

To the Editor

World Journal of Medical Genetics

Object: Manuscript re-submission – NO. 83721, Case Report

“Mosaicism of a novel variant in the *ANKRD11* gene in a child with a mild KBG phenotype”

Dear Editor,

Thank you for giving us the opportunity of submitting a revised version of our manuscript “Mosaicism of a novel variant in the *ANKRD11* gene in a child with a mild KBG phenotype”

We thank the reviewers for their valuable comments, please find enclosed a revised version of our manuscript and answers to the reviewers’ comments.

Please let me know if any additional changes are required.

Yours sincerely,

Evelina Maines

Answers to reviewers’ comments

Reviewer #1

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.

We are very thankful for the valuable comments of the reviewer. The criticisms prompted us to improve our work. We greatly appreciate the opportunity to submit a revised version of our manuscript.

Authors need to cross check the reference section by addressing the cited contents in the introduction and related work part.

Answer: thank you for your suggestion. We checked the references of contents cited in the introduction.

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.

Answer: thank you for your suggestion. We improved the introduction taking into account your comment.

The conclusions in this manuscript are primitive. Rewrite your conclusions.

Answer: thank you for your suggestion. We improved the conclusions taking into account your comment.

References aren't formatted according to rules.

Answer: we checked references according to the guidelines for manuscript type

Reviewer #2

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

We are very thankful for the positive comments of the reviewer.

We performed mild language polishing and we provide a new language certificate along with the manuscript.

Company editor-in-chief

Please provide the original figure documents.

Please prepare and arrange the figures using PowerPoint

If the picture is 'original', the author needs to add the following copyright information to the bottom right-hand side of the picture in PowerPoint (PPT): Copyright ©The Author(s) 2023.

Answer: we checked figure according to the suggestion and we provide figure in PPT format.