Reviewer's report

Title: A novel homozygous frame-shift mutation in the SLC29A3 gene: A new case report and review of literature

Version: 0 Date: 13 Apr 2019

Reviewer: Abraham Zlotogorski

Reviewer's report:

This is an interesting report of four H syndrome patients, with a homozygous deletion in the SLC29A3 gene. These cases highlight the phenotypic variability in this syndrome, and therefore are of importance for clinicians when examining patients with suspected H syndrome. Several points should be addressed by the authors:
P. 3, line 13: Correct "could results" to "could result"
P. 3, lines 13-54: H syndrome, PHID, Faisalabad histiocytosis and DSS should not be described and referred to as different conditions. All these disorders are actually different manifestations of the same condition, and their description as different entities is misleading. Therefore, the introduction should highlight this point, and make clear that all these diseases, with their different names and terminology, are actually the same entity, termed H syndrome. The fact that these are different manifestations of the same condition should also be integrated into the abstract and discussion parts of the manuscript.
P. 3, line 23: Correct comptodactyly with camptodactyly
P. 3, line 30: Correct comptodactyly with camptodactyly
P. 3, line 35: Correct comptodactyly with camptodactyly
P. 7, lines 24-32: Proptosis is a consistent finding in H syndrome patients, seen in more than a quarter of the patients. This is also probably a misnomer, and the correct terminology should probably be exorbitism.
P.7, lines 35-37: It would be of benefit to the readers if clinical representative photos of the camptodactyly will be provided.
P. 7, lines 37-52: Elevated inflammation markers such as CRP and ESR have been reported before for H syndrome patients, and therefore they should not be attributed to RA, but rather to the inflammatory process seen in H syndrome. On this basis, the diagnosis of RA should be put under scrutiny, as joint manifestations are a common finding in H syndrome patients.
P. 8, lines 2-4: There is a HUGE difference between the diagnosis of lymphoma and the lymphadenopathy observed in H syndrome patients, which is absolutely not malignant. The authors should provide more details on the diagnosis of lymphoma in the described patient.
P. 12, Table 1: Include age of onset for the different clinical findings
P. 12, Table 1, footnote: Correct "was died" to "died"
Figure 2 can be deleted, since it repeats the information given in figures 1 and 3.
Table 2 should be deleted, since it doesn't add new information.
The manuscript suffers from many grammatical errors, and should be edited by a native English speaker.
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:

Needs some language corrections before being published

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