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Case Report

Unusual Vascular Manifestations of Noonan Syndrome

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ABSTRACT

Noonan syndrome is a relatively common autosomal dominant disorder. We present a patient with Noonan syndrome and multiple cardiac and vascular manifestations, some of which are unusual and rarely reported. Further research is necessary to determine whether these defects are truly secondary to Noonan syndrome or possibly another underlying congenital abnormality.

Key words: Noonan Syndrome, Coronary Artery Aneurysm, Solitary Coronary Artery.

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INTRODUCTION

A 25 year-old male with a history of Noonan's Syndrome diagnosed at an outside facility, at an early age with a remote history of pulmonic valve replacement was admitted for worsening dyspnea on exertion and pulmonary hypertension. The patient was normotensive with developmental delay, severe S-shaped scoliosis and short stature. Physical exam revealed a faint, short 2/6 systolic ejection murmur. Laboratory test values demonstrated a prothrombin time (PT) of 16.7 seconds, otherwise, lab values were noncontributory.

He was admitted to the Progressive Care Unit (PCU) after having an echocardiogram that demonstrated moderate dilatation of the right heart with severe pulmonary artery hypertension (pressure gradient of 85 mmHg). The right ventricular outflow tract was narrowed with dilatation of the pulmonary trunk and branch pulmonary arteries. A single coronary artery was identified originating from the left coronary sinus.

Cardiac MRI demonstrated right ventricular enlargement with leftward bowing of the intraventricular septum, enlarged main pulmonary artery, and valvular insufficiency of the aortic, tricuspid, and mitral valves (Figures 1A and 1B). There was no delayed myocardial enhancement and normal biventricular systolic function.

Coronary computed tomographic angiography (CCTA) was performed, showing dilation of the main and proximal branch pulmonary arteries. Multiple lobar and segmental pulmonary arterial aneurysms and strictures were also present (Figures 2A and 2B).

The aortic root was dilated measuring 4.1 cm in diameter. Multiple collaterals were seen originating from the descending aorta. A solitary coronary artery originated from the left coronary sinus, giving origin to the left anterior descending (LAD) and left circumflex coronary arteries. The proximal LAD was dilated measuring 8.22 mm in diameter. A dominant left circumflex artery supplied the posterior descending artery and posterior left ventricular branch. The right coronary artery originated from the mid LAD, supplying the right ventricular outflow tract and right ventricle (Figure 3).

Anomalies of the hepatic venous circulation were also identified. The right portal vein was hypoplastic as well as the right hepatic lobe. A fistula between the inferior vena cava and left portal vein was identified, probably a remnant ductus venosus. The main and left portal veins were dilated.

DISCUSSION

Noonan Syndrome is a relatively common disorder, seen in 1 in every 1000 to 2500 live births.[1-5] It is an autosomal dominant disease with variable expressivity and no sex or race predilection.[1-4] Typically, it is easy to observe, as there are several distinct physical features associated with the disease. These features are similar to Turner syndrome, including shortened stature and neck webbing.[1-5] However, there are some facial dimorphisms unique to Noonan Syndrome, such as a deep philtrum and low-set ears.[1-3-5] Like Turner syndrome, there is also the possibility of

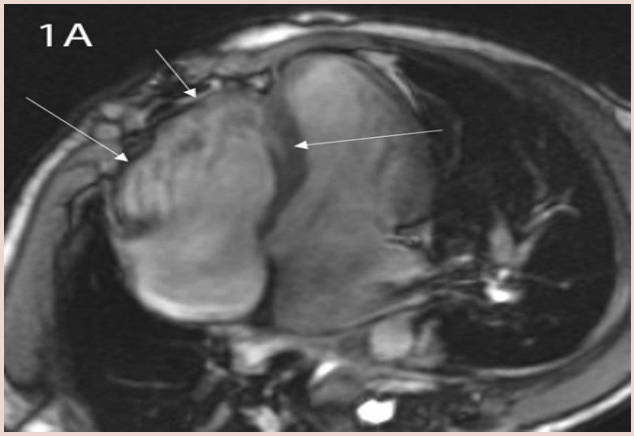

Figure 1a: Bright blood cardiac MR in a 4-chamber projection shows dilation of the right ventricle and right ventricular hypertrophy with septal bowing (thin white arrows).

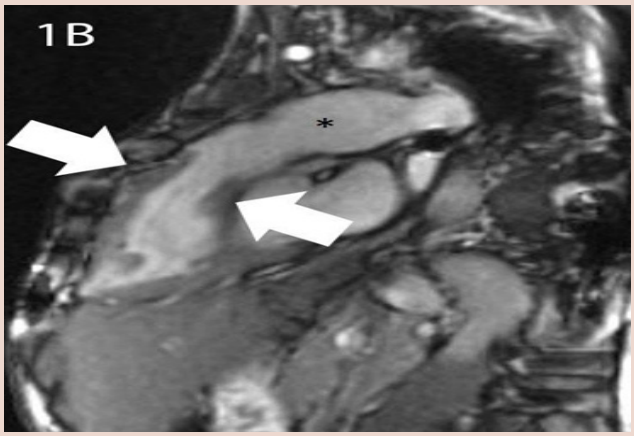

Figure 1b: Another bright blood cardiac MR image demonstrates the RV outflow tract obstruction (thick white arrows) with dilation of the pulmonary trunk (*).

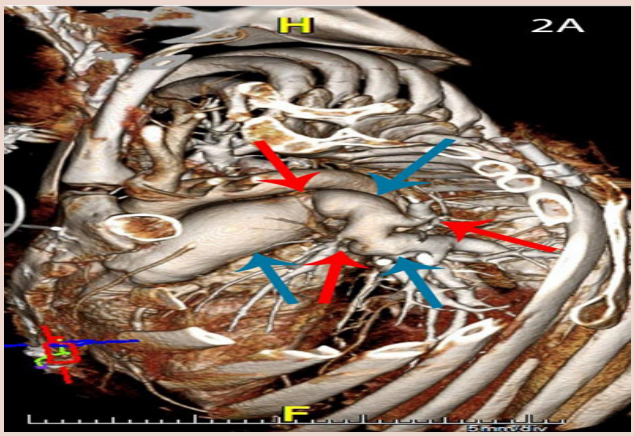

Figure 2a: Volume rendered CT angiogram of the chest shows a tortuous left pulmonary artery and upper lobe branches with multifocal stenosis (red arrows) and aneurysms (blue arrows).

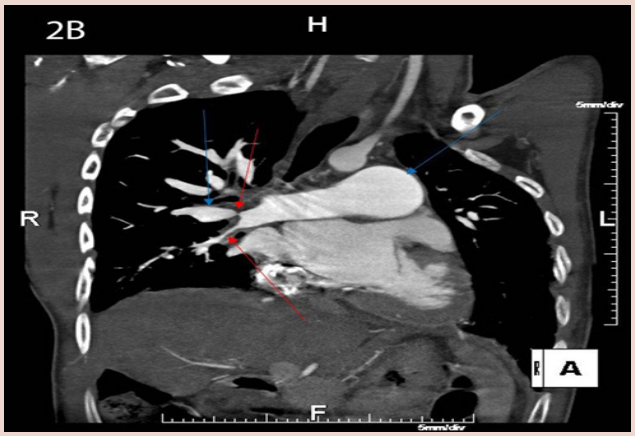

Figure 2b: Maximal intensity projection CT angiogram showing similar findings in the right lung (red and blue arrows).

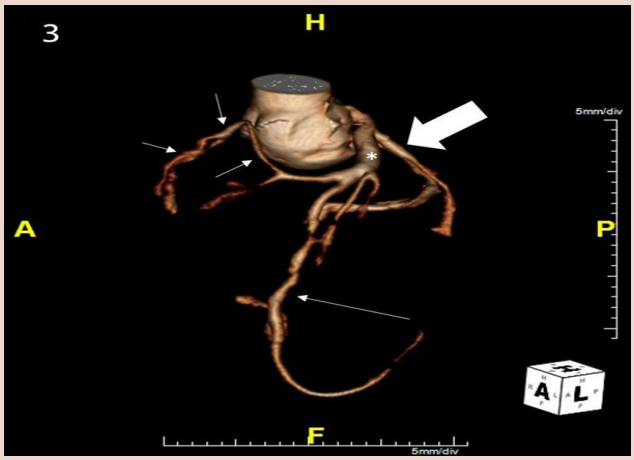

Figure 3: Volume rendered cutaway CT angiogram image of the aortic root and coronary arteries. Note the solitary coronary artery originating from the left coronary sinus giving rise to the left anterior descending (long thin white arrow), left circumflex (thick white arrow) and right coronary (short thin white arrows) arteries. The proximal LAD is ectatic (*). Motion artifact is noted in the mid LAD.

mental or learning disabilities depending on its severity.[2-3-5] Diagnosis can be confirmed through genetic testing in about 70% of cases.[1-3]

It is important to identify those with Noonan syndrome early, as there is a 50%-90% frequency of congenital heart defects in these patients. The most common defect (50%-62% of patients) is valvular pulmonary stenosis, often including valve dysplasia. Another commonly associated defect is hypertrophic cardiomyopathy (20%-30%).[1-5] Though these two are the most common, other defects can be present such as coarctation of the aorta, abnormal systemic and pulmonary venous return, atrial septal defects, fistulas, dilatations, and (rarely) coronary artery anomalies.[1,2-4-6] To date, there have only been a handful of reports of coronary artery anomalies and ectasias in Noonan children (4,7-12). There has only been one previous case reported of a Noonan adult presenting with bilateral coronary ectasia detected on CCTA.[13]

CONCLUSION

While congenital heart defects are common in this condition, our patient shows a remarkable array of rare vascular abnormalities. Further investigation is necessary to determine whether these defects are truly secondary to Noonan syndrome or possibly another underlying congenital abnormality. If these defects are associated with Noonan syndrome, then there may be need for adjustment of current management practices to better identify and address these anomalies in future patients.

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CONFLICT OF INTEREST

No honorarium, grant, or other form of payment was given to anyone to produce the manuscript.

ABBREVIATIONS

PCU: Progressive Care Unit; CCTA: Coronary computed tomographic angiography; LAD: left anterior descending artery.

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