Reviewer's report

Title: A donor splice site mutation in CISD2 generates multiple truncated, non-functional isoforms in Wolfram Syndrome type 2 patients

Version: 1 Date: 31 Oct 2017

Reviewer: Luciana Concetta Rigoli

Reviewer's report:
The manuscript MGTC-D-17-00149R1: "A donor splice site mutation in CISD2 generates multiple truncated, non-functional isoforms in Wolfram Syndrome type 2 patients" is well written and offers interesting insights for understanding the pathogenetic mechanisms of Wolfram 2 syndrome, a very rare and complex genetic disease. The experimental design has been well performed and the results are useful to clarify the role of the c.103+1G>A mutation in WS2.

Therefore I believe that the manuscript is suitable for the publication on BMC Medical Genetics.

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.

Yes

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.
Quality of written English

Please indicate the quality of language in the manuscript:

Acceptable

Declaration of competing interests

Please complete a declaration of competing interests, considering the following questions:

1. Have you in the past five years received reimbursements, fees, funding, or salary from an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

2. Do you hold any stocks or shares in an organisation that may in any way gain or lose financially from the publication of this manuscript, either now or in the future?

3. Do you hold or are you currently applying for any patents relating to the content of the manuscript?

4. Have you received reimbursements, fees, funding, or salary from an organization that holds or has applied for patents relating to the content of the manuscript?

5. Do you have any other financial competing interests?

6. Do you have any non-financial competing interests in relation to this paper?

If you can answer no to all of the above, write 'I declare that I have no competing interests' below. If your reply is yes to any, please give details below.

'I declare that I have no competing interests'

I agree to the open peer review policy of the journal. I understand that my name will be included on my report to the authors and, if the manuscript is accepted for publication, my named report including any attachments I upload will be posted on the website along with the authors' responses. I agree for my report to be made available under an Open Access Creative Commons CC-BY license (http://creativecommons.org/licenses/by/4.0/). I understand that any comments which I do not wish to be included in my named report can be included as confidential comments to the editors, which will not be published.

I agree to the open peer review policy of the journal