

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 62346

Title: 2 + 0 CY 1 Y deletion carrier: A limitation of the genetic testing and counseling:
A case report

Reviewer's code: 04469475

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Poland

Author's Country/Territory: China

Manuscript submission date: 2021-01-07

Reviewer chosen by: Ya-Juan Ma

Reviewer accepted review: 2021-01-26 09:14

Reviewer performed review: 2021-01-27 09:23

Review time: 1 Day

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" 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 Grade E: Do not publish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 checked="" type="checkbox"/> Accept (General priority) <input 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

SPECIFIC COMMENTS TO AUTHORS

The study is well done, the material is large enough and the methods look reliable. However the study is based on extensive and very recent literature, gives some new information and this warrants its publication.

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 62346

Title: 2 + 0 CY 1 deletion carrier: A limitation of the genetic testing and counseling:
A case report

Reviewer's code: 00742209

Position: Editorial Board

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: United States

Author's Country/Territory: China

Manuscript submission date: 2021-01-07

Reviewer chosen by: Ya-Juan Ma

Reviewer accepted review: 2021-01-26 05:48

Reviewer performed review: 2021-02-02 22:28

Review time: 7 Days and 16 Hours

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input checked="" type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input type="checkbox"/> Grade B: Minor language polishing <input checked="" 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 type="checkbox"/> Minor revision <input checked="" 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

Authors should briefly discuss the limitations of the current testing strategy (PCR-based mutation detection methods with sequencing of the entire gene, and multiplex ligation-dependent probe amplification) to detect silent carriers of pathologic genetic disorders, such as gene duplication paired with gene loss on the opposite chromosome or allele (2+0). Authors can suggest a group discussion with the family, and the group includes a representative from the laboratory that performed the genetic testing, a genetic counselor, and the primary care provider. Authors can say in the text of the manuscript “a limitation of the genetic testing” instead of “a trap.” Authors can say “our report” instead of “our study” in the manuscript, such as in the conclusion of the abstract. Authors can clarify the reference to “this possibility” in the middle of the second paragraph of the discussion section in the manuscript. Authors can clarify “detection of the number of copy variants of CYP21A2 gene” in the conclusion section of the discussion.

RE-REVIEW REPORT OF REVISED MANUSCRIPT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 62346

Title: 2 + 0 CY 1 Y deletion carrier: A limitation of the genetic testing and counseling:
A case report

Reviewer's code: 04469475

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Poland

Author's Country/Territory: China

Manuscript submission date: 2021-01-07

Reviewer chosen by: Le Zhang

Reviewer accepted review: 2021-03-09 08:04

Reviewer performed review: 2021-03-09 08:11

Review time: 1 Hour

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" 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 checked="" type="checkbox"/> Accept (General priority) <input type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
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

SPECIFIC COMMENTS TO AUTHORS



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I would like to thank the authors for the effort they made to write such good manuscript.