Reviewer's report

Title: Array-CGH in patients with Kabuki-like phenotype: Identification of two patients with complex rearrangements including 2q37 deletions and no other recurrent aberration

Version: 1 Date: 10 December 2007

Reviewer: Jeffrey Ming

Reviewers report:

The paper by Cusco et al describes the analysis of patients with features overlapping Kabuki syndrome by BAC array-CGH and delineates the identification of 2 patients with a 2q37 rearrangement. The paper is well-written and thorough.

Minor Essential Revisions:

1. In addition to the 2q37 deletion, one patient had a 16p11 deletion and another had a 2q36-q37 duplication. Please provide a more detailed description of the phenotype of previously described cases with rearrangements overlapping those seen in the patients in this paper so that the patients' phenotypes can be compared to previously published 16p11 deletion and 2q36-q37 duplication cases.

2. In Table 2, please clarify if the starting and ending genomic coordinates are those of the deleted (duplicated) probes or if they are the probes flanking the rearrangement. Indicating the neighboring probe that is not deleted (duplicated) is useful for assessing how large the rearrangement could be.

Discretionary:

1. The authors have generally been careful to indicate that some of the patients had "KS-like" features or clinical manifestations overlapping the KS phenotype but may not have had a clear diagnosis of KS. It would be preferable to maintain this approach throughout the manuscript and not refer to the patients as "KS patients" (p. 7) or "atypical" forms of KS (p.10).

2. The authors should indicate in the discussion which specific papers have used a whole genome array-CGH approach to assess KS patients since these papers support the authors' findings that KS is not associated with a currently detectable frequent consistent chromosomal rearrangement.

What next?: Accept after minor essential revisions

Level of interest: An article of importance in its field

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