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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 71314

Title: Heterozygous deletion in the OTC gene results in ornithine transcarbamylase deficiency: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03409194
Position: Peer Reviewer
Academic degree: MD
Professional title: Doctor
Reviewer's Country/Territory: Pakistan
Author's Country/Territory: China

Manuscript submission date: 2021-09-03

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-09-06 06:39

Reviewer performed review: 2021-09-13 09:08

Review time: 7 Days and 2 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Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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

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

SPECIFIC COMMENTS TO AUTHORS

In the manuscript, "A 10-bp deletion in the OTC gene results in ornithine transcarbamylase deficiency by early translation termination: A case report", Wang and colleagues present an affected boy with Ornithine transcarbamylase deficiency (OTCD) due to a novel deletion variation in OTC. Specific comment: There are more than 500 OTC reported pathogenic variants (http://www.hgmd.cf.ac.uk/ac/gene.php?gene=OTC). Therefore, the statement "…results confirm the pathogenic variation in OTC and provide strong evidence for further OTCD screening and clinical consultation.", should be revised to highlight the contribution of the current report. Minor Comment: 1.On page 2, please provide OMIM number for phenotype Ornithine transcarbamylase deficiency (OTCD). 2. On page 2, in the statement "The OTC gene (OMIM:300461) is located on position Xp11.4, contains 10 exons and 9 introns, and encodes 354 amino acids." Please replace "position" with "chromosome" and add "encodes a 354 amino acids protein", instead of the current wording. 3. On page 3, statements "in the neonatal onset group, it was completely lost," and "the late onset group, it was partially lost"; please replace "was" by "is". 4. On page 3, statement "They are normal at birth, but gradually refuse …" please re-word, "they have no symptoms at birth, but gradually refuse…" 5. On page 3: What are the "molecular function experiments" referred to? The data for biochemical investigations, exome sequencing and treatment is presented but there is no experiment regarding function. 6. On page 5, Please state that the variant is "absent" in all publicly available databases including gnomAD instead of writing "included". 7. For page 5: Please deposit the variant in ClinVar or other comparable databases such as LOVD and

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insert the accession number in the manuscript. 8. On page 6: In the statement "other ornithine circulatory disorders; other genetic metabolic diseases, including organic acid hematic disease, fatty acid, beta oxygen defects, …", please replace "other" with "different" and "miscellaneous" respectively, in order to avoid the use of the word "other" multiple times. 9. On page 7: Please re-word the statement "Sanger sequencing fails in the detection of OTCD in approximately 20% of patients [10, 11], whereas NGS has the advantage of detecting small insertions or deletions". In the cited papers, array CGH or multiplex ligation-dependent probe amplification were used to detect relatively large exon level insertions and deletions which are usually missed by both Sanger and exome sequencing. In special circumstances, exome sequencing may be used to detect these exon level duplications and deletions. However, this does not apply in the case presented here since Sanger sequencing is able to detect small 10bp deletion or insertion which is comparable to the detection by exome sequencing. 10.On page 7, "Our results provide evidence for the pathogenicity of our variant and accurate diagnosis for patients with the same variant." Please re-word since the results do not provide evidence of pathogenicity since no such experiments were performed. However, the pathogenicity is inferred due to the extreme severity of the variant which is present in the gene known to cause the phenotype as detected in the patient. 11.On page 6: In the statement: "…there was no response to stimulation, and the patient was in a coma. The patient died soon after discharge." Please clarify; did the patient recover from coma before discharge? Or was he discharged while in a coma? If he had recovered from coma, then please state whether he had a relapse at home. 12. On page 9, in table 2, please specify in the footnote what NE stands for.

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Reviewer's code: 01344350
Position: Peer Reviewer
Academic degree: MD
Professional title: Doctor
Reviewer's Country/Territory: Germany
Author's Country/Territory: China

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Reviewer performed review: 2021-09-17 07:15

Review time: 3 Days

Scientific quality	[Y] Grade A: Excellent [ ] 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
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Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

This is an interesting and clinically relevant study.