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PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 72053

Title: Novel mutations of the Alström syndrome 1 gene in an infant with dilated cardiomyopathy: a case report

Provenance and peer review: Unsolicited manuscript; externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03498422
Position: Peer Reviewer
Academic degree: MD

Professional title: Doctor

Reviewer's Country/Territory: Italy

Author's Country/Territory: China

Manuscript submission date: 2021-10-05

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-10-17 09:59

Reviewer performed review: 2021-10-21 18:05

Review time: 4 Days and 8 Hours

Scientific quality	[ ] Grade A: Excellent [ ] Grade B: Very good [ ] Grade C: Good [Y] Grade D: Fair [ ] Grade E: Do not publish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) [ ] Accept (General priority) [ ] Minor revision [Y] 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 Authors report a novel mutation of the ALMS1 gene causing DCM in a 1-month-old girl. The case is interesting, even though several points should be addressed. 1. Medical therapy included Angiotensin converting enzyme inhibitors, spironolactone, digoxin and diuretics. Overall, this therapy seems quite outdated compared to current guidelines on HF (sacubitril/valsartan, betablockers... are not mentioned). 2. In the manuscript "After standard anti-congestive heart failure treatment, TTE revealed acoustic enhancement in the left ventricular endocardium compared with at admission, which was suspected to have arisen from thickening of the ventricular endocardium as a result of deposition of subendocardial fibrous tissue layers during heart development[8]. Consequently, myocardial fibrosis may also play a role in AS." This concept shoud be removed or toned down, because subendocardial hyperintensity at cardiac ultrasound cannot be used as a surrogate for myocardial fibrosis, which should be demonstrated by the presence of late enhancement at cardiac magnetic resonance. 3. In the summary the terms "antiventricular remodeling treatment" and "acoustic enhancement in the left ventricular endocardium" are quite unusual and difficult to understand; I would strongly recommend to rephrase this sentence ("heart failure therapy" or "anti-remodelling therapy".... and "subendocardial hyperechogenicity"... respectively, even though this last concept should be removed/rephrased as indicated in point 2). 4. There are no biohumoral exams at follow-up (did NTproBNP decrese on medical therapy)? 5. There are no data about the arrhythmic burden; was a 24-holter ECG monitoring performed at baseline and/or at follow-up?

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Manuscript NO: 72053

Title: Novel mutations of the Alström syndrome 1 gene in an infant with dilated cardiomyopathy: a case report

Provenance and peer review: Unsolicited manuscript; externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 03908435
Position: Peer Reviewer
Academic degree: MBBS

Professional title: Consultant Physician-Scientist, Doctor

Reviewer's Country/Territory: India

Author's Country/Territory: China

Manuscript submission date: 2021-10-05

Reviewer chosen by: AI Technique

Reviewer accepted review: 2021-10-17 17:25

Reviewer performed review: 2021-10-28 18:42

Review time: 11 Days and 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	[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

An interesting addition to the existing knowledge on the genotype of Alström syndrome. I'd like to draw the attention of the authors to the following- 1. The authors' statement -// There have been no cases reported with a relationship between DCM and mutation of ALMS1.// is ambiguous. What do the authors try to convey? When cardiomyopathy is an important feature of Alström's, how do the authors say that there has been no case with ALMS1 mutation and DCM? 2. In the case presentation part, the sequence of findings that led to the suspicion of Alström's clinically should be described. This should come prior to the genetic testing part. 3. In discussion, try to add a few words about the differences between phenotypes (if any)when mutation is found in the hot-spot exons and the non-hotspot exons.