Is that SAM?
A Case of Hypertrophic Cardiomyopathy AND Pulmonic Stenosis in a Patient with Noonan Syndrome

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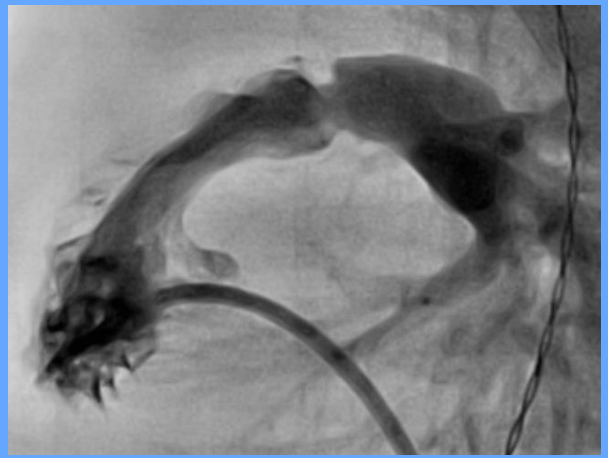
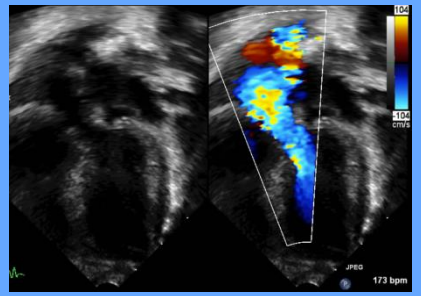
BACKGROUND

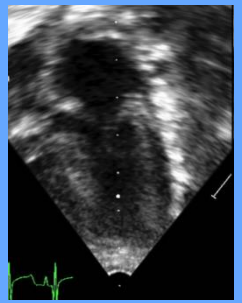
Noonan syndrome is a genetic disease with an incidence of 1:1000 to 1:2500 that is characterized by facial anomalies, short stature, and congenital heart defects. Noonan syndrome follows an autosomal dominant inheritance pattern with variable expression. It is one of the most common genetic disorders associated with congenital heart defects, second only to trisomy 21. Approximately 65-80% of patients will have a cardiac anomaly, with pulmonic stenosis and hypertrophic cardiomyopathy being the most common, but rarely are these two findings found in the same patient. PS in these patients is associated with a thickened, dysplastic valve that is not usually amenable to transcatheter balloon dilatation as was evidenced in our patient. Hypertrophic cardiomyopathy affects the ventricular septum primarily although the ventricular free walls may also be involved.

CASE DISCUSSION

Our patient's initial echocardiogram revealed mild bilateral branch pulmonary artery (PA) stenosis with normal proximal branch PA diameters, mild RPA stenosis, qualitatively normal biventricular systolic function, no significant valvar pulmonary stenosis, and no evidence of hypertrophic cardiomyopathy (HCM). Following a routine cardiology appointment at 4 months of age, the patient was noted to be floppy with perioral cyanosis, acutely became bradycardic, and desaturated. After resuscitation, an echo revealed progressive moderate pulmonary valvar stenosis with peak gradient across the pulmonary valve of 68mmHg (mean of 40mmHg), a mildly prominent muscle bundle in the right ventricular outflow and mild RVH with normal systolic function. The patient underwent a cardiac catheterization which showed a thickened pulmonic valve with additional supravalvar narrowing (Figure 1). Balloon dilation of her congenital subvalvar PS was unsuccessful and the decision was made to proceed with surgery. She underwent an uncomplicated pulmonary valvotomy with transannular patch that extended to the LPA. Two weeks after her cardiac surgery, mild dynamic left ventricular outflow tract (LVOT) obstruction secondary to systolic anterior motion (SAM) of the mitral valve chordae and HCM was noted on follow-up echocardiogram which was an entirely new finding.







CONCLUSION

In summary, patients with Noonan syndrome have the potential to develop hypertrophic cardiomyopathy despite earlier echocardiograms that do not demonstrate this abnormality. Cardiology follow up is vital in this patient population to ensure stable cardiac function even in patients with previously corrected congenital cardiac defects. Systolic anterior motion of the mitral valve can be life-threatening under anesthesia. Commonly used anesthetic agents act as vasodilators which decrease preload and may cause reflex tachycardia, together worsening dynamic left ventricular outflow tract obstruction. Inotropes that may be used to treat this hypotension increases the displacement of the mitral valve into the LVOT creating further cardiovascular collapse.

Anesthetic Hemodynamic Goals

- Minimize sympathetic stimulation
- Maintain preload and afterload
- Decrease contractility while maintaining cardiac output
- Phenylephrine and Beta Blockers are therefore useful agents

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