Reviewer’s report

Title: A Chinese pedigree with Brown-Vialetto-Van Laere Syndrome due to two novel mutations of SLC52A2: clinical course and response to Riboflavin

Version: 0 Date: 05 Mar 2019

Reviewer: Maria Barile

Reviewer's report:

The paper entitled "A Chinese pedigree with Brown-Vialetto-Van Laere Syndrome due to two novel mutations of SLC52A2: clinical course and response to Riboflavin " by Shi K et al., deals with the first Case report in a Chinese pedigree of mutations in the gene encoding the plasma membrane riboflavin transporter SLC52A2, leading to Brown-Vialetto-Van Laere (BVVL) Syndrome, a rare neurological disorder characterized by motor, sensory, and cranial neuronopathies. Clinical outcomes describe here significantly improved by high dose riboflavin supplementation and this stresses the necessity of an early diagnosis and treatment of this rare metabolic disorder. This is what the authors recommend.

I would have expected (preferred) some biochemical characterisation of patient fluid/cell metabolic alterations; nevertheless, in my opinion, the discovery of novel pathogenic variations of SLC52A2 is quite important -per se and for the scientific community - to justify publication in BMC Medical Genetics.

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

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Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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