Reviewer's report

**Title:** Novel SCARB2 mutation in Action Myoclonus-Renal Failure syndrome and evaluation of SCARB2 mutations in isolated AMRF features

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**Reviewer:** Gambardella Antonio

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Hopfner and co-workers in their paper give evidence that a demyelinating neuropathy can be part of the action myoclonus-renal syndrome (AMRF) that has been associated with SCARB2 mutations.

**Major Compulsory Revisions**

1. The authors should mention a recent paper dealing on the same topic (Arch Neurol. 2011, 68: 812-813) that reports a patient with PME due to SCARB2 mutation who also had a demyelinating neuropathy. In such a patient, a peculiarity was the lack of any symptoms suggestive of a sensory, motor, or autonomic neuropathic. Moreover, there was a longitudinal stability of electrophysiological abnormalities. Did the affected AMRF members have a similar phenotype of demyelinating neuropathy?

2. The authors also claimed a possible pathogenic role of SCARB2 in patients with isolated polyneuropathy, as two of 103 patients with a diagnosis of polyneuropathy (PNP) carried a heterozygous mutation of SCARB2. Their data do not support this assumption. It would be important to give more details about the clinical and electrophysiological features of these two individuals. Had both of them a demyelinating polyneuropathy? Were alternative diagnosis ruled out? It is reasonable that these two patients had clinical symptoms of PNP, while this seems not to be the case in the affected members of the family with AMRF.

**Discretionary Revisions**

1. The authors also emphasized that the frequency of SCARB2 mutations in PME may differ between different populations as their findings are not in line with a recent report of high frequency of homozygous or compound heterozygote SCARB2 mutations in patients with isolated PME who are negative for CSTB mutations. Since all these patients were Italian, it would important to specify the ethnic origin of their PME patients.

2. There are several grammatical errors throughout the manuscript (e.g; a 27-years-old..., discussion, page 9).

3. Figure 4 is unnecessary and should be omitted.

**Level of interest:** An article of importance in its field
Quality of written English: Needs some language corrections before being published

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
I declare that I have no competing interests' below.