Reviewer’s report

Title: A very early diagnosis of Alström syndrome by next generation sequencing

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Reviewer's report:

This is a well summarised review, narrated in context of a clinical case. It identifies a novel mutation in ALMS1 gene and serves as a good review of the condition. The introduction is well written and summarizes this condition well in few words. The discussion subsequently dwells into the new variants identified and challenges in diagnosing alstrom syndrome.

I have some minor issue that authors may wish to consider

(1) From an ethics perspective, I note that parents signed a informed consent but considering that this a rare disease and the only case in slovakia, authors need to be aware that anything written in this review can easily be traced back to the patient.

(2) The lines 189 to 193, could help with rewriting to convey the message more clearly.

(3) Far too much of the discussion dwells on investigational medicinal products that are still unproven in terms of clinical benefit. Further trials are underway but clearly we are still in phase II of most studies. Why not spend some time discussing life style modification's and use of standard established treatments for individual organ dysfunction eg good diabetes control, managing hypertriglyceridemia, ACEi for LV dysfunction.

(4) As well outlined by the authors, eye symptoms on most occasions is what tends to be what draws attention to the diagnosis in Asltrom even in our experience. I wonder if authors would consider exploring in more details some of the diagnostic challenges related to the condition.

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No
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Unable to assess

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
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