Author’s response to reviews

Title: Xp11.22 Duplications in Four Unrelated Chinese Families: Delineating the Genotype-phenotype Relationship for HSD17B10 and FGD1

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Author’s response to reviews:

1. Manuscript format
   Response: I have reformatted the main body of text according to Editor’s requests.

2. Consent to participate
   Response: Written informed consent to participate in the genetic testing and this study was obtained from the patients’ families, including patients, the patient’s mothers, the patient’s uncle in family 1, and the maternal grandparents in family 3.

3. Consent for publication
   Response: I have corrected this section according to the Editor’s requests.

4. Case reports
   * Please describe how this case report is exceptional or unusual
     Response: Xp11.22 has been reported to be associated with nonsyndromic ID. HUWE1 gene duplication in this interval has been believed to be responsible for nonsyndromic ID for Xp11.22 microduplication patients. Currently, few Xp11.22 duplication cases were reported in Chinese population with limited knowledge regarding the role of other genes in this interval. Here, we ascertained four unrelated Chinese Xp11.22 duplication male patients and performed a comprehensive clinical evaluation for all patients and discussed the role of other genes in this interval. In this study, we further explored the genotype-phenotype associations and the potential candidate genes, HSD17B10 and FGD1 genes may be involved in the process.
   * Please describe the educational and/or scientific merit of the report
     Response: We reported novel genotypes and phenotypes in Chinese Xp11.22 duplication male patients and HSD17B10, FGD1 genes may be involved.
   * Does the case report include an up-to-date list and review of all previously published cases in the field?
     Response: Yes

5. CARE checklist
   Response: I have corrected this section according to the Editor’s requests.