Reviewer’s report

**Title:** Novel mutations of the GALT gene and mutation spectrum in the Korean population with decreased galactose-1-phosphate uridyltransferase activity

**Version:** 1 **Date:** 27 June 2014

**Reviewer:** Anna Marabotti

**Reviewer’s report:**

Major compulsory revisions.
In my opinion, no major compulsory revisions should be made to the paper.

Minor essential revisions.
1) Background, page 3: In my opinion, reference 3 (Coss et al 2013), that is referred to a very precise population, is not pertinent with the presentation of general long-term complications that are common to galactosemic people worldwide. I’d suggest to replace current reference 3 with Waisbren et al., J Inherit Metab Dis 2012 (PMID: 21779791)

2) Methods: can Authors include information, if known, about compliance of patients to galactose-free diet?

3) Results, paragraph “Mutation spectrum of the GALT gene in Korean patients”, page 8: please correct the mistake in the code c.940A>G.

4) Discussion, page 9: the Authors state that “Our study also revealed that measurement of GALT enzyme activity was not enough to predict the genotypes and to make a decision for treatment in some patients”. I think that the lack of correlation between genotype and phenotype in classic galactosemia is already well known, therefore this study does not reveal any new data on this point. Authors should amend this sentence accordingly.

5) Discussion, page 11: the Authors make the hypothesis that the prevalence of classic galactosemia is increasing in the Korean population, on the basis of the results of newborn screening before 2006 and in 2012. However, this difference could be due to a different panel of population screened, or to different methods used more recently with respect to old methods, or to other variables that they take into account e.g. to discuss the different prevalence of classic galactosemia among different countries. Can Authors rule out these hypotheses before suggesting that the prevalence of classic galactosemia is strikingly increasing in Korea?

6) Table 1, page 19: please explain the meaning of superscript “d” near mutations p.Asn314Asp referred to cases 5 and 6, that is not present in the legend. Maybe this is a mistake? (the symbol should be “§”, as in cases 11, 12 and 13?)

Discretionary revisions
I don’t suggest discretionary revisions for this paper.

**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.

**Declaration of competing interests:**

I declare that I have no competing interests