Reviewer's report

Title: Preimplantation genetic testing for a family with Usher syndrome through targeted sequencing and haplotype analysis

Version: 0 Date: 09 Feb 2019

Reviewer: Christine de Die-Smulders

Reviewer's report:

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This is a comprehensive description of one PGD case. The application of MPS in PGD is recently described by some other authors (Xiang 2015, Zhou 2018). Also other methods based on haplotyping of embryos are known and applied (Renwick 2010, Handyside 2010, 2015). In the introduction or discussion these papers have to be mentioned and also the differences, advantages and disadvantages of MPS based PGD compared to these methods.

In the introduction authors mention that PGD is possible by mutation analyses or linkage analysis. However, mutation analyses is usually combined with haplotyping to prevent misdiagnosis due to ADO.

Methods section is very long and contains a lot of technical details, authors can skip part of it, which will make the paper easier to read. More or less the same applies to the results section, that is many details and overlap with the methods section.

In discussion authors mention an increased risk on chromosomal anomalies after PGD, however this is not proven. Prenatal testing is merely meant for detection of a misdiagnosis after PGD as described earlier (for review see Wilton et al).

Usher syndrome is autosomal recessive disorder. In this family 3 generations are affected, which is really exceptional. Do the authors have an explanation for this? Please mention in the discussion.

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.
Yes

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

Yes

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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:

Needs some language corrections before being published

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