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**Telephone:** +1-925-399-1568
**E-mail:** bpgoffice@wjgnet.com
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# PEER-REVIEW REPORT

**Name of journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 86803

**Title:** Case report：Hematopoietic stem cell transplantation of aplastic anemia by relative with several mutations but normal telomere length

**Provenance and peer review**: Unsolicited Manuscript; Externally peer reviewed

**Peer-review model:** Single blind

**Reviewer's code:** 00415501
**Position:** Peer Reviewer
**Academic degree:** MD
**Professional title:** Professor
**Reviewer's Country/Territory:** Taiwan
**Author's Country/Territory:** China

**Manuscript submission date:** 2023-08-06

**Reviewer chosen by:** AI Technique

**Reviewer accepted review:** 2023-08-07 13:34

**Reviewer performed review:** 2023-08-11 08:44

**Review time:** 3 Days and 19 Hours

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|---|---|
| **Scientific quality** | [ ] Grade A: Excellent [ ] Grade B: Very good [Y] Grade C: Good<br>[ ] Grade D: Fair [ ] Grade E: Do not publish |
| **Novelty of this manuscript** | [ ] Grade A: Excellent [ ] Grade B: Good [Y] Grade C: Fair<br>[ ] Grade D: No novelty |
| **Creativity or innovation of this manuscript** | [ ] Grade A: Excellent [ ] Grade B: Good [Y] Grade C: Fair<br>[ ] Grade D: No creativity or innovation |

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|---|---|
| **Scientific significance of the conclusion in this manuscript** | [ ] Grade A: Excellent [ ] Grade B: Good [ Y] Grade C: Fair [ ] Grade D: No scientific significance |
| **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 |
| **Peer-reviewer statements** | Peer-Review: [ Y] Anonymous [ ] Onymous |
| | Conflicts-of-Interest: [ ] Yes [ Y] No |

## SPECIFIC COMMENTS TO AUTHORS

The authors attempted treatment of a patient a 43-year-old male patient with severe AA-carrying BRIP1, TINF2, and TCIRG1 mutations. Screening of the family pedigree revealed the same TINF2 mutation in his mother and older brother, and his older brother also carried the BRIP1 variant but with normal telomere length and hematopoietic function. Whilst the manuscript had some innovative and useful ideas, this manuscript is not suitable for publication in its current status. This case report had some limitations that should be highlighted. 1. Several recent studies showed heterozygous TINF2 mutation in 1–5% of patients with acquired aplastic anemia (Walne et al, 2008; Du et al, 2009). The subjects of these studies were Caucasian, Black, and Hispanic. Analysis of the TINF2 gene among adult Asian populations of AA and myelodysplastic syndrome (MDS), to the best of our knowledge, had never been done.

2. Mutations in the TINF2 gene have been associated with certain conditions related to telomere dysfunction, such as dyskeratosis congenita (DC) and other disorders that fall under the spectrum of telomere biology disorders. Whether a TINF2 mutation should be considered clinically important depends on the specific variant and its impact on the

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protein's function. Some TINF2 mutations may cause significant disruption to telomere maintenance, leading to severe clinical phenotypes like DC. Other variants might have milder effects and may be associated with less severe or asymptomatic presentations.