Author's response to reviews

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

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Author's response to reviews: see over
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Dear Editors:

First let me apologize for the delay in my response to the timely review of my article by the peer referees. Part of this delay was in attempting to obtain consent for photography from the mother of the child reported in this case report. Unfortunately, we were not able to obtain this consent. However, all other concerns by the reviewers have been addressed and are documented below:

Reviewer 1: No particular concerns noted.

Reviewer 2: 1. A photograph of the child was suggested to ensure the accuracy of the clinical diagnosis. Unfortunately, the family has given us only limited consent and has not consented to the use of a photograph of the child’s face. This child spent a majority of his life in the hospital and the only good photographs were post-mortem which was stigmatizing to this family. However, to alleviate the concern of the reviewer, as stated in the text of the article, the diagnosis was confirmed by a second independent geneticist who was blinded to my initial evaluation.

2. Attempts have been made to more concisely describe the hospital course with the article being shorter than the original.

3. The reviewer reports that abdominal wall defects are not common in Kabuki syndrome. That fact is correct. However, according to the article referenced by Donadio in the American Journal of Medical Genetics, diaphragmatic and abdominal wall defects are seen with a greater incidence in non-Asian patients than in the original descriptions of this syndrome.

Reviewer 3: 1. The cardiac repair was limited to correction of the coarctation of the aorta. No surgical manipulation of the cardiac septum was performed so that iatrogenic causes of arrhythmia would not be considered a high likelihood.

2. The reviewer makes an excellent point with regards to medications which may precipitate arrhythmias. Further review of the chart shows that this child was initially on lidocaine after his first episode of bradyarrhythmia. Lidocaine is known to be arrhythmogenic. However, lidocaine was not continued beyond the initial episode. His cardiac medication during his subsequent episodes included only furosemide which is not associated with arrhythmia. His electrolytes were normal during the episode of bradyarrhythmia and asystole which resulted in his death.
3. An electrophysiologist was consulted after the initial episode of bradyarrhythmia. There were no EKG changes consistent with delayed conduction as reviewed by 4 different cardiologists who managed his care during his hospitalization. His health was fragile enough that EP studies were not conducted prior to pacemaker placement.

4. Figure 3 has been changed to include only the VVG stain at higher power. Further review by the pathologist does not indicate any areas of necrosis. A size bar is placed for reference.

5. With regards to the photograph of this child please see the response to reviewer 1.

6. The child was studied by FISH analysis for the 22q and 10p loci for VCFS syndrome with negative results.

Sincerely,

Maulik R. Shah