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 November 2007

Reviewer: Mariyn Jones

Reviewer's report:

General
Cuscó and colleagues used array CGH analysis in 10 patients with features suggestive of Kabuki syndrome to explore the possibility that this disorder might be caused by a microdeletion or microduplication in the genome. Two of the ten patients were found to have complex rearrangements including a common ~4.5 Mb deletion of terminal 2q37, one of whom also had an interstitial deletion in 16p11.2. (I would agree that these two individuals do not have typical Kabuki syndrome phenotypes.) In addition, three patients with previously undescribed copy number variations were identified. The study adds support to prior investigations refuting the notion that rearrangements at 8p23.1-p22 and 20p12.1 cause Kabuki syndrome.

Although this is primarily a negative study, the authors have added new information to the literature with two further descriptions of individuals with 2q37 deletions, identification of several novel copy number variants, and more importantly further evidence that array based genome wide searches may not provide the key that unlocks the secret of Kabuki syndrome.

The paper is well written. The methodology is clearly addressed. I have no editorial suggestions.

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Major Compulsory Revisions (that the author must respond to before a decision on publication can be reached)

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Minor Essential Revisions (such as missing labels on figures, or the wrong use of a term, which the author can be trusted to correct)

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Discretionary Revisions (which the author can choose to ignore)

What next?: Accept without revision
**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.