Reviewer's report

Title: A novel c.-22T>C mutation in GALK1 promoter is associated with elevated galactokinase phenotype

Version: 4 Date: 30 January 2009

Reviewer: Yoshiyuki Okano

Reviewer's report:

This manuscript described that the increased GALK1 activity was caused in a c.-22T>C mutation of the promoter region in the positive subjects of newborn mass screening for galactosemia. The authors confirmed the increased transcription activity in the promoter analysis. This is the first report for the up-regulation of the GALK1 activity by the molecular basis. Authors answered all the questions and corrected the points in the last manuscript adequately.

Level of interest: An article of importance in its field

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.