Reviewer’s report

Title: Childhood neurodegeneration associated with a specific UBTF variant: a new case report and review of the literature

Version: 0 Date: 02 Oct 2019

Reviewer: Reviewer 2

Reviewer's report:

PEER REVIEWER ASSESSMENTS:

RELEVANCE - Does this case report make a contribution to medical knowledge, have educational value, or highlight the need for a change in clinical practice or diagnostic/prognostic approaches?

Yes, this report contributes to medical knowledge

CASE DESCRIPTION - Are the details of the case sufficiently well described to understand the patient's symptoms and course of treatment?

Yes, the description of the case is sufficient

DIAGNOSIS/INTERPRETATION - Based on the facts presented, are the diagnosis, interpretation, and course of treatment medically sound?

Yes, the work described is medically sound

DISCUSSION OF THE CASE - Does the discussion appropriately analyse the importance of the findings and their relevance to future understanding of disease processes, diagnosis or treatment? Has an adequate literature review pertinent to the case been included?

Yes, the case is discussed fully in the context of the literature
OVERALL MANUSCRIPT POTENTIAL - Could an appropriately REVISED version of this work represent a technically sound contribution?

Yes, this case report is suitable for publication

PEER REVIEWER COMMENTS:

GENERAL COMMENTS: The manuscript is well written and describes in detail the presentation of another patient with the recurrent gain-of-function mutation in UBTF that results in the published phenotype of UBTF associated neuro-regression. This manuscript highlights new phenotypic features including the evolution of clinical and EEG aspect of the disease that are punctuated seizures and accelerated periods of decline during periods of decompensation.

REQUESTED REVISIONS:

Manuscript is suitable as written.

ADDITIONAL REQUESTS/SUGGESTIONS:

Figures #1 could benefit from better labeling.

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

Quality of written English
Please indicate the quality of language in the manuscript:
Acceptable

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