Reviewer's report

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

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Reviewer: Sau W Cheung

Reviewer's report:

The manuscript entitled “Submicroscopic Aberrations in Chinese Patients with Unexplained Developmental Delay/Mental Retardation” by Wu. et al. This report is very well prepared and will add valuable information to the body of literature on microdeletion/duplication of subtelomeric region.

This reviewer has the following comments:

1. It will benefit the reader to define Mental Retardation by including IQ ranges.

2. The conclusion in the abstract needs revision. The following sentence “Four deleted subtelomeric regions and the duplicated region found in this study were smaller.....” The size of the copy number changes may not be as important as the presence or absence of a dosage sensitive gene in the region.

3. Case 1729 with a GAIN on 22q13.3 involving the SHANK gene is very interesting; more discussion on the phenotypic correlation would be helpful.

4. Please discuss case 1467 with 874Kb deletion but no phenotypic descriptions are provided.

5. Cases 1947, 1711, 2126 and 2026 appear to be cryptic unbalanced translocations. For clarification, it is important to perform FISH analysis on the patients as well as their parents.

After these minor revisions, this reviewer recommended for publication.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: Yes, but I do not feel adequately qualified to assess the statistics.