Reviewer’s report

Title: Identification of novel L2HGDH mutation in a large consanguineous Pakistani family- a case report.

Version: 1 Date: 10 Jul 2017

Reviewer: Dinesh Rakheja

Reviewer's report:

Since elevated blood/urine level of L-2-hydroxyglutarate is the defining feature of L-2-hydroxyglutaric aciduria, I think this is an essential piece of information missing from the manuscript. Also, please see attached results from Alamut Visual re: the L2HGDH mutation.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

No

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

No

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published
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