Reviewer’s report

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

Version: 1 Date: 15 February 2010

Reviewer: Pawel Stankiewicz

Reviewer’s report:

The BMC Medical Genetics manuscript “Submicroscopic subtelomeric aberrations in Chinese patients with unexplained developmental delay/ mental retardation” by Wu et al. reports the results of MLPA and SNP array (Affymetrix 6.0) analyses of the subtelomeric chromosome regions in 451 children with DD/MR from mainland China.

1) Page 4, line 9
However, microscopic techniques cannot detect microdeletions and duplications in interstitial regions or at the functional endings of the chromosomes (subtelomeric regions)
change to:
However, microscopic techniques cannot detect interstitial or terminal subtelomeric microdeletions and microduplications

2) Page 4, line 2 from bottom
These aberrant regions associated with DD/MR are likely to contain undiscovered candidate genes
Rewrite this sentence

3) Page 5, line 8
Subtelomeric aberrations were identified, their exact sizes obtained, and possible candidate genes proposed.
change to:
Subtelomeric aberrations were identified, their exact sizes were defined, and possible candidate genes are proposed.

4) Page 5, line 5 from bottom
All 451 subjects were Chinese children from the Departments of Pediatric Neurology
Provide the name of the hospital. In the affiliations, there is no such department.

5) Page 6, lines 5-6
in one of the first genes following the centromeric repeats.
This should read
in one of the genes just proximal to the telomeric repeats.

6) Page 6, line 6 from bottom
combination of a SNP array with aCGH in a single chip meant the size of each aberrant region could be accurately defined
change to:
combination of a SNP array with aCGH probes in a single chip enabled the accurate definition of the size of each aberrant region

7) Page 7, Results, lines 4-7
15pter del in 5; 4pter del in 4; 13pter del in 3
Delete the locations of short arms of acrocentric chromosomes (13pter and 15 ter). They are confusing.

8) Page 8, line 5 from bottom
deletions in 8p23.3, terminal 7q, terminal 15q and 14q32.3, as well as 22q13
Be consistent and provide the (sub)band designation for all subtelomeric regions

9) Page 9
The Authors should cite the following papers:
and

10) Page 12
This paragraph is supposed to describe “Subtelomeric aberrations smaller than previous reports”; however, the Authors discuss also patients with larger rearrangements.

11) Page 12, line 4
The terminal 14q deletion found in this patient is the smallest reported to date, and was in 14q32.33,
Delete “and was in 14q32.33,”

12) Page 12, lines 2-3 from bottom
The deleted region in patient 390 was 2.68Mb, from 4.69 Mb to 4.96 Mb,
Change to
The deleted region in patient 390 was 268 kb in size, extending from 4.69 Mb to 4.96 Mb,

13) Page 12, line 12
22q 13.33 deletion
change to
22q13.33 deletion

**Level of interest:** An article of limited interest

**Quality of written English:** Needs some language corrections before being published

**Statistical review:** No, the manuscript does not need to be seen by a statistician.