

## PEER-REVIEW REPORT

**Name of journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 81960

**Title:** Clinical and genetic features of Kenny-Caffey syndrome type 2 with multiple electrolyte disturbances: A case report with literature review

**Provenance and peer review:** Unsolicited Manuscript; Externally peer reviewed

**Peer-review model:** Single blind

**Reviewer's code:** 05329239

**Position:** Peer Reviewer

**Academic degree:** MD

**Professional title:** Doctor

**Reviewer's Country/Territory:** Romania

**Author's Country/Territory:** China

**Manuscript submission date:** 2022-12-02

**Reviewer chosen by:** AI Technique

**Reviewer accepted review:** 2022-12-02 13:46

**Reviewer performed review:** 2022-12-07 18:59

**Review time:** 5 Days and 5 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 Grade E: Do not publish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 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

<b>Peer-reviewer statements</b>	Peer-Review: [ <input checked="" type="radio"/> ] Anonymous [ <input type="radio"/> ] Onymous
	Conflicts-of-Interest: [ <input type="radio"/> ] Yes [ <input checked="" type="radio"/> ] No

## SPECIFIC COMMENTS TO AUTHORS

I congratulate the authors for a nice clinical description of a case with Kenny-Caffey syndrome and for the appropriate genetic assessment. However I have some issues to discuss: 1. Medical English should be corrected as sometimes there are inconsistencies: e.g.: U/I instead of U/L, etc 2. Based only upon the ultrasound aspect of the ovaries, PCOS can not be excluded. The authors should either take into consideration further dosage of hormones to prove hyperandrogenism (as Guidelines suggest - they said that some high levels of testosterone were detected, but no details about it), or make a discussion upon the possible causes of her menstrual irregularity at 18 years old.

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**Provenance and peer review:** Unsolicited Manuscript; Externally peer reviewed

**Peer-review model:** Single blind

**Reviewer's code:** 02441085

**Position:** Editorial Board

**Academic degree:** PhD

**Professional title:** Associate Professor

**Reviewer's Country/Territory:** Thailand

**Author's Country/Territory:** China

**Manuscript submission date:** 2022-12-02

**Reviewer chosen by:** AI Technique

**Reviewer accepted review:** 2022-12-30 22:48

**Reviewer performed review:** 2023-01-03 02:09

**Review time:** 3 Days and 3 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
Novelty of this manuscript	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Good <input checked="" type="checkbox"/> Grade C: Fair <input type="checkbox"/> Grade D: No novelty
Creativity or innovation of this manuscript	<input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Good <input type="checkbox"/> Grade C: Fair <input type="checkbox"/> Grade D: No creativity or innovation

<b>Scientific significance of the conclusion in this manuscript</b>	<input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Good <input type="checkbox"/> Grade C: Fair <input type="checkbox"/> Grade D: No scientific significance
<b>Language quality</b>	<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
<b>Conclusion</b>	<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
<b>Re-review</b>	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No
<b>Peer-reviewer statements</b>	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 authors detected a missense variant of FAM111A in a patient with an acute onset of psychiatric symptoms combined with short stature, electrolyte disorders including hypomagnesemia and hypocalcemia, and abnormal skeletal manifestations consistent with KCS2. The R569H variant was similar to other reported pathogenic variants and was consistent with the proposed pathophysiological mechanisms. However, the patient showed different manifestations, including overweight, severe electrolyte disorder, and psychiatric disorders. The authors suggested that the FAM111A mutation was more likely to be a de novo mutation.