Reviewer's report

Title: A Novel Mutation in the Glycogen Synthase 2 Gene in a Child with Hypoglycemia Due to Glycogen Storage Disease Type 0

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Reviewer: Tjin-shing Jap

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In this paper, Soggia et al. have reported an interesting case of Glycogen storage disease (GSD0) in a patient with 6 months of age. They found a compound heterozygous for one previously nonsense mutation (c.736 C>T; R243X) which was inherited down from the mother and a novel frame-shift mutation (966_967 delGA/insC) from the father, respectively.

The following ones had been corrected

1. In background, the author had described the properties of hepatic glycogen synthase including 80.9 KD, when the GYS2 gene was cloned, and the number of the exons. In addition, this is the first case with Glycogen storage disease (GSD0) reported in his home country.

2. The gender of patient is female

Suggestion:

1. In Page 4, hepatomegaly

2. In page 5, Glycemia and lactemia were measured by immunoenzymatic method # The plasma glucose and blood lactate were measured by the enzymatic method.

3. In page 7, the first paragraph in the discussion of R246X, the reference should be cited.