Reviewer's report

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

Version: 3 Date: 25 November 2008

Reviewer: Dallas M Swallow

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Minor Essential Revisions
I am still not comfortable with the sentence about researching the difference between the effect of deficiency in children and adults.

It seems plain to me that to be deficient in lactase as a baby will have much more severe consequences than for an adult. Milk is so much more important in the diet. There also will have been less time for the gut bacterial flora to adapt to cope with lactose--as occurs in many adults. It would be preferable to turn this sentence around into such a speculation.

The comment of reviewer 2 can be addressed as follows:

The English is still not perfect so suggest the following version:

"Deletion of five bases c.1692-1696delAGTGG in exon 6 leading to frameshift mutation V565fsX567 (Table 1) was found in Finnish patient heterozygous for the founder mutation Y1390X [10]. The deletion was located in a conserved region of the LCT gene (Figure 1) and (see below). Unfortunately parental DNA was not available for the analysis. The deletion was not found in 98 anonymous Finnish blood donors.""

Also the authors do not make it clear that a truncated protein is predicted--because I assume that this will be the consequence of the change of reading frame. This should be stated.

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

Quality of written English: Acceptable

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