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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 71782

Title: Targeted next-generation sequencing identifies a novel nonsense mutation in ANK1 for hereditary spherocytosis: a case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03353662
Position: Peer Reviewer
Academic degree: PhD
Professional title: Senior Researcher
Reviewer's Country/Territory: United Kingdom
Author's Country/Territory: China

Manuscript submission date: 2021-09-22

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-09-30 21:02

Reviewer performed review: 2021-10-10 13:03

Review time: 9 Days and 16 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Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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 [Y] Grade B: Minor language polishing [ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection
Conclusion	[ ] Accept (High priority) [ ] Accept (General priority) [Y] Minor revision [ ] Major revision [ ] Rejection
Re-review	[ ] Yes [Y] No

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Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The following to be reviewed 1. p3 line 61 hematopathy p77 unsaturatediron. Please review terminology and spelling More details on the instrumnetation used for NGS

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Reviewer's code: 01221812
Position: Peer Reviewer
Academic degree: MPhil, PhD
Professional title: Academic Research, Doctor, Research Scientist, Teacher
Reviewer's Country/Territory: Pakistan
Author's Country/Territory: China

Manuscript submission date: 2021-09-22

Reviewer chosen by: Xin Liu (Online Science Editor)

Reviewer accepted review: 2021-12-17 10:20

Reviewer performed review: 2021-12-24 17:13

Review time: 7 Days and 6 Hours

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish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

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Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

Targeted next-generation sequencing identifies a novel nonsense mutation in ANK1 for hereditary spherocytosis: a case report and a brief review of literature Fu et al. recruit an infant with hereditary spherocytosis (HS) which is characterized by anemia, jaundice, splenomegaly and cholelithiasis. The authors carried out targeted next-generation sequencing and identified a novel nonsense mutation in ANK1. This is an interesting study, however, the manuscript would benefit from the following changes/corrections: 1. Please mention OMIM number with each malformation and gene reported in the text. 2. Please mention the prevalence estimate of Hereditary spherocytosis in the global populations and the study populations. 3. Unclear statement, "a suspicious positive family history, and then finally confirmed by genetic sequencing.". Please elaborate the statement. What is the phenotypic presentation in the father of the index person. 4. The protocol of next-generation sequencing is not described. Please give detail of library preparation, controls, cut-off values, assembly used, etc. 5. The variant filtration strategy has not been given. 6. It is not clear who many rare variants were left in the final filtration scheme. 7. Please give the allele frequency of the variant as reported in public databases like 1000G and ExAC. 8. Discussion part is weak and redundent. Second paragraph of Disucussion may be shifted to Introduction. 9. Strengths and limitations of the study are not given.

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Peer-review model: Single blind

Reviewer's code: 00069130
Position: Editorial Board
Academic degree: BM BCh, PhD
Professional title: Academic Fellow, Assistant Professor
Reviewer's Country/Territory: India
Author's Country/Territory: China

Manuscript submission date: 2021-09-22

Reviewer chosen by: Xin Liu (Online Science Editor)

Reviewer accepted review: 2021-12-21 06:25

Reviewer performed review: 2021-12-27 03:45

Review time: 5 Days and 21 Hours

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

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Peer-reviewer statements	Peer-Review: [Y] Anonymous [ ] Onymous Conflicts-of-Interest: [ ] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The manuscript is well written ad may be accepted for publication. The authors may include in the discussion a section on the mutation-nonsense mutation (exon23:c.G2467T:p.E823X) of ANK1. This would help the general readers about the location, impact and consequences of the mutation. Unsaturated iron on page 77.