Reviewer's report

Title: Submicroscopic Subtelomeric Aberrations in Chinese Patients with Unexplained Developmental Delay/ Mental Retardation

Version: 1 Date: 10 February 2010

Reviewer: Agatino Battaglia

Reviewer's report:

The authors report on "Submicroscopic subtelomeric aberrations in Chinese patients with unexplained DD/MR".

Although dealing with already well known findings, I think that the paper might be of some interest to the Journal's readership.

There are, however, a number of minor essential revisions with which the authors should comply:

1) concerning the MR, were the patients formally tested? If yes, please state, in the METHODS, the tests used for the assessment.

2) in the RESULTS, pag. 7, there is 1 patient missing. The authors report on 23/24 patients. Only later, reading through the paper (particularly studying the TABLE 2), it emerges that most probably, they entered 23/24 patients (leaving out patient 1518), due to the lack of confirmation of CNV by the Affymetrix SNP array 6.0. Is this true? Please specify clearly in the text, to avoid any confusion!!!

3) from TABLE 2 it emerges that patient 419 does not have microcephaly, but, in the DISCUSSION (pag. 9, line 13) it is said that he had microcephaly!!! Please, amend accordingly.

4) from TABLE 2 it emerges that in the deleted interval of patient 419 there are 20 genes, but, in the DISCUSSION (pag. 9, line 17) it is said that "The deleted region contains 21 genes"!!! Please, amend accordingly.

5) in the DISCUSSION-Subtelomeric duplications-, pag. 13, lines 7-9, the last two sentences should be amended as follows "Our finding supports the observation made by Battaglia et al. [49], who suggested that the critical region underlying the phenotype of dup 3q is in 3q29".

6) in the REFERENCES list, references 49 and 50 should become respectively 50 and 51, and the following reference should be added as reference 49: "Battaglia A., Novelli A., Ceccarini C., Carey JC. Familial complex 3q;10q rearrangement unravelled by subtelomeric FISH analysis. Am J Med Genet 2006, 140A: 144-150.

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

Quality of written English: Acceptable
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'