Reviewer’s report

Title: Inclusion of joint laxity, recurrent patellar dislocation, and short distal ulnae as a feature of Van Den Ende-Gupta syndrome: A case report

Version: 0 Date: 15 Nov 2017

Reviewer: Jennifer Below

Reviewer's report:

The authors detail what is presumed to be a unique presentation of Van Den Ende-Gupta Syndrome in a pair of siblings from a consanguineous Saudi family. Phenotypic features include those typical for the syndrome, as well as joint laxity, patellar dislocations, and short distal ulnae. The siblings have a homozygous mutation (c.773G>A) in SCARF2 which is known to cause this syndrome. The authors also present the results of a literature review, grouping clinical features by frequency of observation.

Major critiques:

1. There is no acknowledgement of other potential genetic or environmental explanations for this unique phenotypic presentation. Could this phenotypic presentation be due to interaction or a distinct recessive phenotype in this consanguineous sibling pair? The authors mentioned that the cases have two unaffected siblings. The unaffected siblings' genotypes could provide additional evidence of causality. Did the unaffected siblings share any symptoms with the cases?

2. It is unclear if Marden-Walker syndrome is mentioned because it was considered in diagnosis of the presented cases. This should be clarified.

Minor critiques:

1. Due to the rarity of this syndrome, it may be more useful to specify the number of observations of each phenotypic feature, rather than binning them into such wide categories. This will give a better idea of how many other features are observed as rarely as the unique ones reported here.

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.

No

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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