findings, even though the molecular testing is still ongoing. A detailed genetic analysis (if available) would enhance the depth</p>	

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	<p>of the diagnostic section, especially for an autosomal recessive condition.</p> <p>The intervention section lacks details about other supportive care measures (e.g., respiratory management, physical therapy, palliative care). Since the child is being fed by a gastric tube, nutritional management and gastrointestinal considerations should have been discussed. The choice of anticonvulsants could be expanded to include other options, as well as an explanation of their effectiveness or limitations in this disease.</p> <p>The discussion could be expanded by including a comparison with other reported cases, especially in terms of genetics, clinical variants, or outcomes. The role of novel treatments or gene therapy could be addressed. Furthermore, challenges in managing neurodegenerative diseases like this one should be emphasized more. Addressing the ethical implications of providing care for terminal neurodegenerative conditions in pediatric settings could also be insightful.</p> <p>The conclusion could benefit from clearer clinical takeaways or recommendations for future research. Including a brief note on the future management of such cases (palliative care, genetic counseling) would add value.</p>	
<p>Please write a few sentences regarding the scientific correctness of this manuscript. Why do you think that this manuscript is scientifically robust and technically sound? A minimum of 3-4 sentences may be required for this part.</p>	<p>While the case report provides substantial clinical and diagnostic information, it could be strengthened with a clearer structure, detailed genetic data, additional discussion on management, and inclusion of a patient perspective and informed consent.</p>	
<p>Are the references sufficient and recent? If you have suggestions of additional references, please mention them in the review form.</p>	<p>The references appear comprehensive and relevant.</p>	
<p>Minor REVISION comments</p> <p>Is the language/English quality of the article suitable for scholarly communications?</p>	<p>Need minor revision.</p>	
<p>Optional/General comments</p>		

PART 2:

	Reviewer's comment	Author's comment (if agreed with reviewer, correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)
<p>Are there ethical issues in this manuscript?</p>	<p><i>(If yes, Kindly please write down the ethical issues here in details)</i></p>	

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