Author’s response to reviews

Title: Whole-exome sequencing identifies homozygous mutation in TTI2 in a child with primary microcephaly: a case report

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1. Please provide further justification for the inclusion of NC and BL in the authors list in the Authors' Contributions section.

To clarify the role of NC and BL in the author's contributions, we changed the following sentence: N.C and B.L investigated and followed the patient, designed the study and substantively revised the manuscript.

2. Please crop the images in Figure 3, particularly part B and D. Please ensure that the eyes are cropped out and as much of the hairline as possible.

We did crop the figure 3B and D as much as possible.

3. Please proofread and ensure that when you upload your revised submission it is in the final form for publication. Please remove any tracked changes or highlighting and include only a single clean copy of the manuscript.

We did remove any tracked changes and highlighting.

Ni-Chung Lee (Reviewer 1): All the points had been addressed, though the author did not understand the request of listing the % of patients having a specific phenotype for table 1, so that reader can understand more about the clinical phenotype. It will be great if author can do so.

We thanks the reviewer for this clarification. We added the following sentence at page 6 line 22: The phenotype is similar in all cases and the most frequent findings include primary or progressive microcephaly (80%), dysmorphic features (80%), severe cognitive impairment (70%), severe speech delay (70%), strabismus (70%), movement disorder (60%), short stature (60%) and scoliosis (50%) (Table 1).