Reviewer’s report

Title: Identification of a novel mutation in PEX10 in a patient with attenuated Zellweger spectrum disorder: a case report

Version: 0 Date: 11 Dec 2016

Reviewer: Heiko Reutter

Reviewer's report:

The authors report a case of Zellweger spectrum disorder due to a novel homozygous mutation (c.530T>G) (p.Leu177Arg) (NM_153818.1)) in the PEX10. The authors demonstrate thoroughly the diagnostic workup including biochemical tests, that pointed to the Zellweger spectrum disorder.

Minor comments:

1) Interestingly, the diagnosis was made quite late (age 15 of the patient). It would be interesting to know, which "top" differential diagnosis were discussed during the long diagnostic course of the child, till the diagnosis was made biochemically and genetically.

2) It would be interesting to know, which genetic tests were made before NGS-Panel-testing was performed for suspicion of Zellweger syndrome?

3) Is it possible to get pictures of the patient at different times of development (5 years of age, 10 years of age, etc.) or at least MRIs of the patient?

Level of interest

Please indicate how interesting you found the manuscript:

An article of importance in its field

Quality of written English

Please indicate the quality of language in the manuscript:

Acceptable

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