Reviewer’s report

Title: Leukoencephalopathia, 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: 19 Mar 2019

Reviewer: Bianca Tesi

Reviewer's report:

The authors report about an interesting case with a novel de novo variant in SAMD9L identified in a patient with predominant neurological symptoms, but also a history of transient cytopenia with spontaneous recovery. It is an interesting report since the neurological aspects of SAMD9L-associated syndrome are probably not yet fully described.

I have the following two comments:

1) Chen et al (PMID 27259050) and Tesi et al (PMID: 28202457) should be referenced when talking about SAMD9L mutations in ataxia-pancytopenia syndrome. At the moment only reference to the initial clinical descriptions of this syndrome are included (ref 1 and 2 in the current draft).

2) It has been shown (Tesi et al 2017) that self-resolution from cytopenia is a feature of this syndrome and that it is due to revertant mosaicism in blood cells. I would like the authors to state if additional variants where found in SAMD9L gene that could represent in-cis revertant variants or if there were signs of acquired UPD of chromosome 7. I think this analysis would be a valuable addition to this case report.

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:

Acceptable

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