Reviewer's report

Title: Clinical and molecular characterization of a cohort of patients with novel nucleotide alterations of the Dystrophin gene detected by direct sequencing

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Reviewer: Madhuri Hegde

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the DMD gene by sequencing the coding region of the gene. The authors describe several interesting cases detected by cDNA sequencing. It is important to publish the case reports and mutations in the DMD gene as we accumulate more data. Though the manuscript is generally well written the authors need to address several points in the manuscript

1. The overall conclusion is not appropriate. The authors say that only a small number if mutations have been described in the DMD gene. This is not an accurate statement. The Leiden database and the HGMD mutations has a full catalogue of DMD point mutations. I am assuming that the authors want to say that a only a small number of pre-RNA maturation defects or deep intronic mutations have been described. Only one ref (7) has been cited.

2. In methods under transcript analysis manufacturer of Eurozol not mentioned

3. Prediction analysis (bioinformatics) using ESE finder etc needs to be added to methods.

4. primers for transcript analysis??? Were published primers used?

5. Table 1: Change " kind of mutation" to "Mutation"

6. A comprehensive map of mutations identified along with DMD gene will be helpful

7. Some grammatical errors need to be corrected

Level of interest: An article of outstanding merit and interest in its field

Quality of written English: Needs some language corrections before being published

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests