Reviewer's report

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

Version: 2  Date: 23 April 2015

Reviewer: Fonnet Bleeker

Reviewer's report:

Tai and colleagues present an interesting case of a patient with L2 hydroxyglutaric aciduria. Although his case report is interesting for publication, I suggest major compulsory revisions.

1. Readers would be helped when a figure of the GC-MS spectrum is shown, showing the increased L2HG concentrations.

2. More information is required on how the authors separated the 2HG L-enantiomer from the 2HG D-enantiomer in their GC-MS analyses.

3. Figure 2 needs clarification for BMC Neurology readers. An arrow indicating the mutation on this Sanger plot would help.

4. The finding that this patient has not one, but two mutations in L2HGDH, seems special. The authors should emphasize this more in their report and, if available, relate this to other reports in the field on double-mutated L2HGA patients.

5. The entire manuscript text needs serious editing from someone with good English proficiency. The current manuscript is sometimes barely readable due to grammatical errors and translating-machine artefacts. From the abstract alone:

   who had characteristic developmental delay, ataxia and acrocephaly as the main features. Features = symptoms?

   He also complained ?? paroxysmal headache and palpitation.

   Urine test for organic acids showed a significantly increase (increased?) level of 2-hydroxyglutaric acid

   169G>A in exon 2 and c.542G>T in exon 5, not hitherto ?? described.

   Novel gene mutation and associated clinical symptom ?? can contribute for the understanding and identification of this rare disease. Possible genotype-phenotype correlation waits further investigation ??

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Not suitable for publication unless extensively edited
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