Reviewer's report

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

**Version:** 2  **Date:** 15 December 2008

**Reviewer:** Yoshiyuki Okano

**Reviewer's report:**

This paper shows that elevated GALK activity is found in 13 of 63 subjects which show positive results in newborn mass screening for galactosemia. And authors have identified a c.-22T>C mutation in the promoter region as a cause of up-regulation, and have confirmed the increased transcription activity in the promoter analysis. This is the first report for the up-regulation of the GALK activity by the molecular basis and is found to be interesting.

1) In Abstract, EMSA needs full spelling.
2) In Methods, Authors should describe the method (for example, colorimetric or Paigen method) and the measurement materials for the galactosemia in newborn mass screening in Korea. The values of the measurement materials should be described in 63 positive patients, especially c.-22TC positive patients, to easily understand the situation of the patients.
3) In Table I, 65 subjects are written, but it is 63 subjects in the text.
4) In Figure 3, authors should rewrite c.22T instead of AAT, c.177G instead of ACT, c.27C instead of ACT, c.22C instead of AAC.
5) In Figure 4, please show the lane number (1-10).

**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.