Reviewer's report

Title: Rare ocular features in a case of Kabuki syndrome (Niikawa-Kuroki syndrome)

Version: 4  Date: 5 July 2014

Reviewer: Fowzan Alkuraya

Reviewer's report:

This is a very interesting case that highlights the rarely discussed overlap between Kabuki syndrome and CHARGE syndrome when colobomatous microphthalmia is observed in the former. Unfortunately, this very interesting point was completely missed by the authors who seem to be fixated on the significance of an elevated optic disc instead.

Major comments:
1- Please shift the focus of the paper to focus on the interesting overlap between Kabuki and CHARGE syndromes when the very rare finding of colobomatous microphthalmia in Kabuki syndrome is observed. This will also allow the authors to come up with a much more significant conclusion for their paper than the one in the current version.
2- What is the MLL2 gene mutation? Is it novel? Is it de novo? How did they conclude it is pathogenic?

Minor comments:
1- Please use the term intellectual disability instead of mental retardation.
2- The use of colobomatous microphthalmia in the key words is advised.
3- Change “Despite this, few reports have reported on the ocular findings that may affect vision” to “In addition, ocular findings, usually in the form of strabismus and ptosis, have been reported”.
4- Change “the boy claimed” to “the boy was claimed”.
5- I’m surprised the very obvious microphthalmia in the clinical photograph is not listed in the features observed on external examination. It is obvious without the need for examination under anesthesia.
6- If a photo consent is available it will be helpful to show the full face.

Level of interest: An article of importance in its field

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:

None