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PEER-REVIEW REPORT

Name of journal: *World Journal of Medical Genetics*

Manuscript NO: 83721

Title: Mosaicism of a novel variant in the ANKRD11 gene in a child with a mild KBG phenotype: a case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 00742373
Position: Editorial Board
Academic degree: MD, PhD
Professional title: Professor
Reviewer's Country/Territory: China
Author's Country/Territory: Italy

Manuscript submission date: 2023-02-06

Reviewer chosen by: Geng-Long Liu

Reviewer accepted review: 2023-03-24 02:09

Reviewer performed review: 2023-03-30 08:31

Review time: 6 Days and 6 Hours

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Novelty of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No novelty
Creativity or innovation of this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No creativity or innovation

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Scientific significance of the conclusion in this manuscript	[ ] Grade A: Excellent [Y] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No scientific significance
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
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The manuscript reported a case of somatic mosaicism of KBG syndrome diagnosed in childhood. The study described the patient's physical abnormalities with pituitary hypoplasia and central hypothyroidism. A novel deletion in the ANKRD11 gene (c.4880-4893del.) was found in this patient, which was considered as the causative of the non-specific phenotype of the KBG syndrome. The manuscript concluded that the next generation sequencing may be helpful in genetic diagnosis for those somatic mosaicisms in children. KBG syndrome is a rare autosomal dominant disorder with delay development an variety spectrum of clinical phenotypes. It is likely underdiagnosed because of its features are often mild and non-specific. Furthermore, the definitive diagnosis of KBG syndrome is rarely achieved as early time. This report found the gene mosaicism of the novel variant in ANKRD11 gene in child. It increased the awareness of variant forms of KBG syndrome in children and helpful for readers to recognize the significance of genetic diagnosis. This case report is significant for recognizing KBG syndrome and new diagnosis methods. It provided the detail clinical description and compared typical reports from literature. English writing was well except for mild

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polishing (such as page 2 line 6). Figures and tables were designed in good quality.

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Peer-review model: Single blind

Reviewer's code: 06090125
Position: Peer Reviewer
Academic degree: MD

Professional title: Lecturer, Technical Editor

Reviewer's Country/Territory: Iraq

Author's Country/Territory: Italy

Manuscript submission date: 2023-02-06

Reviewer chosen by: Geng-Long Liu

Reviewer accepted review: 2023-04-21 14:57

Reviewer performed review: 2023-04-23 14:35

Review time: 1 Day and 23 Hours

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Novelty of this manuscript	[Y] Grade A: Excellent [ ] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No novelty
Creativity or innovation of this manuscript	[Y] Grade A: Excellent [ ] Grade B: Good [ ] Grade C: Fair [ ] Grade D: No creativity or innovation

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Conclusion	[ ] Accept (High priority) [Y] Accept (General priority) [ ] Minor revision [ ] Major revision [ ] Rejection
Re-review	[ ] Yes [Y] No
Peer-reviewer statements	Peer-Review: [ ] Anonymous [Y] Onymous
	Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

This manuscript is well and good at innovation and clears the clarity of the reader. It is well structured and well written. The author does a good job of presenting a highly technical and complicated process in an easy-to-understand manner. Authors need to cross check the reference section by addressing the cited contents in the introduction and related work part. The introduction must be an extended version of the abstract. The authors must elaborate on the points highlighted on the abstract and give supportive ideas and references. The conclusions in this manuscript are primitive. Rewrite your conclusions. References aren't formatted according to rules.