

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 62249

Title: New mechanism of partial duplication and deletion of chromosome 8: A case report

Reviewer's code: 02445886

Position: Editorial Board

Academic degree: DSc, PhD

Professional title: Chief Doctor

Reviewer's Country/Territory: Russia

Author's Country/Territory: China

Manuscript submission date: 2021-02-21

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-04-08 10:38

Reviewer performed review: 2021-04-08 11:47

Review time: 1 Hour

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input checked="" type="checkbox"/> Grade A: Priority publishing <input type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No
Peer-reviewer statements	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous Conflicts-of-Interest: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No



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SPECIFIC COMMENTS TO AUTHORS

The title reflects the main subject of the manuscript. Chromosomal microarray analysis (CMA) results of 25-week fetus have shown a deletion/duplication of chromosome 8p/8q

(arr[GRCH37]8p23.3p23.2(158048-3220759)x1,8q21.11q24.3(77115706-146295771)x3).

Systematic ultrasound showed that the fetal ventricles widened bilaterally, the measured value of the septum pellucidum was smaller than the normal, and no gallbladder was screened. Cardiac ultrasound screening suggested fetal venous catheter occlusion or absence that need to be further screened. Noninvasive prenatal testing (NIPT) just suggested that the fetus may have a terminal deletion of chromosome 8p. The results of CMA and NIPT are not consistent with each other. Then placental high-throughput sequencing confirmed that the placental long-arm terminal duplication is 40% mosaic, indicating that NIPT may not have a high detection rate when chromosomal copy number variations are with a low proportion of placental mosaic. The conclusion is that recombinant offspring of the chromosome are rarely seen when the inversion segment is shorter than one third of the chromosome length. The extent of the genetic imbalance of these recombinants depends on the relative size of the inversion segment. In terms of the occurrence mechanism of Chromosome 8 duplication and deletion, attention should be paid to the production of unbalanced gametes by the pairing of homologous chromosome during meiosis, and the possibility of mitotic recombination exchanged as well. Thus, it provides reference to clinical evaluation of recurrence risk. The manuscript is well written, the abstract and key words reflect the main topics of the entire text. The discussion is informative and helpful. The article is well illustrated. It cites 11 relevant and important references, but 10 of them are more than 10 years old. I suggest to accept the manuscript after renewing the references.

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 62249

Title: New mechanism of partial duplication and deletion of chromosome 8: A case report

Reviewer's code: 05524014

Position: Peer Reviewer

Academic degree: PhD

Professional title: Research Scientist

Reviewer's Country/Territory: Iran

Author's Country/Territory: China

Manuscript submission date: 2021-02-21

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-04-13 07:55

Reviewer performed review: 2021-04-30 02:04

Review time: 16 Days and 18 Hours

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No
Peer-reviewer statements	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous Conflicts-of-Interest: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No



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SPECIFIC COMMENTS TO AUTHORS

I have some concerns and elaborating them here point by point and based on the sentence no as mentioned in the manuscript (most of them are minor but needs to be corrected) 1- Sentence no 20 page 2- Few literature is wrong. 2- Sentence no20, 21 and 22 page 2- need references. 3- 3- If this case is reported for the first time, it is better to put a part as novel insights in your manuscript. 4- Sentence no 27 it is better to explain more demography and also add the age of the pregnant woman. 5- Figure 4 add arrow indicated the abnormality in your target. 6- Figure legends - is not written completely.it needs complete rewriting. 7- I think it is better to separate material and method from result.