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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 67278

Title: A novel KDM6A mutation in a Chinese infant with Kabuki syndrome: A case report

Reviewer's code: 05260764
Position: Peer Reviewer
Academic degree: Doctor, MD
Professional title: Surgeon
Reviewer's Country/Territory: Denmark
Author's Country/Territory: China

Manuscript submission date: 2021-04-20
Reviewer chosen by: AI Technique
Reviewer accepted review: 2021-06-29 09:41
Reviewer performed review: 2021-06-29 11:53
Review time: 2 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[ ] Grade A: Priority publishing [ ] Grade B: Minor language polishing [Y] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [Y] Major revision [ ] Rejection
Re-review	[Y] Yes [ ] No
Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

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SPECIFIC COMMENTS TO AUTHORS

To the authors: This is an interesting case report of a child born with KS and a novel frameshift mutation in KDM6. However, the manuscript needs some major revision before being eligible for publication. 1. Core tip is too long and more or less a copy of the abstract. It should be shortened to be more concise 2. The introduction needs a bit of attention. a. In line 80, page 5, you mention KS type 2 – however a type 1 has never been mentioned and how they separate from each other. It is mentioned in the discussion, but should be moved to the introduction. b. It would be nice if the authors briefly described the function of KMT2D and KDM6A in order to better understand the mechanisms of the disease. c. You do not mention anything about prenatal ultrasound as a diagnostic tool for KS. With multi-system manifestations in so many cases, you would think that a prenatal ultrasound in gestational week 20+ would find most of the cases. Then amniotic fluid testing could be done and KS would be diagnosed before delivery, giving the children the most optimal pre- and postnatal care. This is standard in Denmark and many European countries, but I am not familiar with the practice in China. A paragraph regarding this would be advised. 3. The case overall a fine presentation, but there is many repetitions, a very confusing description of the examinations and findings within each organ system where everything is mixed together needs a bit of a revision to make it concise and easily read. a. Line 98-99, page 6 – a description of further prenatal sonographic findings as well as the gestational weeks in which the mother was scanned would be nice to add. b. Line 100-102, page 6 – you mention that the mother did not have a fever. When? During the entire pregnancy, before conception or at delivery? Is there any literature supporting that fever is a risk factor for developing KS, since you have mentioned it? Furthermore, you mention that the father did not use tobacco, alcohol or illegal drugs. Again, does this predispose to KS

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since you mention it? c. Line 107-109, page 6 – you describe that the patient was treated for his anal atresia and spent 3 weeks in the NICU – is there any description regarding the neonatal examination and whether or not a congenital disease was suspected before discharge? d. First line in line 140, page 8 – discussion, should not be in the case presentation e. Line 147-150, page 8 – ethical approval needs to be an independent paragraph. 4. The discussion is far too long for a case report and overall a bit confusing to read. You mention a lot of studies with all sorts of mutations, as well as many different presentations of the KS syndrome as well as the genetic concerns regarding further conception – and it is a bit too much. I would recommend to shorten it as well as keep your focus on the KMT2D and KDM6A mutations, their clinical manifestations and how to evaluate the patient with ultrasound in utero to secure the patient the best possible care following delivery.