**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:** 25 Oct 2017

**Reviewer:** Bhavana Dave

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The case report is regarding a rare autosomal recessive Van Den Ende-Gupta Syndrome (VDGES) in two siblings born to consanguineous parents and presenting with joint laxity, recurrent patellar dislocation, and short distal ulnae, which have hitherto not been associated with the clinical constellation of the syndrome. The report is well presented with appropriately depicted images justifying the inclusion of the phenotypic features in the spectrum of VDGES syndrome. The description and figures are adequately described in support of the diagnosis of VDGFS. The rarity of this recessive disease, family history of consanguineous parents, scarcity of published reports, supportive molecular and clinical findings, and clearly described additional non-typical clinical features in both siblings with pictorial depiction makes this case report interesting.

Some minor suggested changes:

1. Page 5 - The first paragraph should describe how and at what age the cases were referred to the clinician.
2. Page 5 line 91- please include the underlined words in the line. The phenotypes of ......siblings, a male and a female, were…
3. Page 8 line 176 space between Hospital & research Center
4. Page 8 line 187-188. Please include the underlined words in the lines - Authors' contribution: All authors .... and are in agreement…….manuscript. All authors have read…….manuscript.
5. Page 8 line 190- If there are no acknowledgements remove the title and remove the word-None.
6. Page 11 line 249 hands of "male sibling" instead of "brother"
7. Page 11 line 252 Figure legend should read: Figure 2: Radiologic images of classic features of the VAGDS syndrome (instead of demonstration of other….).
8. Page 11 line 257 Figure 3: A) legend should read the appearance…..of the "female sibling" instead of "sister"

9. The clinical features are getting lost in the table. If possible, Table 1 may be reformatted in three columns; each comment can be abbreviated in title. For e.g. "Constant" features in 90-100% cases; "Common" features in 40-89% cases; "Infrequent" features in less than 40% cases. List features under each column.

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.

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:

Needs some language corrections before being published

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