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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 83915

Title: De novo mutation of NAXE (APOAIBP)-related early-onset progressive encephalopathy with brain edema and/or leukoencephalopathy-1: A case report and literature review

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 05426817
Position: Peer Reviewer
Academic degree: MD, PhD
Professional title: Neurosurgeon
Reviewer's Country/Territory: United States
Author's Country/Territory: China

Manuscript submission date: 2023-02-16

Reviewer chosen by: AI Technique

Reviewer accepted review: 2023-02-20 03:43

Reviewer performed review: 2023-02-21 05:50

Review time: 1 Day and 2 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[ ] Grade A: Priority publishing [Y] Grade B: Minor language polishing [ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [Y] Major revision [ ] Rejection

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Re-review	[ ] Yes [Y] No
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

Interesting paper. Major revision. B for all the answers regarding relevance of topic. Very good Authors should investigate other causes of the edema

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Peer-review model: Single blind

Reviewer's code: 02968018
Position: Peer Reviewer
Academic degree: PhD
Professional title: Associate Professor
Reviewer's Country/Territory: Brazil
Author's Country/Territory: China

Manuscript submission date: 2023-02-16

Reviewer chosen by: Geng-Long Liu

Reviewer accepted review: 2023-03-05 19:16

Reviewer performed review: 2023-03-05 19:57

Review time: 1 Hour

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Novelty of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No novelty

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Creativity or innovation of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No creativity or innovation
Scientific significance of the conclusion in this manuscript	[ ] Grade A: Excellent [ ] Grade B: Good [Y] Grade C: Fair [ ] Grade D: No scientific significance
Language quality	[ ] Grade A: Priority publishing [ ] Grade B: Minor language polishing [Y] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [Y] Major revision [ ] Rejection
Re-review	[Y] Yes [ ] No
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The authors present a case report of clinical interest, which is illustrated by genetic information The subject falls within the scope of the journal. Description and discussion of the findings are well done and well-founded. The bibliography is pertinent and current, but can be expanded. However, the text needs improvement, and the wording should be comprehensively reviewed, preferably by a native English-speaking professional. Excerpts that deserve special attention were marked in yellow. Consider present more images, including diffusion weighted images, SWI, and sagittal T2 of the spinal cord. Consider include more Figures, divided in 1, 2, 3. The images presented are too small. Specific points: 1. cerebellar edema and brain hernia (Ascending transtentorial herniation?), 2. MRI scan showed suspicious T2: REMOVE suspicious 3. Acute disseminated encephalomyelitis (ADEM), also named post-infectious encephalomyelitis and immune-mediated encephalomyelitis, could be considered. ADEM is a

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demyelinating disease of the CNS that usually presents as a monophasic event with multifocal neurologic symptoms, that involves multiple areas of the white matter and spinal cord, typically associated with encephalopathy, mainly affecting children and mostly occurring after recent (1–2 weeks prior) viral or bacterial infections or more rarely after vaccinations, though a specific etiologic agent is not always identified . INCLUDE THE FOLLOWING REFERENCES Esposito S, Di Pietro GM, Madini B, Mastrolia MV, Rigante D. A spectrum of inflammation and demyelination in acute disseminated encephalomyelitis (ADEM) of children. Autoimmun Rev. 2015 Oct;14(10):923-9. Pereira FV, Jarry VM, Castro JTS, Appenzeller S, Reis F. Pediatric inflammatory demyelinating disorders and mimickers: How to differentiate with MRI? Autoimmun Rev. 2021 May;20(5):102801. Reis F, Kobayashi E, Maciel EP, Zanardi Vde A, Netto JR, Damasceno BP, Cendes F. [Magnetic resonance imaging and clinical features in adults with monophasic demyelinating disease. Acute disseminated encephalomyelitis or a variant of multiple sclerosis?]. Arq Neuropsiquiatr. 1999 Sep;57(3B):853-9. When polyfocal demyelination is accompanied by encephalopathy, the clinical diagnosis of acute disseminated encephalomyelitis (ADEM) is applied. Callen DJ, Shroff MM, Branson HM, Li DK, Lotze T, Stephens D, Banwell BL. Role of MRI in the differentiation of ADEM from MS in children. Neurology. 2009 Mar 17;72(11):968-73.

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RE-REVIEW REPORT OF REVISED MANUSCRIPT

Name of journal: *World Journal of Clinical Cases*

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Title: De novo mutation of NAXE (APOAIBP)-related early-onset progressive encephalopathy with brain edema and/or leukoencephalopathy-1: A case report and literature review

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Peer-review model: Single blind

Reviewer's code: 02968018
Position: Peer Reviewer
Academic degree: PhD
Professional title: Associate Professor
Reviewer's Country/Territory: Brazil
Author's Country/Territory: China

Manuscript submission date: 2023-02-16

Reviewer chosen by: Ji-Hong Liu

Reviewer accepted review: 2023-03-29 02:52

Reviewer performed review: 2023-03-31 01:51

Review time: 1 Day and 22 Hours

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[ ] Grade A: Priority publishing [Y] Grade B: Minor language polishing [ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [Y] Major revision [ ] Rejection

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Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

Some topics still need to be improved (see attached file). The MRI findings in APOA1BP are consistent with a nonspecific leukoencephalopathy. The pattern of white matter changes is characterized by T2 hyperintensities involving mainly the deep white matter with sparing of the peripheral and subcortical areas. These changes may represent a toxic or ischemic damage to the glial (non-neuronal) cells such as oligodendrocytes or astrocytes and are typically seen in delayed compared with the rapid (within days) clinical deterioration Although, a majority of these syndromes initially present in infancy and are associated with variable brain pathologies affecting the cerebral cortex, cerebellum, white matter, basal ganglia, and other structures, the value of MR imaging in the correct diagnosis of these syndromes is limited due to radiological overlap in various syndromes and the lack of specificity of pathological MR patterns to certain genetic syndromes Spiegel, R., Shaag, A., Shalev, S. et al. Homozygous mutation in the APOA1BP is associated with a lethal infantile leukoencephalopathy. Neurogenetics 17, 187–190 (2016). MR spectroscopy is a potential tool in this setting because it frequently detects abnormal lactate peaks that correlate highly with other clinical markers in all mitochondriopathies. The detection of lactate is neither specific nor found in all patients in the setting of mitochondrial disorders . Elevated brain lactate is only one of the helpful information in the evaluation of these patients, and its sensitivity shows temporal and regional variation, depending on the type of mitochondrial disorder. The serum lactate detection presents low sensitivity in the diagnosis of mitochondriopathies, because it is transported to the liver where it is normally reoxidized into pyruvate. Therefore, the quantification of this

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metabolite in the CSF provides important information about the presence of anaerobic metabolism in encephalic tissues. MRS is more sensitive during episodes of clinical exacerbation Saneto RP, Friedman SD, Shaw DW. Neuroimaging of mitochondrial disease. Mitochondrion. 2008 Dec;8(5-6):396-413. Pereira FV, Jarry VM, Castro JTS, Appenzeller S, Reis F. Pediatric inflammatory demyelinating disorders and mimickers: How to differentiate with MRI? Autoimmun Rev. 2021 May;20(5):102801.