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PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 59438

Title: One case of apert syndrome diagnosed by prenatal ultrasound combined with magnetic resonance image and whole exome sequencing: A case report

Reviewer's code: 02512546
Position: Peer Reviewer
Academic degree: PhD

Professional title: Professor

Reviewer's Country/Territory: Brazil

Author's Country/Territory: China

Manuscript submission date: 2020-09-26

Reviewer chosen by: Le Zhang

Reviewer accepted review: 2020-10-20 12:14

Reviewer performed review: 2020-11-13 01:13

Review time: 23 Days and 12 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Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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

World Journal of Case Report Case Report Title: One Case of Apert Syndrome Diagnosed by Prenatal Ultrasound Combined with Magnetic Resonance Image and Whole Exome Sequencing Authors: Lei Chen, Feixiang Huang The manuscript has great scientific importance, highlighting new aspects in the early diagnosis of AS, combining prenatal ultrasound with MRI and the genetic diagnosis through the whole exome sequence. 1 Title. Does the title reflect the main subject/hypothesis of the manuscript? Yes 2 Abstract. Does the abstract summarize and reflect the work described in the manuscript? Yes 3 Key words. Do the key words reflect the focus of the manuscript? No, see the comments 4 Background. Does the manuscript adequately describe the background, present status and significance of the study? Yes 5 Methods. Does the manuscript describe methods (e.g., experiments, data analysis, surveys, and clinical trials, etc.) in adequate detail? Yes 6 Results. Are the research objectives achieved by the experiments used in this study? What are the contributions that the study has made for research progress in this field? The association of prenatal Ultrasound and RMN in the screening of Apert Syndrome as well as the whole exome sequencing for the genetic diagnosis 7 Discussion. Does the manuscript interpret the findings adequately and appropriately, highlighting the key points concisely, clearly and logically? Are the findings and their applicability/relevance to the literature stated in a clear and definite manner? Is the discussion accurate and does it discuss the paper's scientific significance and/or relevance to clinical practice sufficiently? Yes 8 Illustrations and tables. Are the figures, diagrams and tables sufficient, good quality and appropriately illustrative of the paper contents? Do figures require labeling with arrows, asterisks etc., better legends? There are dozens of gramar mistakes also in the legends 9 Biostatistics. Does the manuscript meet the requirements of biostatistics? Not aplicable 10 Units. Does the

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manuscript meet the requirements of use of SI units? Yes 11 References. Does the manuscript cite appropriately the latest, important and authoritative references in the introduction and discussion sections? Does the author self-cite, omit, incorrectly cite and/or over-cite references? The references must be standardized. 12 Quality of manuscript organization and presentation. Is the manuscript well, concisely and coherently organized and presented? Is the style, language and grammar accurate and appropriate? The language of this manuscript must be edited. Please check your manuscript carefully and throughout. There are dozens of grammar mistakes. 13 Research methods and reporting. Authors should have prepared their manuscripts according to manuscript type and the appropriate categories, as follows: (1) CARE Checklist (2013) - Case report; (2) CONSORT 2010 Statement - Clinical Trials study, Prospective study, Randomized Controlled trial, Randomized Clinical trial; (3) PRISMA 2009 Checklist - Evidence-Based Medicine, Systematic review, Meta-Analysis; (4) STROBE Statement - Case Control study, Observational study, Retrospective Cohort study; and (5) The ARRIVE Guidelines - Basic study. Did the author prepare the manuscript according to the appropriate research methods and reporting? Yes, the authors presented the CARE checklist 14 Ethics statements. For all manuscripts involving human studies and/or animal experiments, author(s) must submit the related formal ethics documents that were reviewed and approved by their local ethical review committee. Did the manuscript meet the requirements of ethics? Yes, the authors presented the signed informed consent COMMENTS TO THE AUTHORS Abstract: Conclusion: We conclude ultrasound combined with MRI are important tools to the prenatal imaging examination technology to find early fetal morphological abnormalities and improve the awareness of AS. Keywords: Apert syndrome, Craniosynostosis, Syndactyly, Prenatal ultrasound, Magnetic resonance image, Whole-exome sequencing The words: Craniosynostosis and Syndactyly do not appear in the abstract and should not be used as key-words.

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Introduction Older paternal age is considered to be a high-risk. Dozens of corrections were effectuated in the manuscript At 31,5/7 weeks of gestation, the patient received ultrasound-guided umbilical vein puncture plus amniotic rivanol cavity The authors must add the name of drug, nethacridine monolactate monohydrate (rivanol) - manufacturer, country of origin 3.5ml umbilical blood was successfully extracted during the operation Do not start the phrase with numbers. A solution of 3.5ml umbilical blood was successfully extracted during the operation. Fig.5 Schematic diagram of Sanger sequencing verification results of the tested subjects and their parents with FGFR2 gene variation The authors should indicate the laboratory where the whole exome was performed. Discussion AS is one of the most common severe cranial maxillofacial deformity syndromes. It is first reported by French pediatrician Apert in 1906 This information was previously cited and must be deleted. Is characterized by craniofacial deformity and syndactyly, and is often accompanied by varying degrees of mental retardation and other abnormalities [ ]. Premature coronal suture closure, acrocephalic head, prominent forehead, abnormal development in the middle of the face, the collapse of nasal root, and syndactyly were found in our case, which was consistent with previous reports. Please, give one or more references for the citation. Older paternal age is considered to be a high-risk factor for AS [ ]. With the increase of paternal age, the frequency of gene FGFR2 mutations was also increased, and the corresponding incidence of AS was also increased. The language of this manuscript must be edited. Please check your manuscript carefully and throughout. There are dozens of grammar mistakes. It is speculated that missed diagnosis may be related to the low incidence, insufficient diagnosis experience, and cognition of the disease. In addition, due to the close proximity between fingers and toes of the fetus in utero and low mobility, ultrasound is not easy to observe, which may also cause some missed diagnosis. Ultrasound is the preferred method of prenatal screening, but it is affected by fetal position and resolution of the

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instrument. Fetal MRI can be used to scan various sections with high soft-tissue resolution. It has little influence on the technical level of operator's instrument The paragraph is lacking in references. Conclusion We conclude ultrasound combined with MRI are important tools to improve the prenatal imaging examination technology to find early fetal morphological abnormalities and improve the awareness of AS. References The references must follow the instructions of the authors. They are out of the standard format of World Journal of Case Report Abstract: Conclusion: We conclude ultrasound combined with MRI are important tools to the prenatal imaging examination technology to find early fetal morphological abnormalities and improve the awareness of AS. Keywords: Apert syndrome, Craniosynostosis, Syndactyly, Prenatal ultrasound, Magnetic resonance image, Whole-exome sequencing The words: Craniosynostosis and Syndactyly do not appear in the abstract and should not be used as key-words. Introduction Older paternal age is considered to be a high-risk. Dozens of corrections were effectuated in the manuscript At 31,5/7 weeks of gestation, the patient received ultrasound-guided umbilical vein puncture plus amniotic rivanol cavity The authors must add the name of drug, nethacridine monolactate monohydrate (rivanol) - manufacturer, country of origin 3.5ml umbilical blood was successfully extracted during the operation Do not start the phrase with numbers. A solution of 3.5ml umbilical blood was successfully extracted during the operation. Fig.5 Schematic diagram of Sanger sequencing verification results of the tested subjects and their parents with FGFR2 gene variation The authors should indicate the laboratory where the whole exome was performed. Discussion AS is one of the most common severe cranial maxillofacial deformity syndromes. It is first reported by French pediatrician Apert in 1906 This information was previously cited and must be deleted. Is characterized by craniofacial deformity and syndactyly, and is often accompanied by varying degrees of mental retardation and other abnormalities [ ]. Premature coronal suture closure, acrocephalic

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