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: 01 Nov 2017

Reviewer: Sulman Basit

Reviewer's report:

The authors have sequenced SCARF2 gene in 2 patients with VDEG syndrome from a consanguineous family and identified a homozygous missense mutation. This mutation has already been reported by Anastasio and colleagues (2010). Authors proposed to include some extra clinical features to the spectrum of VDEG syndrome.

Some limitations exist

Major Concern:

1. In consanguineous marriages, biallelic or even triallelic mutations in two distinct genes or co-inheriting genetic modifiers may well be considered. I would suggest whole exome sequencing to identify other variants segregating with the phenotype which might explain the additional common phenotype in patients.

2. Both patients share some unusual features and SCARF2 protein does not fully explain the pathogenesis of most of the clinical features of VDEGS as mentioned by authors on page 7, 2nd paragraph. This is another reason to consider the possibility of co-inheriting variants.

Minor Comment

I would suggest replacing title with "Inclusion of joint laxity, recurrent patellar dislocation, and short distal ulnae as a feature of Van Den Ende-Gupta syndrome: A case report"

Abstract; conclusion, please replace "short distal ulnae should be included in the VEDGS phenotype" with "short distal ulnae should be included as a part of clinical spectrum of VEDGS"

Page 5, case presentation, last paragraph, reference 4 is wrongly cited. Please cite Anastasio et al., 2010.
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.

No

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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