Reviewer’s report

Title: PHENOTYPIC AND GENOTYPIC ASPECTS OF TOWNES-BROCK SYNDROME: CASE REPORT OF PATIENT IN SOUTHERN BRAZIL WITH A NEW SALL1 HOTSPOT REGION NONSENSE MUTATION

Version: 0 Date: 14 Dec 2016

Reviewer: Yolande Van Bever

Reviewer's report:

Dear authors,

I think the manuscript is well written and I know it's difficult to get case reports published nowadays, even though they are important to enhance recognition of entities and to draw attention to some specific or uncommon feature. However, TBS has been described quite extensively before and I didn't read anything new. Maybe it's difficult to get access to all of the papers, in which case you might find another (Brazilian) journal for publication, or maybe you could try to contact other centers such as the genetic departments of e.g. the USP (SP), or the hospital for craniofacial anomalies in Bauru (SP), to try and collect a larger sample.

Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Unable to assess

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Unable to assess

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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