Reviewer's report

Title: Bronchial isomerism in a Kabuki syndrome patient with a novel mutation in MLL2 gene

Version: 1
Date: 1 December 2013
Reviewer: Fernando Scaglia

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Major Compulsory Revisions

1. The English used for this report is acceptable. However, in some areas of the manuscript, it could be improved.

2. In the background section of the abstract, I suggest using "intellectual disability" instead of "mental retardation".

3. Please use "persistent fetal fingertip pads" throughout the manuscript. Be consistent with it.

4. Make sure that throughout the manuscript the term "eversion of the lateral third of the eyelid" applies specifically to the lower eyelid as it is not used in a consistent way.

5. Please make sure that gene symbols are italicized in the manuscript.

6. Please make sure you do a thorough spell check. There are many typographical errors. For example, in the first paragraph of the clinical report, the word "trough" should be changed to "through".

7. It would of interest to the readers to learn about the growth parameters in this male patient. We are only provided with the parameters at birth but there is no information about growth parameters when he is seen by geneticists.

8. It is unclear what the authors mean by dystrophy in the physical exam.

9. Was there any asplenia or polysplenia on abdominal ultrasound?

10. The authors state that the "mutation strongly affects the physiological function of MLL2". If so, they need to provide the laboratory evidence that they have investigated the in vitro effects of this mutation.

11. The authors also state that "the mutation causes a truncated protein". Do they have any evidence for that. How about nonsense mediated decay as an alternative mechanism?

12. The authors conclude that patients with Kabuki syndrome should be investigated for defects of lung lateralization. Should this be done in a subset of symptomatic patients?
Level of interest: An article whose findings are important to those with closely related research interests

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.

Declaration of competing interests:
I declare that I have no competing interests.