Reviewer's report

Title: Cardiac Conduction Abnormalities and Congenital Immunodeficiency in a Child with Kabuki Syndrome: Case Report.

Version: 1 Date: 2 February 2005

Reviewer: Kim McBride

Reviewer's report:

General
This is an interesting case report on a child with Kabuki syndrome with an unusual cardiac phenotype and immunodeficiencies. The Shone complex variant suggests a left ventricular outflow tract malformation commonly seen in Kabuki syndrome, but the long membranous ventricular septum with malpositioned AV node and bundle of His, and thymus hypoplasia are more compatible with a neural crest defect.

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

Some additional details are required. Information about the cardiac repair should be provided to allow assessment of iatrogenic causes of arrhythmias. The authors do not comment on any medications that could cause arrhythmias that may have been used, nor is there any mention if electrophysiologic studies were performed prior to pacemaker implantation. If there was delayed conduction along this lengthy track, was there evidence of a prolonged PR interval on 12 lead EKG?

I find figure 3 somewhat hard to interpret. There appears to be some increased eosinophilic material in the upper portion of the muscular septum, but at this power I cannot tell if this represents necrosis or not. If there is necrosis, would this be the cause of the bradyarrhythmias? Some labels on this figure (including a size bar) and an inset at higher power would be helpful.

Since the diagnosis of Kabuki syndrome is purely clinical, pictures of the facial features, if available and parental permission can be obtained, would be helpful.

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

Please review the abstract for apostrophe errors.

Discretionary Revisions (which the author can choose to ignore)

Given the overlapping phenotype, did the authors also perform FISH on the DiGeorge II locus on 10p12?

What next?: Unable to decide on acceptance or rejection until the authors have responded to the major compulsory revisions
Level of interest: An article whose findings are important to those with closely related research interests

Quality of written English: Acceptable

Statistical review: No

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