Reviewer's report

Title: A de novo ANK1 mutation associated to hereditary spherocytosis: a case report

Version: 1 Date: 06 Nov 2018

Reviewer: Susana Rocha

Reviewer's report:

General considerations:

The subject matter of this work is well within the scope of the BMC Pediatrics Journal and it is a very to the point, clear-cut paper as any case report should be.

The novelty of the work is to present a new mutation underlying HS, proving further knowledge about the physiopathology of this hereditary disease.

Specific considerations:

In the overall text, the authors present the expression "de novo" without being in italic in several instances. As is it a Latin expression, it should be denoted differently from the rest of the text (in English).

Line 1 - In my opinion, "risk factor" is something that increases predisposition or susceptibility of developing a disease, such as, a habit or an environmental condition. A mutation is something that is intrinsic to the individual thus calling it a risk factor is a bit exaggerated. I suggest changing the title to "… mutation underlies a new case of …." or "… mutation associated to…"

Lines 39 and 143 - What do the authors mean by "intractable"? Are they saying that some hemolytic anemias are difficult to diagnose by routine means or do they mean that these HA are not possible to be cured, as they are hereditary diseases? The authors should rephrase the sentence to be better understood.

Lines 50-51 - As Band 3 is more common name for the protein encoded by the "SLC4A1" gene and it is usually what is described in HS articles, the authors should add "or Band 3" after "soluble carrier family 4, number 1".

Line 76 - Because blood transfusions can adulterate the analytic results it should be specified that the routine blood examinations were performed before the transfusion (if it was the case, as I believe so).
Lines 104-105 and 155 - RBCs acronym is widely used to refer to "red blood cells" and not "spherical-shaped blood cells". As it can cause confusion, I urge the authors to choose another abbreviation.

Line 108 - delete "all"

Line 121 - delete "ankyrin"

Line 140 - substitute "database" by "databases"

Line 232 - substitute "2018:1-4" by "2018; 23:413-16"

Line 245 - substitute "Spherocytosis are indicated by an arrow" by "Spherocytes are indicated by arrows"

Line 247 - substitute "HS" by "gene mutation"

Line 248 - substitute "indicated" by "indicates"

Line 249 - Change to the title of the table to something similar to "Laboratory test results of the patient at time of birth" and RBC value is within the reference range, so the arrow should be deleted.

Line 255 - Change to the title of the table to something similar to "Laboratory test results of the patient at eleven months of age"

Line 258 - substitute "G-6PD" by "G-6-PD"; also, in the table.

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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