

PEER-REVIEW REPORT

Name of journal: World Journal of Clinical Cases

Manuscript NO: 81694

Title: Clinical and genetic diagnosis of autosomal dominant osteopetrosis type II in a

Chinese family: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06307616

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: China

Author's Country/Territory: China

Manuscript submission date: 2022-11-23

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-11-25 01:32

Reviewer performed review: 2022-11-25 01:40

Review time: 1 Hour

| Scientific quality | [] Grade A: Excellent [] Grade B: Very good [Y] Grade C: Good [] Grade D: Fair [] Grade E: Do not publish |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) [Y] Accept (General priority) [] Minor revision [] Major revision [] Rejection |



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

SPECIFIC COMMENTS TO AUTHORS

This study suggested that there is no straightforward genotype-phenotype correlation between CLCN7 variants and osteopetrosis. This is a rare report of an ADO-II patient carrying both TCIRG1 and CLCN7 mutations, which also highlights that genetic analysis is be beneficial for the classification and therapeutic decision of osteopetrosis.



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Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06411308

Position: Peer Reviewer

Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: United States

Author's Country/Territory: China

Manuscript submission date: 2022-11-23

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-11-30 09:22

Reviewer performed review: 2022-12-03 14:07

Review time: 3 Days and 4 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| Scientific quality | [] Grade A: Excellent [] Grade B: Very good [Y] Grade C: Good [] Grade D: Fair [] Grade E: Do not publish |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 |
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SPECIFIC COMMENTS TO AUTHORS

1. Authors should describe the background in detail why the patient has been "accidentally" (page 1) or "mistakenly" (page 2) diagnosed with ADO-II. 2. It would be helpful if authors would provide the histologic results of pathological study to evaluate the cortical bone sclerosis and some thickened bone trabeculae. 3. It would be great if authors could suggest differential diagnosis except for ADO-II. 4. While this case report described long about general information of ARO and ADO for osteopetrosis, relatively in the case, rationale arriving to the final diagnosis seem not to be well-described based on the detailed results of diagnostic work-up.