Reviewer's report

Title: Clinical features and genetic analysis of two siblings with Startle disease in an Italian family: a case report.

Version: 0 Date: 28 Jan 2019

Reviewer: Amrik Sahota

Reviewer's report:

Comments:

Line 30: Change "mutations in other 4 genes" to "mutations in 4 other genes"

Line 39: Change "Center" to "center"

Lines 73, 74: Were both probands older than 18 at the time of the DNA study or was consent for both of them given by the parents? You may need to clarify this.

Lines 91, 92: "all of the patients' parents and probands were screened using Sanger sequencing:" This is confusing. You may wish to change it to: "DNA samples from the probands and their parents were screened using Sanger sequencing:"

Line 104: Is the MAF for c.587C>A listed in the ExAC database? If so, it should be reported here.

Line 129: Change "heterozygote" to "heterozygous". Also, are you making a distinction between recessive and compound heterozygous mutations? The latter simply means that the proband inherited two different mutant alleles, one from each parent. The inheritance pattern is still recessive.

Lines 135, 136: The sentence "GLRA1 accounts for about 80% of cases and pathogenic variants in another 4 genes." is not clear.
Line 144: Change "Mother's patients " to "Patients' mother "

Lines 148-150: If the mother is showing mild symptoms, it means the mutation is showing variable expressivity rather than incomplete penetrance. Since the father showed no symptoms, non-penetrance is correct terminology for him.

Line 157: " compound heterozygosis mutation in GLRA1 is the same." This is not clear. Do you mean: " compound heterozygous mutations in GLRA1 are the same." Note spelling in bold.

Line 170: Change "variable penetrance” to "variable expressivity"

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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