Reviewer’s report

Title: Three novel mutations in the lactase gene (LCT) underlying congenital lactase deficiency (CLD)

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Reviewer: Songbin Fu

Reviewer’s report:

In the manuscript of “Three novel mutations in the lactase gene (LCT) underlying congenital lactase deficiency (CLD)”, the authors amplified and re-sequenced the coding region of LCT in four CLD patients. By sequencing they identified three novel mutations and predicted the function of these mutations by the bioinformatics programs. It is an interesting paper and the main suggestions are listed below:

Major compulsory revisions:

1. By sequencing, the authors found three novel mutations in four patients. In the section of results, the authors pointed out that these mutations were not found in the control subjects. However this was not listed in the methods, the authors should introduce it in the methods.

2. Although the authors predicted the function of these mutations by bioinformatics programs, it remained quite challenging to understand precisely some statements in this paper. In addition, it is questionable if this functional prediction was sufficiently compelling and novel. Importantly, I would strongly suggest that the authors make a considerable effort to clean up the language, such as the first paragraph in page 13. The authors should use shorter sentence to express themselves effectively in the whole paper.

3. The mutations occurred in the upstream of LCT in the patients of the adult-type hypolactasia mostly, I think the author should explain why they did not sequence the upstream of LCT in the CLD patients.

Minor essential revisions

The table should be three-line formatted and the figure should be more attractive. There are some errors in grammar usage throughout the manuscript. I urge the authors to have it edited in detail by a native English speaker if at all possible.

Discretionary revisions

There should be more functional analysis about the three mutations in addition to the prediction by the program.

The manuscript could be considered for publication after revision.

Level of interest: An article whose findings are important to those with closely
related research interests

**Quality of written English:** Not suitable for publication unless extensively edited

**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