Author's response to reviews

Title: Identification of a rare 17p13.3 duplication including the BHLHA9, YWHAE genes in a family with developmental delay and behavioural problems

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Author's response to reviews:

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Dear Editor,

Attached please find the revised manuscript n. 8281239926995772 entitled “Identification of a rare 17p13.3 duplication including the BHLHA9, YWHAE genes in a family with developmental delay and behavioural problems” by Capra et al., for publication in BMC Medical Genetics.

Reviewer: Damien Bruno

Major compulsory revision

We have included cases 9 and 11 reported from the study by Bruno et al. in our report comparing them with our 17 p13.33 duplication case and subjects 1 and 2 from the study by Bi et al.

The manuscript has been reviewed by a native English speaker. (This revision was required also by Reviewer Roberto Ravazzolo)

Background

We have rewritten this section differentiating the submicroscopic duplications from the deletions in 17p13.3. We have also distinguished and described the classe I and class II of 17p13.3 duplication discussing the associated phenotypes.

Discussion

1. We have corrected the genomic description of MDS.
2. We have read the report of Albert et al., Nat. Genet 2012 and we have deleted the reference to the molecular mechanism underpinning TAR syndrome.
3. We have modified the conclusion to give a more comprehensive assessment of the minimal region of overlap for class I duplication after inclusion of this case.
4. In the discussion we have discuss the clinical complexities in this family, the genetic and environmental contributors to the phenotype of the probando and his mother. (This revision was required also by reviewer Roberto Ravazzolo)

We have accepted all the minor essential revisions and we have corrected the text as suggested.

We are grateful to Reviewer 1 and 2 for their precious suggestions that allowed us to improve the understanding and the quality of our manuscript.

We are looking forward to hearing from you at your earliest convenience.

My best regards

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