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# PEER-REVIEW REPORT

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

**Manuscript NO:** 66269

**Title:** Autosomal dominant tubulointerstitial kidney disease with a novel heterozygous missense mutation in the uromodulin gene: A case report

**Reviewer's code:** 03714155
**Position:** Peer Reviewer
**Academic degree:** DPhil
**Professional title:** Doctor
**Reviewer's Country/Territory:** Japan
**Author's Country/Territory:** China

**Manuscript submission date:** 2021-03-29
**Reviewer chosen by:** AI Technique
**Reviewer accepted review:** 2021-06-26 07:39
**Reviewer performed review:** 2021-07-04 11:39
**Review time:** 8 Days and 4 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[ ] Grade D: Fair [ ] Grade E: Do not publish |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** | [ ] Accept (High priority) [Y] 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**

This report represents an effort to accomplish this, as these data will most likely prove to be a unique resource well into the future. I think this paper should be published.

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# PEER-REVIEW REPORT

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

**Manuscript NO:** 66269

**Title:** Autosomal dominant tubulointerstitial kidney disease with a novel heterozygous missense mutation in the uromodulin gene: A case report

**Reviewer's code:** 05758726
**Position:** Peer Reviewer
**Academic degree:** MD
**Professional title:** Doctor
**Reviewer's Country/Territory:** Japan
**Author's Country/Territory:** China

**Manuscript submission date:** 2021-03-29
**Reviewer chosen by:** Ze-Mao Gong
**Reviewer accepted review:** 2021-07-22 03:52
**Reviewer performed review:** 2021-07-22 07:52
**Review time:** 3 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[ ] Grade D: Fair [ ] Grade E: Do not publish |
| **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 reported a family case with a new UMOD mutation. 1. About "DNA analyses", the authors should provide more minute explanation of its method. 2. Description concerning genetic counseling is totally lacking. The authors should explain this aspect of issue within genetic examination.

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# PEER-REVIEW REPORT

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

**Manuscript NO:** 66269

**Title:** Autosomal dominant tubulointerstitial kidney disease with a novel heterozygous missense mutation in the uromodulin gene: A case report

**Reviewer's code:** 05278701
**Position:** Editorial Board
**Academic degree:** MD
**Professional title:** Professor, Staff Physician
**Reviewer's Country/Territory:** Brazil
**Author's Country/Territory:** China

**Manuscript submission date:** 2021-03-29
**Reviewer chosen by:** Ze-Mao Gong
**Reviewer accepted review:** 2021-07-23 00:25
**Reviewer performed review:** 2021-07-23 12:50
**Review time:** 12 Hours

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|---|---|
| **Scientific quality** | [ Y] Grade A: Excellent [ ] Grade B: Very good [ ] Grade C: Good<br>[ ] Grade D: Fair [ ] Grade E: Do not publish |
| **Language quality** | [ Y] Grade A: Priority publishing [ ] Grade B: Minor language polishing<br>[ ] Grade C: A great deal of language polishing [ ] Grade D: Rejection |
| **Conclusion** | [ ] Accept (High priority) [ Y] Accept (General priority)<br>[ ] 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**

I would like to congratulate the article written by Santao Ou et al. The article is well written, presenting genetic and pathophysiological aspects of a unique condition, and is very well documented. We recommend the publication of the paper

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# PEER-REVIEW REPORT

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

**Manuscript NO:** 66269

**Title:** Autosomal dominant tubulointerstitial kidney disease with a novel heterozygous missense mutation in the uromodulin gene: A case report

**Reviewer's code:** 05223442

**Position:** Editorial Board

**Academic degree:** FICS, MD, MSc

**Professional title:** Academic Fellow, Lecturer, Senior Researcher, Surgeon

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

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

**Manuscript submission date:** 2021-03-29

**Reviewer chosen by:** Ze-Mao Gong

**Reviewer accepted review:** 2021-07-28 18:27

**Reviewer performed review:** 2021-07-29 19:00

**Review time:** 1 Day

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

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**SPECIFIC COMMENTS TO AUTHORS**

It is still possible to achieve a renal biospsy in an 8.3 cm kidney which is the 3 cm below the average lenth. Findings from renal biopsy in patients with ADTKD-UMOD could have shown aggregates of uromodulin in the enoplasmic reticulum and disruption of the epithelial cells of the thick ascending limb (TAL) of the loop of Henle. After considering ADTKD as a probable diagnosis, urine uromudolin would have been helpful while awaiting genetic sampling.