Reviewer’s report

Title: De novo truncating variant in WHSC1 gene leading to mild Wolf-Hirschhorn syndrome phenotype: a case report

Version: 0 Date: 11 May 2019

Reviewer: Nobuhiko Okamoto

Reviewer’s report:

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The authors reported a boy with de novo truncating variant in WHSC1 with similar but mild clinical features comparing with typical 4p16.3 deletion related WHS. This is an interesting report covering WHSC1 mutations.

1. Please show the SD score or percentile of occipitofrontal circumference.

Growth parameters should be included in the Table.

2. How about the endocrinological data including GH?

3. Figure of NSD2 with its domains and positions of reported variants will be a help for readers.

"de novo" "de novo"

Page numbers of the references lack coherence.

Please check other minor errors.

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?
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No

**Are the conclusions drawn adequately supported by the data shown?**
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Not relevant to this manuscript

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