Reviewer’s report

Title: Xp11.22 Duplications in Four Unrelated Chinese Families: Delineating the Genotype-phenotype Relationship for HSD17B10 and FGD1

Version: 2 Date: 13 Mar 2020

Reviewer: Research Square Reviewer 1

Reviewer's report:

PEER REVIEWER ASSESSMENTS:

OBJECTIVE - Full research articles: is there a clear objective that addresses one or several testable research questions? (Brief or other article types: is there a clear objective?)
No - there are minor issues

DESIGN - Is the current approach (including controls and analysis protocols) appropriate for the objective?
No - there are minor issues

EXECUTION - Are the experiments and analyses performed with sufficient technical rigor to allow confidence in the results?
No - there are minor issues

STATISTICS - Is the use of statistics in the manuscript appropriate?
N/A - there are no statistics in this study

INTERPRETATION - Is the current interpretation/discussion of the results reasonable and not overstated?
No - there are minor issues

OVERALL MANUSCRIPT POTENTIAL - Has the author addressed your concerns sufficiently for you to now recommend the work as a technically sound contribution? If not, can further revisions be made to make the work technically sound?
Probably - with minor revisions

PEER REVIEWER COMMENTS:

GENERAL COMMENTS: paper is ok as far as they followed reviewers guidelines which they always did. However, they did not answer all comments (see below). Also as they did not state how they identified the 4 families it either may be due to studying patients with similar symptoms or due to screening many patients with different symptoms - this needs to be made clear in study design.

REQUESTED REVISIONS:
There are Study design problems. Also authors need to include - as requested by Reviewer 1 in Discussion some sentences about: "The authors should discuss whether some of the CNVs could be mediated by these polymorphisms." The same holds true for comment of Reviewer 1 for FDG1 and HUWE1 genes - here the authors cannot just answer this point in the answer to the reviewers
comments and include not all what they answered completely in the article; this must be added in discussion - and it seems that this was not really done yet completely.

In reviewing the previous reviewers' comments and reading the manuscript, my main concern is about being able to document the number of women who chose to receive treatment, which would allow the readers to interpret all prevalence data in a more informed context. I understand the reasoning for not including this information in the regression analysis, but could the authors include simple percentages of women who, after being referred to treatment, followed through with referral? Perhaps I have missed it, but I did not find this information easily available in the paper.

ADDITIONAL REQUESTS/SUGGESTIONS:

n.a.

**Are the methods appropriate and well described?**
If not, please specify what is required in your comments to the authors.

No

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

No

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

No

**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**
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:

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

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5. Do you have any other financial competing interests?

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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.

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