Incidental finding of severe aortic root dilation and aortic insufficiency in a child with undiagnosed connective tissue disorder

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BACKGROUND

- 11-year-old male was incidentally found to have an aortic aneurysm with a severely dilated aortic root, severe aortic insufficiency and extensive aortic to aorto-pulmonary collateral vessels resulting in a "left to left" shunt.
- It was subsequently discovered that the patient had a strong family history of connective tissue disorders.
- Patients with connective tissue disorders are at a high risk for complications in the perioperative period, including aortic root dilation, aneurysms, aortic valve insufficiency, and aortic dissections, as well as cerebrovascular events and sudden death.[1-2]
- This case is unique because of the urgent management and anesthetic considerations required to address the aortic pathology and resultant valve insufficiency, but also due to the nature of how this child presented despite a known, a strong family history of probable connective tissue disorder.

CASE DESCRIPTION

- The patient presented with a left wrist fracture from a football injury and was found to have a pathologic diastolic murmur.
- Transthoracic echocardiogram showed severely dilated aortic root and annulus with severe aortic insufficiency..
- Chest CT showed large ascending aortic aneurysm and multiple aorto-pulmonary collaterals.
- He had extensive family history with autosomal dominant pattern of severe aortic dilation and aneurysm requiring root replacement and systemic artery to pulmonary collaterals, as well as systemic arterio-venous malformations.
- There was no definitive genetic testing, but probable connective tissue disorder now termed "Familial Multiple Organ Arterial Ectasia with Massive Hemoptysis" in a case report of his family.[3]
- MRI additionally confirmed multiple collateral arteries arising from descending aorta that branched in the hilar region of both lungs creating a left to left shunt estimated at 53% of the net ascending aortic flow.
- Prior to operative repair of aortic root and aneurysm, he underwent cardiac catheterization for coiling of multiple aorto-pulmonary collateral vessels to reduce risk of bleeding during cardiopulmonary bypass..
- Ultimately, the patient underwent a valve sparing aortic root replacement with 28 mm Hemashield graft and coronary re-implantation. Of note, the surgeon identified a contained dissection in the root. He had an unremarkable post-operative course and was discharged on post operative day #3.

RESULTS

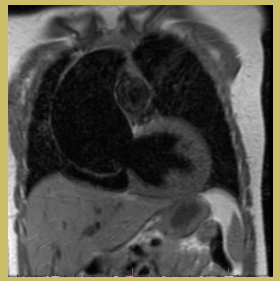

Figure 1. Coronal view on MRI of severely dilated aortic root and dilated aorta.

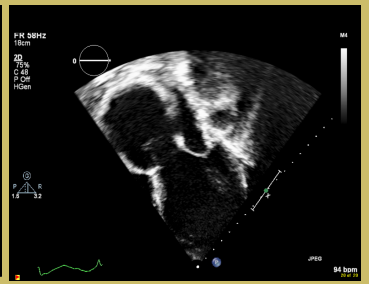

Figure 3. Still frame of the dilated ascending aorta and aortic root compared to the heart on transthoracic echocardiogram in an apical five chamber view.

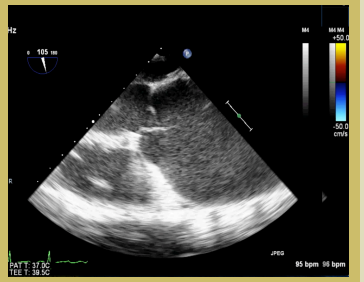

Figure 5. The relative diameter of the aortic root in comparison to the valve as well as effacement of sinotubular junction.

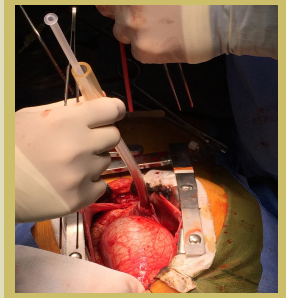

Figure 2. Intraoperative photograph of dilated aorta during aortic cannulation.

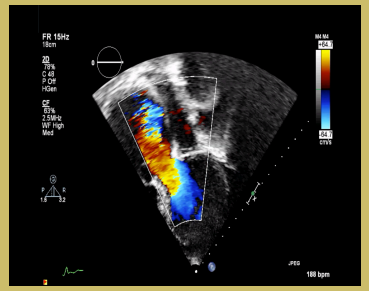

Figure 4. Still frame of the dilated ascending aorta and aortic root with color doppler over the aortic valve, showing severe aortic regurgitation on transthoracic echocardiogram in an apical five chamber view.

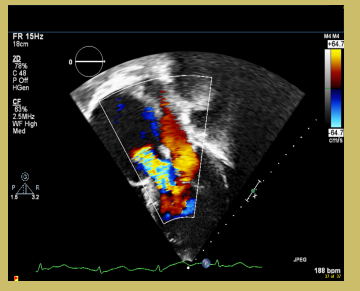

Figure 6. Transesophageal echocardiogram long axis view with color Doppler demonstrating less severe aortic insufficiency with decreased afterload under anesthesia.

DISCUSSION

- The case described is remarkable due to the severity of aortic root dilation and aneurysm serendipitously found after sustaining a left wrist fracture, leading to urgent management requiring anesthesia for multiple procedures..
- This patient was at risk for sudden death from the dissection was well as worsening aortic regurgitation.
- The anesthetic management for patients with connective tissue disease includes consideration of bleeding tendency, risk of aortic rupture or dissection, and hemodynamic management of severe aortic insufficiency.
- Blood pressure and heart rate must be tightly controlled with the primary aim to avoid hypertension.. An arterial line is recommended even for non cardiopulmonary bypass.[4]
- Blood products should be available for most cases.[4]
- This case underscores the importance of a obtaining a full history, including family history, and complete physical exam even in routine procedures.
- In this case, astute physicians diagnosed a potentially fatal disease, where standard management, including any hypertensive episodes, for an orthopedic surgical case could have had devastating consequences, including death, for this patient.

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