



PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 77064

Title: Two novel mutations in the VP 3B gene in a Chinese patient with ARC : A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06124644

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Iran

Author's Country/Territory: China

Manuscript submission date: 2022-04-13

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-05-04 09:09

Reviewer performed review: 2022-05-04 09:23

Review time: 1 Hour

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input checked="" 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 Grade E: Do not publish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 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



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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

Dear author. The case can be considere a rare one, but it is not well written and does not reveal what you want to explain. The explanation is not enough



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Peer-review model: Single blind

Reviewer's code: 05204241

Position: Editorial Board

Academic degree: FRCP (C)

Professional title: Staff Physician

Reviewer's Country/Territory: Malaysia

Author's Country/Territory: China

Manuscript submission date: 2022-04-13

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-05-11 13:46

Reviewer performed review: 2022-05-15 15:13

Review time: 4 Days and 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 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 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



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SPECIFIC COMMENTS TO AUTHORS

This is an interesting read of a child with cholestasis and hypothyroidism that turned out to have 2 compound heterozygous in the VPS33B gene. Their finding adds on to the understanding of genetic mutations involved in ARCS1. Nevertheless, quite a number of sentences are unclear of their meaning in the case description. Sentence structure needs some revision. Perhaps, this could be relook into by the authors so that the content remains valid and not affected by translation work. Some examples:". Breath sounds in both lungs were rough, and dry and moist rales were not heard., Embrace reflex and sucking reflex could elicit., The ends of the limbs were warm.." and many more.



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Peer-review model: Single blind

Reviewer's code: 05086048

Position: Editorial Board

Academic degree: PhD

Professional title: Postdoctoral Fellow

Reviewer's Country/Territory: Germany

Author's Country/Territory: China

Manuscript submission date: 2022-04-13

Reviewer chosen by: Dong-Mei Wang

Reviewer accepted review: 2022-06-21 15:42

Reviewer performed review: 2022-06-24 19:29

Review time: 3 Days and 3 Hours

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



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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

It was a pleasure to read this article. Everything seems well written and, in the near future, information present in this article may be useful for clinical application. Thus, manuscript may be accepted in the present form.



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Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06079635

Position: Peer Reviewer

Academic degree: BSc, MSc, PhD

Professional title: Assistant Professor

Reviewer's Country/Territory: Pakistan

Author's Country/Territory: China

Manuscript submission date: 2022-04-13

Reviewer chosen by: Dong-Mei Wang

Reviewer accepted review: 2022-06-23 11:12

Reviewer performed review: 2022-06-27 08:20

Review time: 3 Days and 21 Hours

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 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



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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 case-report, 'Two novel mutation in the VPS33B gene in a Chinese patient with ARCS1 : A case report', presented here has identified two novel mutations in the gene responsible for the ARCS1 disease. The case-report presents here two new mutations with a potential for employed in genetic counselling and testing. This is an interesting work of investigation and few comments for the clarity of work presented are: 1) The manuscript file attached is not the final formatted version and still incorporates the highlighted changes. 2) The manuscript could benefit from minor language editing, for example, the word 'mutation' in the title should be 'mutations' to show the plural nature of the work described 3) Introduction section has not detailed the role of genes mentioned and just alluded the names as VPS33B and VPS33B. They should be elaborated with their role in the disease pathogenesis. 4) There are no details about the whole-exome sequencing and CNV analysis being referred to, that should also be clearly explained with the methodology adopted. 5) The manuscript has not detailed the role of reported mutations in the pathogenicity of the disease and how they concluded their role in this particular phenotype. 6) Figure 2 and Figure 3 have not detailed the methodology used to obtain those chromatograms shown.



RE-REVIEW REPORT OF REVISED MANUSCRIPT

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Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06079635

Position: Peer Reviewer

Academic degree: BSc, MSc, PhD

Professional title: Assistant Professor

Reviewer's Country/Territory: Pakistan

Author's Country/Territory: China

Manuscript submission date: 2022-04-13

Reviewer chosen by: Han Zhang

Reviewer accepted review: 2022-07-27 09:34

Reviewer performed review: 2022-07-27 09:56

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	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous



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

SPECIFIC COMMENTS TO AUTHORS

The manuscript has been revised and authors have made the suggested changes. The manuscript still needs a thorough editing for grammar and typos particularly the section on physical examination. Also, the primer sequences should be added that were used to sequence the region of the gene mentioned in the manuscript. It will also be good to incorporate the images to show the predicted structural variation upon analysis by REVEL.



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Reviewer's code: 05086048

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Academic degree: PhD

Professional title: Postdoctoral Fellow

Reviewer's Country/Territory: Germany

Author's Country/Territory: China

Manuscript submission date: 2022-04-13

Reviewer chosen by: Han Zhang

Reviewer accepted review: 2022-07-27 19:54

Reviewer performed review: 2022-07-27 19:57

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 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
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Peer-reviewer	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous



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

SPECIFIC COMMENTS TO AUTHORS

Manuscript may be accepted in the present form



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Reviewer's code: 05204241

Position: Editorial Board

Academic degree: FRCP (C)

Professional title: Staff Physician

Reviewer's Country/Territory: Malaysia

Author's Country/Territory: China

Manuscript submission date: 2022-04-13

Reviewer chosen by: Han Zhang

Reviewer accepted review: 2022-07-27 13:29

Reviewer performed review: 2022-08-03 00:05

Review time: 6 Days and 10 Hours

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 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 checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
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Conflicts-of-Interest: [] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

There are major typo errors in this version. "There were no obvious abnormalities in routine blood examination. Liver function showed: total bilirubin (TBiL) 229.7 +ACY-mu+ADs-mol/L, direct bilirubin (DBil) 152.4 +ACY-mu+ADs-mol/L, and total bile acid (TBA) 50.6 +ACY-mu+ADs-mol/L. Neonatal jaundice was diagnosed.+ADw-/p+AD4APA-/html+AD4-" i am not sure if this errors occurred due to the incompatible format but this makes the whole manuscript difficult to read. otherwise, the scientific content is acceptable