

PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 80226

Title: Clinical manifestations of adult hereditary spherocytosis with novel SPTB gene mutations and hyperjaundice: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 05393679

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: India

Author's Country/Territory: China

Manuscript submission date: 2022-10-05

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-10-19 00:52

Reviewer performed review: 2022-10-30 14:26

Review time: 11 Days and 13 Hours

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|--------------------|---|
| Scientific quality | <input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish |
| Language quality | <input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection |
| Conclusion | <input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection |
| Re-review | <input checked="" type="checkbox"/> Yes <input type="checkbox"/> No |

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|-----------------------------|---|
| Peer-reviewer statements | Peer-Review: [<input type="checkbox"/>] Anonymous [<input checked="" type="checkbox"/>] Onymous |
| | Conflicts-of-Interest: [<input type="checkbox"/>] Yes [<input checked="" type="checkbox"/>] No |

SPECIFIC COMMENTS TO AUTHORS

Is a good study of identification of a new mutation on chromosome 14 resulting in a variant of hereditary spherocytosis resulting in appropriate management of the clinical problem. My concern is why cholecystectomy was not done during splenectomy which is commonly carried out in Hereditary spherocytosis with gallstones. Eventhough there was no anaemia hemolysis was certainly present. Otherwise this is a good research .

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Peer-review model: Single blind

Reviewer's code: 03730113

Position: Peer Reviewer

Academic degree: PhD

Professional title: Academic Research

Reviewer's Country/Territory: India

Author's Country/Territory: China

Manuscript submission date: 2022-10-05

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-11-08 05:17

Reviewer performed review: 2022-11-17 03:44

Review time: 8 Days and 22 Hours

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|--------------------|---|
| Scientific quality | <input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish |
| Language quality | <input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection |
| Conclusion | <input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection |
| Re-review | <input checked="" type="checkbox"/> Yes <input type="checkbox"/> No |

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|-------------------------------------|---|
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| | Conflicts-of-Interest: [<input type="checkbox"/>] Yes [<input checked="" type="checkbox"/>] No |

SPECIFIC COMMENTS TO AUTHORS

The case report by Ni Jiang et al., is a good read. Few suggestions to improve the work are given below: Comments • In the case summary (Page 2) it is advised to add the source of DNA i.e., either blood or tissue. Also, the genotype of the variant (heterozygous or homozygous) needs to be given. • It is surprising that the variant was not seen in the parents. The authors should clarify whether they are the biological parents. Has adoption been ruled out? If so that needs to be mentioned. Have the authors confirmed the variant by drawing an independent blood sample? • The authors in the introduction mention that HS is an autosomal recessive hereditary disease. However, they have identified a heterozygous variant in the SPTB gene (c.1801C>T). How do they justify the causal role of the variant? • In personal and family History (Page 4), although the personal history of the patient is given no mention as to any other family members being affected is not mentioned. • Under Physical examination, have the authors collected the Waist circumference and Hip circumference? • Although the authors mention that “High-throughput sequencing of a liver panel” on Page 5, there is no mention of the panel they have used, the genes they have screened. It would be beneficial to mention the liver panel genes and the platform for the high through-put sequencing that was used (Illumina/Ion torrent). • Were any other variants identified in the patient? Any VUS variants identified? • The rsID for the identified variant should be given. • It is preferable to call the changes in DNA as variants rather than mutation. • The authors mention that high-through put sequencing was used to identify the variant. However, electropherogram (Sanger’s sequencing) is given. This is leading to confusion. In case they have validated the variant that was identified in the high through put

sequencing by Sanger's sequencing they need to mention this. Was high through put sequencing done in all the family members or only the identified variant was screened?