Reviewer’s report

Title: Genetic analysis of 62 Chinese families with Duchenne muscular dystrophy and strategies of prenatal diagnosis in a single center

Version: 0 Date: 31 May 2019

Reviewer: Éliane Chouery

Reviewer's report:

I have reviewed carefully the manuscript. My comments are the following:

1- The paper should be reviewed for the English by a native English speaker.

2- Authors should replace small mutation by point mutations

3- In the methods, abstract section: authors should state briefly the type pf NGS performed (Panel, WES,...) and the used technology.

4- In the results, abstract section: authors should add the mutations identified at the protein level. same section to replace "mothers showed the same mutations as the probands" by mothers were carriers for the same mutations as probands.

5- authors should in the abstract precise that introns of DMD gene were not sequenced.

6- in the methods section: NGS design (primers design), number of samples per run, NGS analysis should be more detailed. authors describe this section in general. They should add more details.

7-results, line 149-150: the sentence "of all the exons, exon 49 and 50 ..... a history of DMD" is not clear.

8- results line 164: why they screened for point mutations in the mothers by NGS not by fluorescent sequencing?

9- discussion: authors didn't explain why molecular diagnosis for these 62 families was not performed before this study? at least MLPA which was available since many years.

10- line 192-194: it could be more interesting to put some numbers when authors compare their results to other publications, instead of saying lower and higher than other studies.
11-line 216: to replace new mutation by de novo mutation.

12- line 223: to replace "and the remaining had no pathogenic mutation" by "the remaining were normal for the identified mutations in the families respectively"

13-did authors identified after studying the fetuses a family with germline mutation in mosaic state?

14- why they didn't include the introns in their study?

15- in the title authors mention "a strategy for prenatal diagnosis" , in all the paper authors didn't mention the needed time to perform the technique and the analysis.

16- authors should change all their conclusion.

17- it could be intereseting to put some clinical info for the patients: age, clinical signs, CPK value. And also it can be interesesting to comapre the severity of the disease in patients with large rearrangeemnts compared to patients with point mutations.

18- to replace in the tables the codon stop X by

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.

Not relevant to this manuscript
Quality of written English
Please indicate the quality of language in the manuscript:

Not suitable for publication unless extensively edited

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