

PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 79911

Title: Compound heterozygous mutations in PMFB Y cause acephalic spermatozoa syndrome: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03208929

Position: Peer Reviewer

Academic degree: PhD

Professional title: Professor

Reviewer's Country/Territory: China

Author's Country/Territory: China

Manuscript submission date: 2022-09-11

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-09-11 16:34

Reviewer performed review: 2022-09-18 15:18

Review time: 6 Days and 22 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 Grade E: Do not publish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 checked="" type="checkbox"/> Yes <input type="checkbox"/> No

**Peer-reviewer
statements**

Peer-Review: [☒ Y] Anonymous [☐] Onymous
Conflicts-of-Interest: [☐] Yes [☒ Y] No

SPECIFIC COMMENTS TO AUTHORS

The title, abstract, key words and background expression are accurate and clear, the biostatistics, references, research methods and reports are reasonable, and the manuscripts are organized and presented in an orderly, concise and coherent manner. This study proposed new findings and new phenomena, but the manuscript did not describe the experimental method in sufficient detail and did not carry out experimental verification on the new findings. Two newly discovered heterozygous mutants: c414+1G>T (p.?) and c.393del (p.C132Afs * 3) Will really affect the function of PMFBP1? The current results seem to have insufficient support for the conclusion, so the author is suggested to conduct further experimental verification. In terms of discussion, discussion on newly discovered heterozygous mutants should be added.

1、 This study proposed new findings and new phenomena, but the manuscript did not describe the experimental method in sufficient detail and did not carry out experimental verification on the new findings.

We add the experimental method in detail: **whole-exome sequencing (WES)、 DNA Library Preparation、 Targeted genes enrichment and sequencing、 Bioinformatics analysis、 Variants Selected。**

2、 The current results seem to have insufficient support for the conclusion, so the author is suggested to conduct further experimental verification. In terms of discussion, discussion on newly discovered heterozygous mutants should be added.

In this case, the gene mutation was associated with abnormal sperm morphology, which will be confirmed by gene knockout experiment in the next step.

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Title: Compound heterozygous mutations in PMFB cause acephalic spermatozoa syndrome: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 04153245

Position: Editorial Board

Academic degree: BM BCh, MD, MSc, PhD

Professional title: Professor

Reviewer's Country/Territory: Egypt

Author's Country/Territory: China

Manuscript submission date: 2022-09-11

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-10-03 05:21

Reviewer performed review: 2022-10-03 17:53

Review time: 12 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 type="checkbox"/> Minor revision <input checked="" type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No

**Peer-reviewer
statements**

Peer-Review: [☐] Anonymous [☒] Onymous
Conflicts-of-Interest: [☐] Yes [☒] No

SPECIFIC COMMENTS TO AUTHORS

Scientific language contains some errors. Please, re-check. Abstract: The mentioned conclusion is a recommendation & not a conclusion. Please, add it before recommendation. This phrase "To date, the following have" is to be corrected to "To date, the followings have ". History of past illness: It has been mentioned that the wife underwent an induced abortion. Please, clarify its cause for induction of abortion in this case. This phrase "and 200 sperm were counted" is to be corrected to "and 200 sperms were counted". This phrase "there were no normal sperm in the semen" s to be corrected to "there were no normal sperms in the semen". This word "progressivey" is wrong. Please, correct it. This "headless sperm tails" is wrong. Please, delete the word "tails". Add this reference to discussion that some authors attributed decreased rate of spermatogenesis in some cases of idiopathic infertility to high rates of apoptosis during normal spermatogenesis. Hegazy R, Hegazy A, Ammar M, Salem E. Immunohistochemical measurement and expression of Mcl-1 in infertile testes. *Frontiers of Medicine*. 2015 Sep;9(3):361-7.

- 1、 This phrase "To date, the following have" is to be corrected to **"To date, the followings have "**.
- 2、 History of past illness: It has been mentioned that the wife underwent an induced abortion. Please, clarify its cause for induction of abortion in this case.
They don't want to have children themselves and choose abortion. It's not fetal problem
- 3、 This phrase "and 200 sperm were counted" is to be corrected to **" 200 sperms were counted".**
- 4、 This phrase "there were no normal sperm in the semen" s to be corrected to **"there were no normal sperms in the semen"**
- 5、 This "headless sperm tails" is wrong. **delete the word "tails".**
- 6、 Add this reference to discussion that some authors attributed decreased rate of spermatogenesis

in some cases of idiopathic infertility to high rates of apoptosis during normal spermatogenesis. Hegazy R, Hegazy A, Ammar M, Salem E. Immunohistochemical measurement and expression of Mcl-1 in infertile testes. *Frontiers of Medicine*. 2015 Sep;9(3):361-7.

This study mainly discusses the problem of gene and sperm. It has little relevance to this literature and, therefore, it was not added