Reviewer's report

Title: A novel compound heterozygous mutation in a Chinese boy with L-2-hydroxyglutaric aciduria: a case study

Version: 2 Date: 2 May 2015

Reviewer: nadege kammoun

Reviewer's report:

Dear Editor

Many thanks for inviting me to review the manuscript: A novel compound heterozygous mutation in a Chinese boy with L-2-hydroxyglutaric aciduria: a case study

Article title: A novel compound heterozygous mutation in a Chinese boy with L-2-hydroxyglutaric aciduria: a case study

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Find attached my reviewer report

1. The author must indicate the effect of the substitution of amino acid glycine by valine on the enzyme since the variation c.542G>T in exon 5 had not been reported before.

2. the author can improve figure 2 by adding the chromatograms of parents and mention which one of the 2 chromatogram is c.542G>T and which one is c.169G>A.
Level of interest: An exceptional article

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.