Reviewer’s report

Title: Insertion of an extra copy of Xq22.2 into 1p36 results in functional duplication of the PLP1 gene in a girl with classical Pelizaeus-Merzbacher disease

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Reviewer: Nicole Wolf

Reviewer’s report:

The authors describe a girl with Pelizaeus-Merzbacher disease (PMD) due to the insertion of an extra copy of PLP1 on chromosome 1p36. The article is interesting and worthwhile to publish. I would, however, suggest some minor essential revisions.

1. The abstract is, regarding quality of language, not optimal. There are several grammatical mistakes, and the use of present and past tense is not consequent. There are language mistakes throughout the article, but not as prominent as in the abstract.

2. OL is used for oligodendrocytes, but the authors should not abbreviate this word.

3. The insertion of PLP1 on chromosome 1 would have missed had the authors not performed FISH. Do they recommend using FISH to confirm "ectopic" PLP1 insertion especially in girls?

4. The readers of BMC Medical Genetics might not be accustomed to evaluate MR images. Perhaps the authors could provide the respective MR images of an age-matched healthy child?

3.

Level of interest: An article of importance 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.