Reviewer's report

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

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Reviewer: Flavio Medina

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The submitted manuscript “Rare ocular features in a case of Kabuki syndrome (Niikawa-Kuroki syndrome)” by Yi-Hsing Chen, Ming-Hui Sun, Shao-Hsuan Hsia, Chi-Chun Lai and Wei-Chi Wu presents a case report of a patient of Kabuki syndrome with the rare ocular findings, as well as a comprehensive review of current literature. It is an article of importance in its field. However I am unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions.

Major Compulsory Revisions

In the second paragraph of case report, authors describe patient fundus as retina and choroid coloboma in both eyes, optic disc coloboma in the right eye, and an elevated optic disc without central cupping in the left eye.

We notice a bilateral posterior bulging in orbit CT. Could this left eye optic disc aspect be related to a peripapillary stafiloma, or to the colobomatous defect itself?

Minor Essential Revisions

In the third paragraph of case report, authors mention that a diagnosis of Kabuki syndrome was made after a genetic examination. Should not diagnostic criteria for Kabuki syndrome rely on the clinical observation of 5 cardinal findings?

Discretionary Revisions

Consider inserting a discussion section and moving part of conclusion section to it.

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