Cardiology in the Young

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Out of the blue: inflammatory myofibroblastic tumour identified during repair of tetralogy of Fallot with absent pulmonary valve

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Brief Report

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Abstract

Inflammatory myofibroblastic tumour of the heart is an exceedingly rare benign neoplasm. While benign, without prompt management its impact can be devastating. Tetralogy of Fallot with absent pulmonary valve is a rare form of CHD. We present the first documented case of inflammatory myofibroblastic tumour of the heart in the presence of tetralogy of Fallot with absent pulmonary valve.

Introduction

Primary cardiac tumours are rare in infants, with an incidence of 0.13%.[1] The most common include rhabdomyoma, myxoma, fibroma, and teratoma.[2,3] Cardiac inflammatory myofibroblastic tumour accounts for <5% of all primary cardiac tumours.[2] It is typically considered a low-grade neoplasm; however, left untreated intracardiac tumour can lead to significant morbidity and mortality.

Tetralogy of Fallot with absent pulmonary valve is a rare variant characterised by right ventricular hypertrophy, malaligned ventricular septal defect, overriding aorta, absent or hypoplastic pulmonary valve leaflets, and resultant pulmonary insufficiency in utero. This creates pulmonary arterial dilation and can compromise the airways, leading to broncho- or tracheomalacia. Tetralogy of Fallot with absent pulmonary valve is a rare form of CHD that affects 0.002–0.004% of babies born in the United States each year.[7,8] We present the first documented case of a patient with both tetralogy of Fallot with absent pulmonary valve and cardiac inflammatory myofibroblastic tumour.

Case description

We present a female infant born at 39 weeks with tetralogy of Fallot with absent pulmonary valve. Prenatal echocardiography revealed significant pulmonary stenosis, free pulmonary insufficiency, and severely dilated main and branch pulmonary arteries. This was confirmed postnatally. A 3/6 harsh systolic ejection murmur heard loudest at the left upper sternal border as well as a 1-2/4 diastolic murmur were noted on exam. Initial oxygen saturations were in the 70% range with minimal improvement with non-invasive support, so the patient was intubated. CT showed no significant airway compression.

The infant was followed closely in outpatient cardiology clinic and continued to grow and develop well the first 2 months of life. Parents reported occasional noisy breathing but denied hypoxic spells, dyspnoea, diaphoresis, or difficulty feeding. Home pulse ox readings were consistently >90% SpO_2. The infant was evaluated by otolaryngology for noisy breathing and was not found to have any signs of airway obstruction. Surgical repair with ventricular septal defect closure and transannular patch was scheduled for 2.5 months of life as she was greater than 4 kg, with severe pulmonary stenosis at the annulus.

Intraoperative transesophageal echocardiogram revealed findings concerning for a mass versus clot in the right atrium (Supplementary Video 1). The patient was placed on cardiopulmonary bypass, and the right atrium was carefully accessed. Upon direct exposure, a mass was attached to the right atrium between the appendage and the superior caval vein requiring a small patch of atrial wall be excised to fully remove the mass. The atrium was repaired with autologous pericardium. The ventricular septal defect was closed with a Gore-tex patch, and the right ventricular outflow tract obstruction was addressed with a transannular patch and resection of infundibular muscle. The patient's CT at birth, thorough exam with our otolaryngology colleagues, and echocardiographic measurements of branch pulmonary arteries

with Z score between +3 and +4 did not suggest a need for plication or other manoeuvre to decrease the size of the branch pulmonary arteries as is sometimes required in this variant.

Histologic examination of the tumour was performed by both our institution and an outside consultant. The mass is described as polypoid, well circumscribed, moderately cellular, and composed of spindle cells with fusiform nuclei, small nucleoli, and eosinophilic cytoplasm. There are scattered mitoses of normal configuration, along with foci of necrosis. Neither atypical mitoses nor cytologic atypia are present. Immunohistochemistry demonstrates diffuse positive expression of actin and focal expression of epithelial membrane antigen. Stains for ALK1, cytokeratin cocktail, desmin, MYOD1, myogenin, CD30, CD31, CD34, CD163, and HHV8 are negative. The proliferation rate is 45%, as estimated from the Ki-67 nuclear labelling index.

On review of the prior echocardiograms, the mass was not evident on the study at birth, and the most recent study at 1 month of life was limited by patient movement. In retrospect, the mass was present in one clip but smaller and appeared to be a part of the atrial wall. The patient recovered smoothly from surgery and was discharged on post-operative day 3. She is now 15 months old with free pulmonary insufficiency, evidence of normal right ventricular pressures, and excellent biventricular function.

Discussion

Tetralogy of Fallot with absent pulmonary valve is a rare variant. Interestingly, only 50% of infants born with tetralogy of Fallot with absent pulmonary valve present with respiratory symptoms, and severity ranges from stridor to respiratory failure. Those without respiratory symptoms can be closely followed by outpatient cardiology until elective surgical repair at 4 to 6 months, similar to other tetralogy of Fallot patients. Cardiac inflammatory myofibroblastic tumours are rare without a clear predilection for a specific atrium, ventricle, or valve. Cardiac inflammatory myofibroblastic tumours do not metastasize or invade the heart and have the potential for regression both spontaneously or in response to steroids. The presenting symptoms are closely related to the size and location of the tumour and may present similarly to CHD. Symptoms commonly reported in infants include respiratory distress, diaphoresis, cyanosis, murmur, tachycardia, and decreased oral intake. Cardiac inflammatory myofibroblastic tumour is a difficult diagnosis to make and requires histological examination for definitive diagnosis. Histologically, it is characterised as a polypoid mass with spindle cells in a myxoid stroma consisting of lymphocytes and occasional plasma cells. Immunohistochemical analysis demonstrates that tumour cells express actin and vimentin, inconsistently express epithelial membrane antigen, CD31 and CD163, and do not express ALK1, S100, or CD34.[1–6]

Whenever possible, complete resection of cardiac inflammatory myofibroblastic tumour is the definitive treatment.[6] When complete resection is not an option, partial resection while preserving cardiac function is the goal. Alternative or complementary treatments to partial resections included steroids, radiation therapy, chemotherapy (protein kinase inhibitor), and heart transplant. There is only one documented recurrence after resection, with no re-recurrence 18 months after repeat resection.[3] Failure to treat these benign neoplasms can be catastrophic due to direct coronary invasion, venous thromboembolism, or sudden death.[3,4] When managed surgically, cardiac inflammatory myofibroblastic tumours have a very favourable outcome.

Conclusion

This is the first documented case of a patient with both tetralogy of Fallot with absent pulmonary valve and cardiac inflammatory myofibroblastic tumour. Had our patient's CHD been less severe, surgical management of her CHD would have occurred later, and the cardiac inflammatory myofibroblastic tumour would have likely gone unnoticed until it progressed enough to cause symptoms.

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Competing interests. None.

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