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7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA
Telephone: +1-925-399-1568
E-mail: bpgoffice@wjgnet.com
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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 79361

Title: Identification of 1q21.1 microduplication in a family

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06137207
Position: Peer Reviewer
Academic degree: MD
Professional title: Doctor, Postdoc, Research Fellow
Reviewer's Country/Territory: Morocco
Author's Country/Territory: China

Manuscript submission date: 2022-08-17

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-09-08 06:18

Reviewer performed review: 2022-09-08 13:01

Review time: 6 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Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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) [Y] Accept (General priority) [ ] Minor revision [ ] Major revision [ ] Rejection
Re-review	[ ] Yes [Y] No
Peer-reviewer	Peer-Review: [Y] Anonymous [ ] Onymous

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statements	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The 1q21.1 microduplication is a chromosomal change in which a small amount of genetic material on chromosome 1 is abnormally copied leading to developmental delay and intellectual disability. Some are born with malformations of the heart. The authors report a case of an 8-month-old girl suffering from convulsion for the past 2 months and they analyzed the clinical features of 1q21.1 microduplication syndrome, and carried out a literature review to determine the correlation between 1q21.1 microduplication and its phenotypes. The study's aim is clearly stated and logical. This is a rare clinical case well written and scientifically discussed by the authors with the conclusions drawn that supported the data. The tables and Figures are well labelled and comprehensive. The study method is sufficiently detailed to allow any other researcher to easily carry on the same study. The authors were able to demonstrate that 1q21.1 microduplication syndrome is really a rare copy number variant disease that can be manifested as multiple congenital developmental disorders. These include developmental delays, autism spectrum disorders, congenital malformations, and congenital heart defects with genetic heterogeneity and various clinical manifestations. The method allows to compare and conclude of whole exon sequencing combined with qPCR providing an accurate molecular diagnosis for children carrying this genetic mutation. There is no evidence-based guidelines or standard recommendations in the literature for the treatment of 1q21.1 microduplication syndrome. Thus, this approach of the management add much to the current available data in the literature. It opens the doors for further investigations. The references are appropriate in number and up-to-date. The conclusion in the abstract is too long. Please be so kind and summarize it. Thank you.