Reviewer's report

Title: A Novel Single Base Pair Duplication in WDR62 Causes Primary Microcephaly

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Reviewer: Bassam R. Ali

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A Novel Single Base Pair Duplication in WDR62 Causes Primary Microcephaly
Rupp et al

The authors report in this case report the identification of a novel mutation in the WDR62 gene underlying microcephaly in a consanguineous Pakistani family. Reporting of clinical and molecular data of heterogeneous conditions such as microcephaly is often a useful addition to the literature. The manuscript is well written. However, I have the following comments for the authors to address:

Major Compulsory Revisions
None

Minor Essential Revisions
1. Lanes 30 and 147: change confirmation to conformation
2. Lanes 32-33: Delete “It also opens many insights into disease understanding.” The sentence is not that accurate
3. Lanes 51-52: Delete “(Roberts et al. 1999)”
4. Table 1. Since the authors are listing all the reported mutations in table 1, they should perform a more thorough search of the literature. For examples, the mutation reported by McDonell et al (2014) is not listed.
Therefore, the authors should carry full literature review and add any missing mutations to table 1.
5. Figure 3. I prefer it if the pedigree is presented earlier; perhaps this could be by joining the clinical data (photos in figure 1) and the pedigree (figure 3) in a single figure 1.

Discretionary Revisions
1. Figures 2 (homozygosity mapping) and 4 (the Sanger sequencing chromatograms) can be merged into a single figure

2. The authors could re-check the English throughout the manuscript to eliminate any errors

**Level of interest:** An article of limited interest

**Quality of written English:** Needs some language corrections before being published

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

**Declaration of competing interests:**

I declare that I have no competing interests