Reviewer’s report

Title: A novel mutation within the lactase gene (LCT): the first case report of congenital lactase deficiency diagnosed in Central Europe

Version: 1 Date: 16 June 2015

Reviewer: Mamata Sivagnanam

Reviewer’s report:

Minor Essential Revisions:

1. This manuscript would be enhanced by putting this mutation in the context of other mutations. For example- are other mutations deletions, missense or nonsense mutations? Is there any other deletion mutation leading to a premature stop?

2. Please include Family history (other children affected?). Depending on the nature of the consanguineous relation in the family/inclusion of a pedigree would be of interest.

3. The authors state that biopsy testing should replaced by genetic testing. This is a strong statement and should be qualified. Genetic testing AND clinical improvement (including growth improvement) must take place for endoscopy to be bypassed.

Discretionary Revisions

1. Line 134 gastroscopy should be esophagogastroduodenoscopy or at least gastroduodenoscopy.

2. Line 146 add gastroenterologists

3. Line 113 Please define colics.

4. In addition to the genetics leading to easier diagnosis, it also aids in the ability to guide the family with genetic counseling and for family planning. Addition of these sentiments would improve the discussion.

Level of interest: An article of limited interest

Quality of written English: Acceptable

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 interest