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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 73811

Title: Novel mutation in the SAL gene in a four-generation Chinese family with uraemia: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03761355
Position: Editorial Board
Academic degree: FASN, FRCP, MD
Professional title: Professor
Reviewer's Country/Territory: United States
Author's Country/Territory: China

Manuscript submission date: 2021-12-21
Reviewer chosen by: Xin Liu (Online Science Editor)
Reviewer accepted review: 2022-03-06 03:03
Reviewer performed review: 2022-03-06 22:27
Review time: 19 Hours

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[Y] Grade A: Priority publishing [ ] 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

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

SPECIFIC COMMENTS TO AUTHORS

In this report. the authors present a patient with kidney failure and morphological abnormalities with evidence for mutation in SALL1 gene confirming a diagnosis of Townes-Brocks Syndrome (TBS) first described in 1972. There was 4 generation involvement. This case highlights the importance of clinicians to think "out of the box" in order to make diagnosis of unusual clinical entities, that may have impact on other family members. The article is written well. I would recommend the following points to increase the usefulness for the readers: 1. It will be useful to add any data available on outcomes of kidney transplantation in patients with TBS. 2.Rarely a mutation in the DACT1 gene can also be causative, this should be mentioned. 3.The group of related disorders with physical features similar to TBS (including oculo-auriculo-vertebral spectrum disorders, Okihiro syndrome, bracheo-oto renal syndrome, VACTERL association and STAR syndrome) should mentioned in discussion.

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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 73811

Title: Novel mutation in the SAL Y gene in a four-generation Chinese family with uraemia: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 00503228
Position: Editorial Board
Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Iran

Author's Country/Territory: China

Manuscript submission date: 2021-12-21

Reviewer chosen by: Xin Liu (Online Science Editor)

Reviewer accepted review: 2022-03-08 03:06

Reviewer performed review: 2022-03-08 06:14

Review time: 3 Hours

Scientific quality	[Y] Grade A: Excellent [ ] Grade B: Very good [ ] 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	[Y] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [ ] Major revision [ ] Rejection
Re-review	[Y] Yes [ ] No

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

SPECIFIC COMMENTS TO AUTHORS

An extremely interesting case and good presentation; there are some more delicate issues that deserves consideration: 1. The type of kidney malformation has not been well defined. In the TBS, the kidneys could present with a wide spectrum of malformations including multicystic kidneys & hypoplastic kidneys. A CT scan could provide a proper illustration. More detailed data on the cause of ear problems would also be most interesting. 2. Heart defects including TF and ventricular wall defects are amongst the presenting features of this syndrome. An echocardiography could easily diagnose the potential cardiac defects in the proband and the other affected individuals in the pedigree. 3. Even more importantly, we need to know about the broband's daughter's defects, most especially her renal system. She already has proteinuria, and no data has been provided how she is under surveillance or gets preventive measures and treatment for avoiding (or at least postponing) the renal failure. Has she been prescribed, for example, captopril or ARBs (which are the standard Rx for patients presenting with proteinuria?) Or if the literature provides any information for preventive strategies in this context? Now I think that the girl's fate is the most important issue to focus on. And you may come up with invaluable data on the potential preventive measures for the kidney failure in patients diagnosed early with this syndrome.

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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 73811

Title: Novel mutation in the SAL gene in a four-generation Chinese family with uraemia: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 02726701
Position: Editorial Board
Academic degree: MD
Professional title: Professor
Reviewer's Country/Territory: Chile
Author's Country/Territory: China

Manuscript submission date: 2021-12-21

Reviewer chosen by: Xin Liu (Online Science Editor)

Reviewer accepted review: 2022-03-08 12:06

Reviewer performed review: 2022-03-08 15:57

Review time: 3 Hours

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] Grade C: Good [ ] Grade D: Fair [ ] Grade E: Do not publish
Language quality	[Y] Grade A: Priority publishing [ ] 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

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

SPECIFIC COMMENTS TO AUTHORS

Comments on Novel mutation in the SALL1 gene in a four-generation Chinese family with uremia: A case report The manuscript is very well written. It describes a very rare genetic, apparently, monogenic disease, in a clear and concise way. I suggest: 1. To improve the description of the photographs in order to highlight what are the abnormalities. 2. To intend to teach the readers that even in a chronic ambulatory dialysis facility it is possible to find rare diseases. For example: Ask patients for family history of kidney diseases, see if the patient has ocular or hear pathologies, see if there is poliglobulia or abdominal masses or has family history of cerebral aneurisms, verify that if there is neuropathy or cardiopathy they do not obey to uremia or hypertension alone. Please, add a comment about the most convenient treatment for the patient´s kid who showed proteinuria.

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Title: Novel mutation in the SAL Y gene in a four-generation Chinese family with uraemia: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03815828
Position: Editorial Board
Academic degree: MD

Professional title: Full Professor, Senior Lecturer

Reviewer's Country/Territory: Turkey

Author's Country/Territory: China

Manuscript submission date: 2021-12-21

Reviewer chosen by: Xin Liu (Online Science Editor)

Reviewer accepted review: 2022-03-13 15:47

Reviewer performed review: 2022-03-13 15:56

Review time: 1 Hour

Scientific quality	[ ] Grade A: Excellent [Y] Grade B: Very good [ ] 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	[Y] Accept (High priority) [ ] Accept (General priority) [ ] Minor revision [ ] Major revision [ ] Rejection
Re-review	[ ] Yes [Y] No

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

SPECIFIC COMMENTS TO AUTHORS

The authors have reported a four-generation Chinese family in which four individuals had a novel SALL1 mutation and presented with uraemia or abnormal urine tests. Index patient was a 32-year-old man presented with end-stage renal disease with a 4-year history of dialysis. His father and paternal aunt both had a history of unexplained renal failure with haemodialysis, and his 10-year-old daughter presented with proteinuria. The patient had multiple congenital abnormalities, including bilateral overlapping toes, unilateral dysplastic external ears and sensorineural hearing loss. His family members also presented with similar defects. Genetic testing revealed that the proband carried a novel heterozygous shift mutation in SALL1_exon 2 (c.3437delG), and Sanger sequencing confirmed the same mutation in all affected family members. At the end of the study the authors concluded that a novel SALL1 exon 2 (c.3437delG) mutation and clinical syndrome with kidney disease, bilateral overlapping toes, unilateral dysplastic external ears and sensorineural hearing loss in a four-generation Chinese family. I think this case report is quite educational and interesting. It can open new horizons for clinicians. This is a well written manuscript.

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PEER-REVIEW REPORT

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Title: Novel mutation in the SAL gene in a four-generation Chinese family with uraemia: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 05040165
Position: Editorial Board
Academic degree: MD
Professional title: Associate Professor
Reviewer's Country/Territory: Thailand
Author's Country/Territory: China

Manuscript submission date: 2021-12-21

Reviewer chosen by: Xin Liu (Online Science Editor)

Reviewer accepted review: 2022-03-07 04:51

Reviewer performed review: 2022-03-16 04:20

Review time: 8 Days and 23 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 [ ] Grade B: Minor language polishing [Y] 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

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

SPECIFIC COMMENTS TO AUTHORS

The paper is reporting a novel mutation in the SALL1 gene in a four-generation Chinese family with uraemia: A case report. The topic is interest and despite the data are not adding much to the current data reported so far, it is well written. Her are my concerns:

1. Please provide data of important investigations for differential diagnosis and confirm cause of kidney disease/failure i.e., imaging, work up for KUB anomaly, tissue biopsy or specific serology, etc. explicit. 2. The references are outdated. More than 2/3 of the references are more than 10 years. Please add and summarize the results of recent studies in 2021. 3. Finally, since I am not a native English user, I did not check for grammatical errors thoroughly. This should be done by an appropriate language reviewer.