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# PEER-REVIEW REPORT

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

**Manuscript NO:** 70809

**Title:** A Case of Autosomal Dominant Osteopetrosis Type II Mutations in the CLCN7 Gene in a Patient with Osteopetrosis and Literature Review

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

**Peer-review model:** Single blind

**Reviewer's code:** 05910223
**Position:** Peer Reviewer
**Academic degree:** MD

**Professional title:** Doctor

**Reviewer's Country/Territory:** Japan

**Author's Country/Territory:** China

**Manuscript submission date:** 2021-09-15

**Reviewer chosen by:** AI Technique

**Reviewer accepted review:** 2021-09-18 04:28

**Reviewer performed review:** 2021-09-19 03:31

**Review time:** 23 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[ ] Grade D: Fair [ ] Grade E: Do not publish |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** | [Y] Yes [ ] No |

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|---|---|
| **Peer-reviewer statements** | Peer-Review: [ Y] Anonymous [ ] Onymous<br>Conflicts-of-Interest: [ ] Yes [ Y] No |

**SPECIFIC COMMENTS TO AUTHORS**

Thank you for having an opportunity to review this case report by Dr. Song, et al. They reported a rare case of autosomal-dominant osteopetrosis with Type II mutations in the CLCN7 gene. The authors emphasized the rarity of de novo missense mutation in the CLCN7 gene to further understand this rare disease with the literature review. Although their work is valuable, there are several major and minor points which should be addressed for the acceptance. Major 1. In Table 1, if possible, the authors should summarize and provide the data about main symptoms, treatments and prognosis of the osteopetrosis patients with CLCN7 mutation in previous reports. The additional data can be beneficial for early diagnosis and management of autosomal-dominant osteopetrosis with CLCN7 mutations. 2. In Background of Abstract section, the authors should mention briefly about clinical unsolved problems related to this rare disease and the reason why they want to report this case, which will be linked to the proof of clinical value of this case report. Minor 1. In Table 1, the authors should fill in the blank in the line of reference #11. 2. In Table 1, "united kingdo" is typo in reference #15. 3. The authors should refer to the acquisition of informed consent from the patients and the family in the manuscript. 4. In Discussion section, page 8, the authors should provide the percentage of gender ratio in the line where they referred to it. 5. In Discussion section, page 9, next-generation sequencing should be replaced to "NGS" after abbreviated. 6. If possible, the imaging results at the 2-year follow-up should be presented in Figure 1. 7. In Supplementary Table 1, the legends of Hb and ALT should be provided in the footnote.

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

**Peer-review model:** Single blind

**Reviewer's code:** 03593092
**Position:** Editorial Board
**Academic degree:** MD, PhD

**Professional title:** Associate Professor, Chief Physician, Neurosurgeon

**Reviewer's Country/Territory:** Taiwan

**Author's Country/Territory:** China

**Manuscript submission date:** 2021-09-15

**Reviewer chosen by:** AI Technique

**Reviewer accepted review:** 2021-09-17 18:23

**Reviewer performed review:** 2021-09-27 17:56

**Review time:** 9 Days and 23 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>[ ] Minor revision [Y] Major revision [ ] Rejection |
| **Re-review** | [ ] Yes [Y] No |

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|---|---|
| **Peer-reviewer statements** | Peer-Review: [ Y] Anonymous [ ] Onymous<br>Conflicts-of-Interest: [ ] Yes [ Y] No |

**SPECIFIC COMMENTS TO AUTHORS**

This case report presented a 5-year-old boy diagnosed with autosomal dominant osteopetrosis type II (ADO II), who had a de novo heterozygous missense mutation c.746C>T (p. P249L) in the CLCN7 gene as revealed by whole-exome sequencing. However, authors stated in the text that "To date, nearly forty mutations in CLCN7 have been identified linked to ADO II (page 8)," and "mutation c.746C>T (p.P249L) of the CLCN7 gene (NM_001287.5) has been previously described in patients with ADO II (References # 5 and 7) (page 6), so identification of this missense mutation c.746C>T in the CLCN7 gene in patient with ADO II is not a novel finding. In contrast, de novo mutation is the novelty of this case report authors should emphasize, thereby highlighting the fact that this mutation c.746C>T may or may not be inherited. The take-home message of this case report appears to be that "Even without the family history of ADO II, the possibility of de novo mutation in the CLCN7 gene and subsequent ADO II cannot be ruled out. In addition to the conventional neurological, imaging, and biochemical examinations, whole-exome sequencing is critical for final diagnosis." In addition, please consider the following suggestions to strengthen the present case report for readers. 1. As mentioned in the Core tip: "CLCN7 mutation can be due to de novo variants or due to inherited variants." The first statement in the Discussion section, "Osteopetrosis is an inherited disorder with symptoms including failure of osteoclasts and impaired bone resorption (page 7)" will be better to be amended to emphasize the possibility of de novo mutation in the first statement. 2.

The de novo CLCN7 mutation has been report, although mutation sites are different. The possibility of de novo CLCN7 mutation should be briefly discussed with the

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following citation. Nicoli ER, Weston MR, Hackbarth M, Becerril A, Larson A, Zein WM, Baker PR 2nd, Burke JD, Dorward H, Davids M, Huang Y, Adams DR, Zerfas PM, Chen D, Markello TC, Toro C, Wood T, Elliott G, Vu M; Undiagnosed Diseases Network, Zheng W, Garrett LJ, Tifft CJ, Gahl WA, Day-Salvatore DL, Mindell JA, Malicdan MCV. Lysosomal Storage and Albinism Due to Effects of a De Novo CLCN7 Variant on Lysosomal Acidification. Am J Hum Genet. 2019 Jun 6;104(6):1127-1138. doi: 10.1016/j.ajhg.2019.04.008. Epub 2019 May 30. PMID: 31155284; PMCID: PMC6562152. 3.

To stay focus, it is not relevant to mention "infantile malignant autosomal recessive osteopetrosis (ARO) and intermediate ARO (IARO) [1, 10] (page 7)" in this case report. 4.

(page 8) It should read as "…there have been more reports of CLCN7 mutations causing cases of ADO II in the Asian population than in Western countries over the last decades." 5. (page 9) "Biochemical markers have been considered for the diagnosis of osteopetroses, such as elevated creatine kinase MB isoenzyme (CK-MB), serum LDH and AST [27, 28]." So, the extent to which the serum levels of creatine kinase, LDH and AST of this boy facilitates the final diagnosis of ADO II should be illustrated in the Results section. 6. Table I summarized the findings from the literature review. However, some drawbacks were observed as follows: a) The starting time point of your literature review is unclear in the text. b) Most importantly, in-table citations do no match their references list. c) Why only case report studies were included in Table 1? If patients' demographic and clinical characteristics were clearly presented in the population-based studies, those patients should be included in Table 1 for comparison. d) Table 1 should be updated for comprehensive comparison and correct citation.