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

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Reviewer: Maria Clara Bonaglia

Reviewer's report:

General
The paper deals with the molecular study on 16 new patients with phenotype suggestive of KS. Firstly the author exclude by microsatellite analysis the presence of 8p23.1-22 duplications in all patients. 10 patients were analysed by array-CGH analysis to search for putative microrearrangements causative for KS. The authors identified two KS-like subjects with an overlapping 2q37 terminal deletion, one of them a patient with a reciprocal translocation (15q;17q) and an additional deletion on chromosome 16p11.2.

The paper is well written. The results are well presented and discussed. The data led to further exclude the duplication at 8p23.1-22 as common cause of the KS. This study highlights the importance to perform Array-CGH analysis before taking for granted an apparently simple cytogenetic investigation with the obvious consequences on a correct clinical diagnosis, as is well represented by the case KS14. As demonstrated by the KS2 case, molecular karyotyping led to a fast and precise definition of a hidden complex chromosome rearrangement.

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

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

The authors performed array-CGH analysis using the 44k Agilent platform (Agilent, G4410B) with a resolution of about 35 Kb.

Considering that three consecutive probes are necessary to make a software call, I suggest to consider the average resolution about 100 Kb.

TAB 1
Patients KS4 : Microcephaly was indicate as “SI” but the abbreviation is not reported in the legend.
Discretionary Revisions (which the author can choose to ignore)

**What next?:** Accept after minor essential revisions

**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.

**Declaration of competing interests:**

I declare that I have no competing interest.