

**Review Form 3**

Journal Name:	<b>Asian Journal of Pediatric Research</b>
Manuscript Number:	<b>Ms_AJPR_122370</b>
Title of the Manuscript:	<b>Autosomal recessive spastic paraplegia type 51 caused by homozygous mutation of the AP4E1 gene in a 17 years old boy</b>
Type of the Article	

### Review Form 3

#### **PART 1: Review Comments**

<b>Compulsory</b> REVISION comments	<b>Reviewer's comment</b>	<b>Author's Feedback</b> <i>(Please correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)</i>
<b>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.</b>		
<b>Is the title of the article suitable? (If not please suggest an alternative title)</b>		
<b>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.</b>		
<b>Are subsections and structure of the manuscript appropriate?</b>		
<b>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.</b>		
<b>Are the references sufficient and recent? If you have suggestions of additional references, please mention them in the review form.</b>		

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<p>Minor REVISION comments</p> <p><b>Is the language/English quality of the article suitable for scholarly communications?</b></p>		
<p>Optional/General comments</p>	<p>The text provides a detailed and informative case report on Autosomal Recessive Spastic Paraplegia Type 51 (SPG51). Overall, it's well-structured and covers key aspects of the disease, but there are some areas where improvements can be made to enhance clarity, coherence, and precision. Below are some suggestions for revision:</p> <p>### General Comments:</p> <ol style="list-style-type: none"><li><b>Consistency</b>: There are some inconsistencies in terminology, such as the use of "homozygous" and "compound heterozygous" mutation. Ensure that these terms are used correctly and consistently throughout the text.</li><li><b>Grammar and Syntax</b>: Some sentences are lengthy and complex, making them hard to follow. Breaking them into shorter sentences can improve readability.</li><li><b>Redundancy</b>: Certain points are repeated multiple times (e.g., the role of the AP4E1 gene), which can be streamlined for a more concise narrative.</li><li><b>Clarity</b>: Some statements could be clarified, especially those involving complex medical terminology or genetic mechanisms.</li><li><b>Formatting</b>: Ensure that section headings are consistently formatted (e.g., capitalize "Conclusion") and that references are uniformly cited according to a specific style guide.</li></ol> <p>### Detailed Suggestions:</p> <p>#### Title:</p> <ul style="list-style-type: none"><li><b>Current</b>: "Autosomal recessive spastic paraplegia type 51 caused by homozygous mutation of the AP4E1 gene in a 17 years old boy."</li><li><b>Suggested</b>: "Autosomal Recessive Spastic Paraplegia Type 51 Due to Homozygous Mutation in the AP4E1 Gene: A Case Report of a 17-Year-Old Boy."</li></ul> <p>#### Abstract:</p> <ul style="list-style-type: none"><li><b>Sentence Simplification</b>: Some sentences are too dense with information. Simplify them to improve understanding.</li><li><b>Redundancy</b>: The first two sentences both mention the mutation in the AP4E1 gene and chromosome 15q21. Consider combining them for clarity.</li><li><b>Example</b>:<ul style="list-style-type: none"><li><b>Current</b>: "Autosomal recessive spastic paraplegia-51 (SPG51) is caused by a homozygous mutation in the AP4E1, adaptor related protein complex 4 subunit epsilon 1 gene, on chromosome 15q21."</li><li><b>Suggested</b>: "Autosomal recessive spastic paraplegia-51 (SPG51) is a rare neurodevelopmental disorder caused by a homozygous mutation in the AP4E1 gene, located on chromosome 15q21."</li></ul></li></ul> <p>#### Introduction:</p> <ul style="list-style-type: none"><li><b>Content Order</b>: Consider starting with a brief definition of HSP before diving into its classification. This sets a clearer context for the reader.</li><li><b>Example</b>:<ul style="list-style-type: none"><li><b>Current</b>: "Hereditary spastic paraplegia (HSP) is a group of neurodegenerative diseases with genetic and clinical heterogeneity characterized by spasticity and weakness of the lower limbs."</li><li><b>Suggested</b>: "Hereditary spastic paraplegia (HSP) refers to a group of genetically and clinically heterogeneous neurodegenerative disorders characterized primarily by spasticity and weakness in the lower limbs."</li></ul></li></ul> <p>#### Case Report:</p> <ul style="list-style-type: none"><li><b>Grammar/Clarity</b>: The sentence "A compound heterozygous mutation in the AP4E1 gene</li></ul>	

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	<p>was found in genetic analysis" might contradict the title mentioning "homozygous mutation."          Clarify whether the mutation is homozygous or compound heterozygous.</p> <ul style="list-style-type: none"> <li>- <b>Medication List:</b> The list of previous medications can be confusing. Consider rephrasing for clarity.</li> <li>- <b>Example:</b> <ul style="list-style-type: none"> <li>- <b>Current:</b> "An anticonvulsant medication with Perampanel was initiated after past therapy with Sultiam, Lacosamide, Lamotrigine, Oxcarbazepine, Levetiracetam and Zonisamide."</li> <li>- <b>Suggested:</b> "Perampanel was initiated as an anticonvulsant therapy after previous treatments with Sultiam, Lacosamide, Lamotrigine, Oxcarbazepine, Levetiracetam, and Zonisamide."</li> </ul> </li> </ul> <p><b>Discussion:</b></p> <ul style="list-style-type: none"> <li>- <b>Clarity:</b> Some sections are dense and difficult to follow. For example, the description of the AP-4 complex's role in vesicular trafficking could be simplified.</li> <li>- <b>Example:</b> <ul style="list-style-type: none"> <li>- <b>Current:</b> "The AP-4 complex subunit <math>\epsilon</math>-1 (AP-4E1) plays a role in recognizing and binding sorting signals in cells."</li> <li>- <b>Suggested:</b> "The AP-4E1 subunit is involved in recognizing and binding sorting signals within cells, which is crucial for proper protein localization in neurons."</li> </ul> </li> </ul> <p><b>Conclusion:</b></p> <ul style="list-style-type: none"> <li>- <b>Structure:</b> Strengthen the conclusion by briefly summarizing the key findings of the case and their implications.</li> <li>- <b>Example:</b> <ul style="list-style-type: none"> <li>- <b>Current:</b> "In conclusion, spastic paraplegia type 51 in childhood is rarely found and diagnosed and often found with clinical inconsistencies, leading to overlook or misdiagnosis."</li> <li>- <b>Suggested:</b> "In conclusion, SPG51 is a rare and often underdiagnosed condition in childhood, frequently presenting with clinical inconsistencies that can lead to misdiagnosis. This case highlights the importance of genetic testing and careful clinical evaluation in suspected cases of HSP."</li> </ul> </li> </ul> <p><b>Additional Minor Corrections:</b></p> <ul style="list-style-type: none"> <li>- "17 years old" should be "17-year-old" in both the title and the text.</li> <li>- Avoid passive voice where possible for stronger statements (e.g., "SPG51 is caused by..." could be "A mutation in the AP4E1 gene causes SPG51...").</li> <li>- Clarify the ambiguous use of "on/off effect" in the Case Report section; it's unclear in this context.</li> </ul> <p><b>Conclusion:</b>          The case report is well-constructed but could benefit from tightening the language, removing redundancies, and ensuring clarity in the presentation of clinical and genetic details. Addressing these points will enhance the overall quality and readability of the manuscript.</p>	
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**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)
Are there ethical issues in this manuscript?	<i>(If yes, Kindly please write down the ethical issues here in details)</i>	

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