Reviewer's report

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

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Reviewer: Kent Lai

Reviewer's report:

Overall, it is an interesting and informative manuscript. The authors did an excellent job in the writing; very easy to understand and good use of the English language. A major concern for this paper lies on the lack of expression studies of the three nucleotide changes in the GALT gene. Although the clinical data and the use of computer modeling programs predicted that these changes are detrimental, these results are by no means definitive, especially due to the wide range of GALT activity in the normal controls. Since there are only a handful of "mutations", this reviewer believes that the authors should make an effort to express these variant GALT genes to see if the nucleotide changes are truly detrimental. Without definitive proofs, it is too early to claim that these changes are mutations. Also, the inclusion of expression studies will improve the overall quality of the paper.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: Yes, and I have assessed the statistics in my report.

Declaration of competing interests:

No competing interests to declare.