Reviewer’s report

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

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Reviewer: Ricardo Fujita

Reviewer's report:

This manuscript describe the diagnosis of DMD in patients with dystrophinopathies in 62 families followed by the corroboration of the respective mutation in the carrier mothers and prenatal diagnosis in amniotic fluid, all performed in a four years period in a department of the Nanjin Medical University.

- The authors say that single exon deletion are the most common type of deletion, however it is known that this kind of mutation can often be an artifact of the MLPA process (mutation in the probe annealing region), since they have primers for each exon, this should be corroborated.

- There are samples that are negative for the test of DMD by MLPA and NGS. Since the authors used the "Ion AmpliSeq Inherited Disease Panel", have they checked other neuromuscular diseases in the panel?. If yes and still negative for the other diseases, it should be stated. If no, they should be checked.

- In Discussion in line 206 the statement "All the six novel nonsense mutations" is not true. There are four small deletions or insertions that led to premature stop codons and two nonsense mutations.

- In Table 2 is written "Nonsence" instead of "Nonsense". Anyway, this information are already included in table 1 and can be distinguished by using bold or other distinctive mark.

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.

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

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Please indicate the quality of language in the manuscript:

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