Reviewer's report

Title: Familial imbalance in 16p13.11 leads to a dosage compensation rearrangement in an unaffected carrier

Version: 2 Date: 1 September 2014

Reviewer: SANDESH CHAKRAVARTHY SREENATH NAGAMANI

Reviewer's report:

The authors have addressed all of my comments from the review. I personally think that the Manuscript reads much clearer in the present format and emphasizes the uniqueness of rearrangement while still alluding to the phenotypic consequences of CNVs of the region.

Minor essential Revisions

Figure 2: Seems to be truncated. I could not see the STR markers in the proband and his brother and the upper portions of the grandparental markers again were not seen. I think this must be due to the way the PDF was built. I would encourage the authors to make sure that this is corrected at the later stages.

Figure 3: Not been cited in the text.

Minor text changes
1) Clinical report section
"Samples from patients and their families were obtained after informed consent."
Samples from the proband and his family were obtained after informed consent.

2) "A prenatal ultrasound at 28 weeks gestation revealed an agenesis of corpus callosum." Please delete "an"

3) Results section
Array comparative genomic hybridization in the patient revealed a gain of 1,78 Mb on '..
Should be 1.78 Mb

Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:
No conflicts of interest to disclose