Reviewer's report

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

Version: 2 Date: 28 June 2012

Reviewer: Roberto Ravazzolo

Reviewer's report:

Major Compulsory Revisions
The authors present a case with a 17p13.3 rearrangement involving a region in which other rearrangements have been reported. Considering the peculiar phenotypic aspects of the patient and inheritance of the rearrangement from the mother, the Reviewer thinks that this case is worthy to be published, also to allow comparisons with other cases carrying similar rearrangements.

This family appears to suffer several environmental problems, then reference to the genetic anomaly should be discussed in term of genetic contribution to a phenotype that, especially for the mother, seems to be due to different components.

The manuscript should be extensively revised for English language and many mistakes. It is not publishable in its present form.

Minor Essential Revisions
Correction of several typing errors

Discretionary Revisions
“Genes BHLHA9, YWHAE, and CRK contribute to the phenotype of our patient” (in the abstract) is a categorical statement that should be expressed in a more hypothetical sentence.

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

Quality of written English: Not suitable for publication unless extensively edited

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' below