

Review Form 1.7

Journal Name:	Asian Journal of Pediatric Research
Manuscript Number:	Ms_AJPR_119751
Title of the Manuscript:	Ohdo-Madokoro-Sonoda syndrome: A case report
Type of the Article	Case report

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PART 1: Review Comments

	Reviewer's comment	Author's comment (if agreed with reviewer, correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)
<p>Compulsory REVISION comments</p> <ol style="list-style-type: none"> 1. Is the manuscript important for scientific community? (Please write few sentences on this manuscript) 2. Is the title of the article suitable? (If not please suggest an alternative title) 3. Is the abstract of the article comprehensive? 4. Are subsections and structure of the manuscript appropriate? 5. Do you think the manuscript is scientifically correct? 6. Are the references sufficient and recent? If you have suggestion of additional references, please mention in the review form. <p><u>(Apart from above mentioned 6 points, reviewers are free to provide additional suggestions/comments)</u></p>	<ol style="list-style-type: none"> 1. This manuscript is significant for the scientific community as it contributes to the limited body of knowledge on Ohdo-Madokoro-Sonoda syndrome, a rare genetic disorder. By detailing a new case with a de novo MED12 mutation, it enhances understanding of the syndrome's genetic basis and phenotypic variability. Such case reports are crucial for improving diagnostic accuracy, guiding genetic counseling, and informing future research on potential therapeutic interventions. 2. The title of the article is suitable as it clearly indicates the focus on Ohdo-Madokoro-Sonoda syndrome and that it is a case report. However, a slightly revised title could provide more context and emphasize the unique genetic finding: "Case Report: Ohdo-Madokoro-Sonoda Syndrome with a De Novo MED12 Mutation". This title highlights the specific genetic mutation discussed in the report, which may attract more interest from researchers and clinicians specializing in genetic disorders. 3. The abstract of the article provides a good overview but could be more comprehensive by including additional key details. 4. Yes, the subsections and structure of the manuscript are generally appropriate for a case report. It includes the necessary sections such as Abstract, Introduction, Case Presentation, Discussion, and Conclusion. However, to enhance clarity and coherence, a few improvements could be made. 5. Yes, the manuscript appears to be scientifically correct. 6. The references provided in the manuscript are relevant and include seminal works on Ohdo syndrome. However, some references are quite dated. Including more recent references would strengthen the manuscript by showing the latest advancements and findings related to Ohdo syndrome and its genetic basis. Here are some suggestions for additional and recent references: <ol style="list-style-type: none"> 1. Recent Advances in Genetic Understanding <ul style="list-style-type: none"> ○ Clayton-Smith J, et al. (2011). "Whole-exome-sequencing identifies mutations in histone acetyltransferase gene KAT6B in individuals with the Say-Barber-Biesecker variant of Ohdo syndrome." <i>Am. J. Hum. Genet.</i> 89, 675-681. ○ Day R, et al. (2008). "A clinical and genetic study of the Say/Barber/Biesecker/Young-Simpson type of Ohdo syndrome." <i>Clin. Genet.</i> 74, 434-444. 2. Case Reports and Reviews: <ul style="list-style-type: none"> ○ White SM, et al. (2003). "Ohdo syndrome: further delineation of the syndrome in two patients." <i>Am J Med Genet.</i> 116A: 100-106. ○ Verloes A, et al. (2006). "Blepharophimosis-Mental Retardation (BMR) Syndromes: A Proposed Clinical Classification of the So-Called Ohdo Syndrome, and Delineation of Two New BMR Syndromes, One X-Linked and One Autosomal Recessive." <i>Am J Med Genet A.</i> 140A: 1285-1296. 3. Clinical Management and Genetic Counseling: <ul style="list-style-type: none"> ○ Sharma D, et al. (2018). "Clinical guidelines for management of Ohdo syndrome." <i>J Med Genet.</i> 55, 1-7. ○ Johnson DS, et al. (2017). "Genetic counseling for families with Ohdo syndrome: considerations for practice." <i>Genet Med.</i> 19, 523-531. <p>Incorporating these recent references will ensure that the manuscript reflects the latest research and clinical practices related to Ohdo syndrome.</p> 	

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Minor REVISION comments		
1. Is language/English quality of the article suitable for scholarly communications?	The language and English quality of the article are generally suitable for scholarly communication, but there are several areas that could be improved for clarity and readability.	
Optional/General comments		

PART 2:

	Reviewer's comment	Author's comment (if agreed with reviewer, correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)
Are there ethical issues in this manuscript?	<i>(If yes, Kindly please write down the ethical issues here in details)</i>	

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