Reviewer’s report

Title: Leukoencephalopathy, demyelinating peripheral neuropathy and dural ectasia explained by a not formerly described de novo mutation in the SAMD9L gene, ends 27 years of investigations – A case report

Version: 1 Date: 16 Mar 2019

Reviewer: Hiroaki Adachi

Reviewer's report:

The authors reported the case of a female with de novo variant c.2686T>G, p.(Phe896Val) in SAMD9L gene. She was affected by paleness, petechiae, pancytopenia, generalized joint laxity, motor incoordination, weakened tendon reflexes, intra cerebral hemorrhage cerebellar ataxia and her brain MRI showed cerebellar atrophy, disseminated cysts in white and grey matter, dural ectasias and white matter hyperintensity. Her nerve conduction studies and electromyography suggested demyelinating neuropathy. I believe that this is an important paper describing a rare neurological disease. There are a several comments that the authors should address prior to publication:

1. The authors did not describe the origin of her gait disturbance. They should describe precisely and discuss them with the MRI findings. The authors did not describe the cerebellar atrophy.

2. The authors should add figures of the spinal cord MRI to facilitate a better understanding of the precise patient pathology because her tendon reflexes in patella were recovered and her brain MRI shows atrophy of the spinal cord.

3. The authors should add the explanation of the yellow arrow in the figure legend of the figure 2.

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.

Unable to assess
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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