CLINICAL VIGNETTE

A Hole lot of Heart

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A 64-year-old male with BPH, hyperlipidemia, hypertension, and urinary retention was admitted with a complicated urinary tract infection. He had a foley urinary cathether placed 1 month prior to admission. In subsequent weeks, he developed suprapubic pain radiating to both of his flanks.

His past medical history was notable for a "hole in his heart", with no prior interventions or therapies. Review of systems was notable for low functional capacity at baseline with self-limited activity since childhood. He denied dyspnea at his current level of activity.

Physical exam included normal temperature, heart rate of 72, blood pressure 110/70, and an oxygen saturation of 92% on room air. His cardiac exam was remarkable for an RV heave, loud P2, a 2/6 holosystolic murmur at the left lower sternal border (LLSB) that increased with inspiration, and a soft 1/6 diastolic decrescendo murmur at the left upper sternal border (LUSB). JVP was 7 cm. Lungs were clear to auscultation bilaterally without wheezes. There was mild suprapubic tenderness to palpation without CVA tenderness. Extremities were free of clubbing, cyanosis or edema.

Basic metabolic panel and CBC were normal. UA was remarkable for many WBCs, bacteria, nitrites and leukocyte esterase.

CXR revealed cardiomegaly without focal consolidations or edema. EKG has right axis deviation, right atrial enlargement, and a prominent R wave in lead V1. CT angiogram revealed subacute and chronic pulmonary emboli. Transthoracic echocardiogram showed borderline LV systolic function with ejection fraction of 50 to 55%, moderately dilated right atrium, right ventricular hypertrophy (RVH), mild-moderate tricuspid regurgitation and pulmonic regurgitation, severe pulmonary hypertension, and a suggestion of a VSD with left to right shunt. Transesophageal echocardio-gram revealed an inlet VSD 1.7 cm in diameter, with left to right shunting during systole and bidirectional shunting in diastole. The velocity across the VSD in systole was approximately 2 m/sec. His systolic blood pressure was 90 mmHg, and the estimated pulmonary artery systolic pressure was 70 mmHg based on echocardiography. The pulmonary artery was dilated at 36 mm. The ratio of the blood flow in the pulmonary circulation (Qp) to the blood flow in the systemic circulation (Qs) was estimated at 2.5:1.

The clinical and pathophysiology features in this case are fascinating and provide a few pearls to clinicians. The clinical exam shows evidence of pulmonary hypertension and right ventricular hypertrophy, most notably the RV heave and loud P2 component of S2. His EKG provides support for RVH with a prominent R wave in V1, right axis deviation and right atrial enlargement. The sensitivity of these EKG criteria for RVH is 20-40%, but the specificity is quite high, around 90%[1]. His cardiac murmurs probably represent tricuspid regurgitation (2/6 holosystolic murmur at LLSB that increases with inspiration, also known as Carvallo's sign) and pulmonic regurgitation (1/6 diastolic decrescendo blowing murmur at LUSB, also known as the Graham-Steell murmur). The VSD murmur in this patient is not prominent as larger defects with small pressure gradients (near equalization between LV and RV pressures) and velocities are usually not audible. In general, smaller VSDs cause louder high-frequency holosystolic murmurs because of the pressure gradient between the left and right ventricles. However, as pulmonary hypertension develops over time, the pressure gradient between the two chambers decreases and the murmur may soften or become inaudible[2].

Equally interesting in this patient are the physical exam findings that are absent given his age, size of his VSD, and the significant pulmonary hypertension and RVH. Notably, he has not developed Eisenmenger's syndrome, whereby the shunting of blood is reversed (right to left) taking deoxygenated blood to the systemic circulation and leading to central and peripheral cyanosis, and—in many cases—clubbing of the extremities[2,3]. And although his RV is hypertrophied, it has not yet failed so elevated jugular venous pressures, hepa-tomegaly, ascites, and lower extremity edema are not present.

What's the primary culprit for his pulmonary hypertension?

The WHO has classified pulmonary hyper-tension into five groups on the basis of mechanisms[4]:

1. Pulmonary arterial hypertension (idiopathic, collagen vascular disease, portal, congenital left-to-right shunts, HIV)
2. Pulmonary hypertension with left-heart disease
3. Pulmonary hypertension associated with lung disease and/or hypoxemia
4. Pulmonary hypertension associated with chronic thromboembolic disease
5. Miscellaneous (tumors, sarcoid, fibrosing mediastinitis, etc.)

Our patient has a mixed picture of pulmonary hypertension given his congenital heart defect and lifelong shunt through his VSD (group 1) as well as subacute and chronic thromboembolic disease (group 4). Mechanistically, with a large VSD and left to right shunt, the RV dilates to accommodate the increased volume of blood. This increased blood flow leads to pathologic changes in the pulmonary vasculature leading to medial hypertrophy and intimal fibrosis (among other changes) and eventual pulmonary hypertension[5,6]. At initial presentation, it is impossible to determine what component of his pulmonary hypertension is from thromboembolic disease and how much is from his chronic left to right shunt through the VSD. By definition, pulmonary hypertension from chronic thromboembolic disease persists 6 months after diagnosis and treatment of pulmonary embolism[5], requiring re-evaluation before proceeding with further interventions and/or therapies.

To repair or not to repair?

VSD is a common congenital defect in newborns, occurring in approximately 3 infants per 1000 live births[7]. However, we rarely see this defect in our adult population because nearly half close spontaneously during childhood and most large defects are already surgically corrected. In this patient, the decision to close the VSD or pursue medical management will be based on the extent of the shunt and the presence of irreversible pulmonary hypertension. The most recent ACC/AHA guidelines recommend closing a VSD when the pulmonary-to-systemic blood flow ratio (Qp:Qs) is greater than 2 and there is clinical evidence of volume overload, or if the patient has a history of infective endocarditis[7]. Closure is not recommended in patients with severe irreversible pulmonary hypertension. Our patient will need to undergo right heart catheterization before any consideration of VSD closure. The decision will be based on: (1) the extent of his pulmonary hypertension, (2) any reversibility in his pulmonary hypertension (tested with vasodilators), and (3) the Qp:Qs ratio. After several months of treatment with anticoagulants for his pulmonary emboli, his physicians will have to determine if the "hole in his heart" can be corrected.

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