Reviewer’s report

Title: Mitochondrial mosaics in the liver of 3 infants with mtDNA defects

Version: 4 Date: 24 December 2008

Reviewer: Richard rodenburg

Reviewer’s report:

Most answers are satisfactory. The following issue remains:

Major comment

Several clinical and biochemical features of patient #2 are in agreement with the assumption that the splice site change in one of the POLG1 alleles of this patient is pathogenic. I can agree that detailed studies of POLG1 mRNA expression are beyond the scope of this manuscript, but some additional research should be done in order to confirm the pathogenicity of the splice site variant. For example, online available resources could be consulted, e.g. to verify evolutionary conservation and to predict the effect of the change on splicing. And has it been checked that the splice site variant is not on the same allele as the 1399 G>A mutation?

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.