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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 57995

Title: Submicroscopic 11p13 deletion including the elongator acetyltransferase complex subunit 4 gene in a girl with language failure, intellectual disability and congenital malformations: A case report

Reviewer's code: 00564092
Position: Peer Reviewer
Academic degree: PhD
Professional title: Professor
Reviewer's Country/Territory: United States
Author's Country/Territory: Mexico

Manuscript submission date: 2020-07-13
Reviewer chosen by: AI Technique
Reviewer accepted review: 2020-07-13 16:35
Reviewer performed review: 2020-07-13 16:41
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Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
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: [ ] Anonymous [Y] Onymous

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

SPECIFIC COMMENTS TO AUTHORS

This is a case report that is interesting and useful due to the link to a chromosomal abnormality. There are two details I suggest the authors to revise. In the title, delete the "case report and literature review." In the conclusion, delete the word "special" when referring to treatment.

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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 57995

Title: Submicroscopic 11p13 deletion including the elongator acetyltransferase complex subunit 4 gene in a girl with language failure, intellectual disability and congenital malformations: A case report

Reviewer's code: 03258487
Position: Peer Reviewer
Academic degree: MD
Professional title: Professor
Reviewer's Country/Territory: Thailand
Author's Country/Territory: Mexico

Manuscript submission date: 2020-07-13
Reviewer chosen by: AI Technique
Reviewer accepted review: 2020-07-14 11:18
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Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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	[Y] Yes [ ] No
Peer-reviewer	Peer-Review: [ ] Anonymous [Y] Onymous

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

SPECIFIC COMMENTS TO AUTHORS

The authors indicate that their main objective is to describe a girl with dysmorphic features, intellectual disability and congenital malformations without aniridia, and with microdeletion of 11p13 involving ELP4 gene. I found the manuscript is very interesting. Some points require further clarification and the manuscript may benefit by adding more focused discussion about the findings. Case presentation • Line 10-12 on page 7, "(hg 19)" can be added at the end of the sentence "The gene content…… NCBI build 37." And The preceding sentence, "The reference……. (hg 19)." can be removed. Result section. • The exact location of 11p13 microdeletion and LOH of Xq should be given, to be more specific nt start and nt end of the deletion and the LOH. Discussion I suggest the authors adding further discussion on the following issues. • Other possible etiology underlying the unusual phenotypes (cleft lip/palate and neuromigration defect including heterotopia) other than what already been said, for example, a coincidental disorder causing cleft lip/palate and heterotopia such as Baraitser–Winter cerebrofrontofacial syndrome (Verloes et al., Eur J Hum Genet. 2015; 23: 292–301). • Among the 6 genes with X-linked recessive inheritance, is there any gene associated with both cleft lip/palate and neuromigration abnormalities. I suggest the authors go over these 2 papers, Kasper, et al., Epilepsy & Behavior 69 (2017) 104–109 and Ge´cz, et al., Eur J Hum Genet. 2006; 14: 1233–1237. • Page 9. The word "The researchers" is ambiguous which researchers, the authors the submitted manuscript or of which paper. • Page 11, Paragraph 2 Line 3-4 is redundant with Line 3-5 from the bottom of page 7, please consider rewrite it. Line 2-6, there are 6 genes but 7 disorders mentioned, so it is not clear which disorder was associated with which gene. A reference is needed for "Borjeson-Forssman-Lehman syndrome has been associated with cleft palate" Other minor comments • "upward-sloping" should be "upward-slanting" • Author contributions "Molecular Karyotyping"

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should be “molecular karyotyping” • Line 2 from the bottom of page 8, “ELPA” should be “ELP4”

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RE-REVIEW REPORT OF REVISED MANUSCRIPT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 57995

Title: Submicroscopic 11p13 deletion including the elongator acetyltransferase complex subunit 4 gene in a girl with language failure, intellectual disability and congenital malformations: A case report

Reviewer's code: 03258487
Position: Peer Reviewer
Academic degree: MD

Professional title: Professor

Reviewer's Country/Territory: Thailand

Author's Country/Territory: Mexico

Manuscript submission date: 2020-07-13

Reviewer chosen by: Le Zhang

Reviewer accepted review: 2020-09-10 05:47

Reviewer performed review: 2020-09-10 08:58

Review time: 3 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) [Y] Accept (General priority) [ ] Minor revision [ ] Major revision [ ] Rejection
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

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SPECIFIC COMMENTS TO AUTHORS

RE: the revised version of the manuscript number 57995 The authors have addressed to all of my concerns. Thank you. There are few suggested changes, as shown below. ABSTRACT Case summary • Her motor and intellectual development was delayed. • A submicroscopic deletion……...was detected. CASE PRESENTATION • …. Does not sit unsupported. RESULTS • One deletion of 31 kb, (arr [hg38] 11p13: 31509401-31540684)x1 dn, involved exon 1 of IMMP1L, and exon 1 to 3 of the ELP4 gene. • PHF6: Borjeson–Forssman–Lehman syndrome (BFS), HPRT1: Lesh Nyhan syndrome) (Figure 3) and were considered to possess uncertain significance. The parents were clinically healthy with no…… DISCUSSION • four of them, OCRL, AIFM1, PHF6 and HPRT1 genes (HP: 0001249 in OMIM), have been associated with intellectual disability (figure 3).