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# PEER-REVIEW REPORT

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

**Manuscript NO:** 62415

**Title:** Geleophysic dysplasia caused by a mutation in FBN1: a case report and review of the literature

**Reviewer's code:** 03815241
**Position:** Peer Reviewer
**Academic degree:** MD, PhD
**Professional title:** Associate Professor
**Reviewer's Country/Territory:** Greece
**Author's Country/Territory:** China

**Manuscript submission date:** 2021-02-23
**Reviewer chosen by:** AI Technique
**Reviewer accepted review:** 2021-04-22 19:14
**Reviewer performed review:** 2021-04-23 06:55
**Review time:** 11 Hours

| | |
|---|---|
| **Scientific quality** | [ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good<br>[ ] Grade D: Fair [ ] Grade E: Do not publish |
| **Language quality** | [ ] Grade A: Priority publishing [Y] Grade B: Minor language polishing<br>[ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection |
| **Conclusion** | [Y] Accept (High priority) [ ] Accept (General priority)<br>[ ] Minor revision [ ] Major revision [ ] Rejection |
| **Re-review** | [Y] Yes [ ] No |
| **Peer-reviewer statements** | Peer-Review: [Y] Anonymous [ ] Onymous<br>Conflicts-of-Interest: [ ] Yes [Y] No |

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**SPECIFIC COMMENTS TO AUTHORS**

Dear Authors, You have just described a very rare genetic anomaly. The description is quite clear and the ivestigation as well.However, I am wondering if the relatives of this child have been investiged as well.

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# PEER-REVIEW REPORT

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

**Manuscript NO:** 62415

**Title:** Geleophysic dysplasia caused by a mutation in FBN1: a case report and review of the literature

**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:** 2021-02-23
**Reviewer chosen by:** AI Technique
**Reviewer accepted review:** 2021-04-16 04:30
**Reviewer performed review:** 2021-05-06 07:23
**Review time:** 20 Days and 2 Hours

| | |
|---|---|
| **Scientific quality** | [ ] Grade A: Excellent [ ] Grade B: Very good [ Y] Grade C: Good<br>[ ] Grade D: Fair [ ] Grade E: Do not publish |
| **Language quality** | [ ] Grade A: Priority publishing [ Y] Grade B: Minor language polishing<br>[ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection |
| **Conclusion** | [ ] Accept (High priority) [ ] Accept (General priority)<br>[ Y] Minor revision [ ] Major revision [ ] Rejection |
| **Re-review** | [ ] Yes [ Y] No |
| **Peer-reviewer statements** | Peer-Review: [ Y] Anonymous [ ] Onymous<br>Conflicts-of-Interest: [ ] Yes [ Y] No |

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**SPECIFIC COMMENTS TO AUTHORS**

The authors report on a 9-year-old girl with Geleophysic dysplasia with the c.5243G>T (p.C1748F) mutation in FBN1 (fibrillin 1). This is an interesting study. However, the manuscript would benefit from the following changes/amendments: 1. Please mention OMIM number with each malformation described in the text. 2. Since it is a case report of Geleophysic dysplasia (GD) with mutation in FBN1 gene, it is not prudent to mention acromicric dysplasia (AD) and Weill-Marchesani syndrome (WMS) in the introduction and Abstract. Please omit them from the Introduction and Abstract. These can be mentioned in the Discussion as differential diagnosis. 3. It would be worthwhile to report the origin, basic demographics and ethnicity of the subject. 4. Please mention the parental parameters/symptoms and consanguinity. 5. Table 1. There are many typos in the table. Almost all the terms used in first column are problematic. 6. It is not clear who is Paitent 1 and Patient 2 in Table 1. 7. It is not clear what is the relationship of respiratory tract infections (RRTIs) with GD. The authors may like to put light on this. 8. Unfortunately the mutation detected in this patient is already reported (see Wang Y, Zhang H, Ye J, Han L, Gu X. Three novel mutations of the FBN1 gene in Chinese children with acromelic dysplasia. J Hum Genet. 2014 Oct;59(10):563-7. doi: 10.1038/jhg.2014.73. Epub 2014 Aug 21. PMID: 25142510.). The authors need to explicitly mention this in the text.