Reviewer’s report

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

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Reviewer: Thong Meow Keong

Reviewer's report:

The manuscript is well written and comprehensive from the laboratory aspects.

However, the major shortcoming is the lack of clinical data regarding the other 5 family members. The clinical information obtained from these extended family members will be interesting to clinicians and other families as it would contribute to genotype-phenotype correlation. The authors mentioned that to date, there was no clear correlation in other reports. It is recommended that further efforts be made to contact the health providers or family members for an update on the clinical condition of the other 5 individuals.

In addition, there are reports in the literature on the use of riboflavin in this condition. The authors mentioned 'appropriate medication for symptomatic treatment' was given. It will be useful to the clinicians to provide the latest review or information on the use of riboflavin and other co-factors in this condition.

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

Yes

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.

Yes

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:

Acceptable

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