Reviewer's report

Title: Dopamine-agonist responsive parkinsonism in a patient with the SANDO syndrome caused by POLG mutation

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Reviewer: Matthias Synofzik

Reviewer's report:

The authors report a case with SANDO plus parkinsonism due to recessive POLG mutations. Parkinsonism showed a response to dopamine agonists.

MAJOR REVISIONS

I am not sure whether the case report entails any fundamental novel aspect. Neither the finding of parkinsonism, nor the finding of SANDO nor the finding of the combination of both of them is novel with respect to POLG.

1) Throughout the last 10 years, parkinsonism has been well reported to be part of the POLG-associated plethora, also in combination with SANDO (Luoma et al., 2004; Davidzon et al., 2006; Invernizzi et al.; 2008; Remes et al., 2008; Hudson et al., 2007; Synofzik et al., 2010). Also good and sustained response to dopamine-agonists has already been well documented three years ago (Synofzik et al., 2010). Even pathological findings on substantia nigra changes in POLG Patient with and without Parkinonism are available (Tzoulis et al., BRAIN).

2) It has been well reported that SANDO is caused by POLG (see Schulte et al, 2009; Neurology, demonstrating that 80% of all SANDO cases are caused by POLG). I am surprised that the authors neglect this literature and discuss their case as if further evidence for this well-known fact would be needed.

MINOR REVISIONS

1. The improvement in UPDRS was about 30%. This should not be called „dramatic“ as this is the standard improvement seen in idiopathic Parkinson’s Disease.

2. Lease provide the exact MMSE value rather than stating that cognition was „affected upon MMSE“ testing.

Level of interest: An article of limited interest

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.