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IMAGING VIGNETTE

CLINICAL VIGNETTE

Type A Aortic Dissection in a 24-Year-Old Patient With Kabuki Syndrome

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ABSTRACT

Our case report documents the first type A aortic dissection in a patient with Kabuki syndrome (KS) and emphasize the need for intensive cardiovascular risk monitoring in patients with KS. It stresses the importance of further research to establish a correlation and awareness for patients with KS. (J Am Coll Cardiol Case Rep 2024;29:102149)
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Kabuki syndrome (KS) is a rare genetic disorder affecting multiple organs. Around 70% of the patients have cardiovascular malformations. Coarctation of the aorta is the most common, but bicuspid aortic valve and aortic dilatation can also occur.[1] Aortic dissections were never documented in patients with KS before. A case report, however, did describe a carotid dissection in a patient with KS.[2]

CASE PRESENTATION

A 24-year-old male patient underwent magnetic resonance imaging for shoulder pain. Surprisingly, it showed a type A aortic dissection. This diagnosis was 5 weeks after a COVID-19 infection for which patient underwent computed tomography. This scan showed stable aortic diameters and no aortic dissection. Further evaluation at our tertiary center confirmed the type A aortic dissection, which spanned from the root to the aortic arch with involvement of the right coronary artery (**Figure 1**).

The patient's medical history was complex, with cognitive developmental delay, coarctation of the aorta, bicuspid aortic valve, hypoplastic arch, ureteral issues, and genetically confirmed diagnosis of KS (pathogenic *KMT2B* mutation). His surgical history included an extended end-to-end anastomosis for coarctation of the aorta as an infant and aortic arch reconstruction at the age of 17 years. The latter procedure was complicated with a tamponade and sternum instability due to infection, necessitating multiple surgeries.

The patient had stable aortic diameters during recent follow-up (**Table 1**). A computed tomography scan 16 months before this presentation reported a sinus diameter of 39 mm (index of 2.8 cm/m^2). Despite being

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ABBREVIATIONS AND ACRONYMS

KS = Kabuki syndrome

below intervention thresholds, a potential procedure was discussed.[3] Prior surgical experiences, however, made the patient, physician, and family reluctant.

MANAGEMENT

Both surgical and conservative strategies were discussed with the patient and his family. Given the subacute nature of the type A dissection, potential surgical challenges and comorbidities, shared decision for a conservative approach was made. Unfortunately sudden bradycardia and shock, probably due to right coronary artery occlusion, led to the patient's demise.

DISCUSSION

This case describes the first recognized type A dissection in a patient with KS. Considering the European Society of Cardiology guidelines, this patient did not meet the surgical criteria based solely on aortic diameters.[3]

Patients with KS often exhibit cardiovascular anomalies paralleling Turner syndrome. For patients with Turner syndrome, there are specific guidelines that suggest aortic surgery when the indexed aortic diameter exceeds 2.75 cm/m^2.[3] Some international guidelines advocate intervention at even smaller diameters.[4-6] The indexed diameters of our patient exceeded these thresholds. This underlines the importance of considering patient height in aortic diameter evaluation. Whereas patients with KS might naturally be predisposed to aortic dissections, a correlation until now remained unconfirmed due to its rarity. In syndromic cases with significant aortic anomalies and shorter stature, like KS, current guidelines should be employed cautiously, emphasizing the role of patient height in aortic diameter evaluation.

CONCLUSIONS

KS requires intensive cardiovascular monitoring. This case report raises the awareness for the risk of aortic dissection in relative small diameters of the aorta. Guidelines such as those for Turner syndrome might be worth considering for patients with KS.

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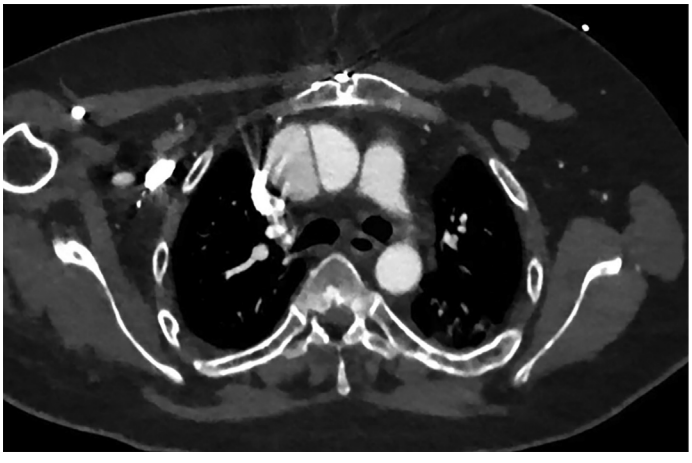
FIGURE 1 CT Scan Showing Type A Aorta Dissection in Patient With KS



A transversal coupe of a computed tomography (CT) scan showing a type A aorta dissection in a 24-year-old patient with Kabuki syndrome (KS).

TABLE 1 Measurements of Aorta Diameters Prior to a Type A Aorta Dissection

Aorta Diameters	41 mo Prior to Type A Aorta Dissection	33 mo Prior to Type A Aorta Dissection	16 mo Prior to Type A Aorta Dissection
Aorta sinus	29 × 38 × 33	29 × 38 × 35	33 × 39 × 38
Ascending aorta	27	28	30
Aortic arch	19	22	20
Descending aorta	15	16	16
Aorta diaphragm level	12	13	14
Abdominal aorta (renal arteries level)	Unknown	9	9
Abdominal aorta proximal to bifurcation	Unknown	9	9

Measurements are millimeters.

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KEY WORDS bicuspid aortic valve, cardiovascular abnormalities, coarctation of the aorta, Kabuki syndrome, Turner syndrome, type A aortic dissection