Reviewer’s report

Title: MLPA identification of dystrophin mutations and in silico evaluation of the predicted protein in dystrophinopathy cases from India

Version: 0 Date: 16 Jan 2017

Reviewer: Atsushi Asakura

Reviewer's report:

In this manuscript, the tested 415 diagnosed patients for dystrophin mutations by multiplex ligation dependent probe amplification (MLPA). Phenotype-genotype correlation was examined by PROVEAN, hydrophobicity and eDystrophin analysis. As a result, around 75% patients, deletions and duplications were identified 91.6% and 8.3%, respectively. Among the hot spot found in exon-45-52, exon 50 was the most frequently deleted. PROVEAN, hydrophobicity and eDystrophin analyses provided more detailed predicted dystrophin protein structures for mutations. However, the authors noticed that the results were not convincing on a case by case basis, indicating the limitation in the currently available tools for genetic analysis.

Overall this manuscript has high quality data and important for clinical DMD filed.

Minor issues:

1. It is nice to provide more information about PROVEAN, hydrophobicity and eDystrophin analyses.

2. In Fig 3, panels J, K, L and V show different background from other panels. It is better that the authors would either recapture images or retry immunostaining.

3. In Fig. 5, numbers for year are overlapped with the markers.

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

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I am able to assess the statistics

Quality of written English
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