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An Unusual Congenital Heart Disease in a Patient with Noonan Syndrome: Isolated Parachute-like Asymmetric Mitral Valve

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Abstract Noonan syndrome is an inherited disorder that involves various organs, such as dysmorphic facial features, cardiac defects, and cryptorchidism, among others. Its pathophysiology is linked to germ-line mutations in the RAS/mitogen-activated protein kinase (MAPK) pathway. The most frequent cardiac defect in this syndrome is pulmonary valve stenosis. Conversely, mitral valve involvement is rare, and a parachute-like asymmetric mitral valve presentation is even rarer. In cases where surgery is necessary, mitral valve repair is preferred over mitral valve replacement. Here we present a case of a 20-year-old gentleman with Noonan syndrome who was found to have a parachute-like mitral valve evidenced by a 2D-Echocardiogram.

Keywords: *Noonan syndrome, MAPK pathway, cardiac defects, parachute mitral valve, parachute-like asymmetric mitral*

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1. Introduction

This is a 20-year-old Dominican Republic male with a past medical history of Noonan syndrome with an unspecified congenital heart defect, intellectual disability, and history of CVA without residual deficits at the age of 15, epilepsy, hypothyroidism, and infective endocarditis who recently moved to the US in November 2021 and presented to our outpatient clinic for establishing primary care. Upon presentation, the patient denied chest pain, shortness of breath, dizziness, palpitations, reduced exertional tolerance, or focal weakness. The patient's mother stated that cardiac surgery was offered at some point, however, because of his intellectual disability he was precluded to undergo any cardiac surgery.

The patient was hemodynamically stable with a blood pressure of 108/65 mmHg, heart rate of 75 beats per min, respiratory rate of 20 breaths per min, and saturating 98% on room air. On physical examination, the patient had typical facial features of Noonan syndrome characterized by a high anterior hairline, triangle-shaped head, and prominent nasolabial folds. Furthermore, the patient had a short stature for his age. On cardiac auscultation, a 4/6 systolic murmur and a 2/6 systolic murmur was appreciated in the pulmonic and tricuspid region, respectively. The rest of the physical exam was grossly unremarkable.

The initial set of labs revealed a TSH level of 1.64 [0.34 – 5.6 uIU/ML], free T4 of 0.70 [0.61 – 1.12 ng/dl], the rest of the lab work including complete blood count, and the comprehensive metabolic panel was unremarkable. EKG showed normal sinus rhythm with sinus arrhythmia, right atrial enlargement, and early repolarization changes. A 2D-Echocardiogram revealed fibroelastic deficiency of the valves and changes consistent with a parachute-like mitral valve (Figure 1).

Since the patient had no signs or symptoms of mitral valve dysfunction, he was instructed to follow up closely for periodic transthoracic echocardiograms (TTE) to monitor the mitral valve and to seek medical attention if he develops symptoms such as shortness of breath, chest pain, or dizziness.

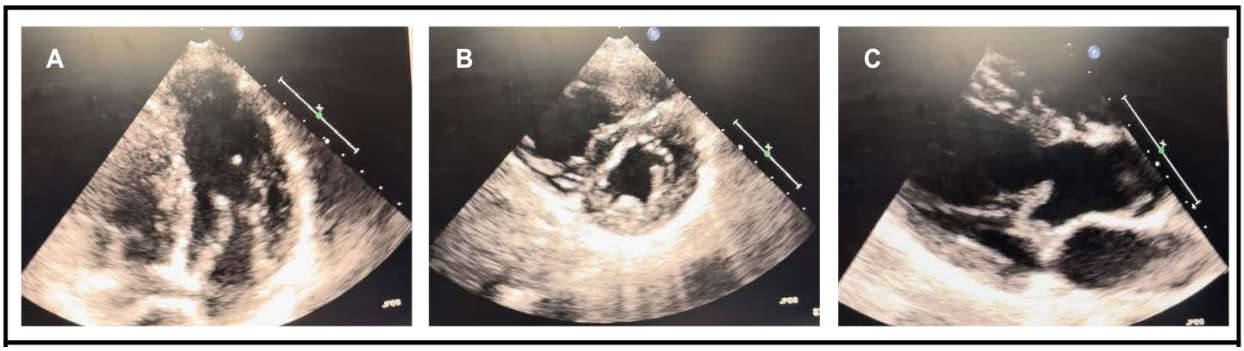

Figure 1. 2D-Echocardiogram showing parachute-like mitral valve in a four-chamber view (panel A) parasternal short axis view (panel B), and parasternal long axis view (panel C)

2. Discussion

Noonan syndrome is an autosomal dominant inherited disease that occurs in around 1 in 1000 to 2500 people. It is a RASopathy, a group of genetic syndromes that involve germ-line mutations in the RAS/mitogen-activated protein kinase (MAPK) pathway. The RAS/MAPK pathway plays a crucial role in cell division, proliferation, differentiation, and migration. Mutations causing Noonan syndrome are considered a gain of function mutation, leading to inappropriate prolongation of the RAS/MAPK signaling. The prolongation of the RAS/MAPK pathway results in the pleomorphic characteristics found in Noonan syndrome [1].

The effects of Noonan syndrome are diverse and seen in various organ systems, with complications beginning as early as during pregnancy. A clinical study of Noonan syndrome conducted by Sharland et al. diagnosed Polyhydramnios in 33% of pregnancies. Common cardiac lesions include pulmonary stenosis(62%) and hypertrophic cardiomyopathy (20%). Other commonly seen abnormalities include feeding difficulties, short stature, low weight and head circumference, motor milestone delay, and abnormal vision and hearing. Cryptorchidism is also seen in males [2].

The disease presents with facial anomalies, and webbed neck and chest deformities. The phenotypic presentation of Noonan syndrome varies significantly, with some patients showing only a few features of the disease [3].

Parachute Mitral Valve is an abnormality in which the mitral valve (MV) chordae insert into a single papillary muscle (anterolateral papillary muscle [ALPM] or posteromedial papillary muscle [PMPM]). Parachute-like asymmetric mitral valve (PLAMV) is a similar anomaly in which chordae are distributed unequally between two identifiable papillary muscles. Usually, the dominant papillary muscle is normal and the other is elongated and displaced toward the mitral valve annulus. Although both mitral valve anomalies have predominant unifocalization of chordae, “true” PMV has only one papillary muscle to which all chordae are attached. When both types of PMV occur, the chordae are short and thickened, limiting the movement of normal mitral valve leaflets, and resulting in functional and hemodynamic abnormalities. Although clinical presentations may be similar, “true” PMV and PLAMV are defined separately because the valves originate and develop morphologically in different ways.

A 2004 study conducted at the University of Toronto, Canada aimed to determine the factors linked to outcomes in patients diagnosed with pulmonary atresia with intact ventricular septum (PA-IVS), excluding those with atrioventricular septal defect. The study included 84 patients, of whom 64% were male, with a median age of 3 days (range, birth to 5.4 years) at presentation. Almost all (99%) of the patients had associated cardiac anomalies, with aortic coarctation, atrial septal defect, bicuspid aortic valve, patent ductus arteriosus, and ventricular septal defect being the most common. Left-heart obstructive lesions were present in 80% of the patients, and 26% had extracardiac anomalies. The survival of patients with PA-IVS was significantly associated with the spectrum of associated cardiac lesions, with those having left ventricular hypoplasia and atrial septal defect showing poorer survival rates. The risk of mitral valvulotomy was higher in patients with subaortic stenosis and in the absence of aortic coarctation. In conclusion, outcomes for patients with PA-IVS depend on the associated cardiac lesions' spectrum, with left ventricular hypoplasia and atrial septal defect being independent predictors of poor survival. The majority of patients will not require valvulotomy as the degree of mitral valve obstruction tends to remain stable [4].

The parachute mitral valve, supramitral membrane, and double-orifice mitral valve are commonly associated with stenotic physiology. A comprehensive study of articles published since 2000 examined 149 cases of parachute mitral valves. The study found that most patients with this condition had either stenotic or regurgitant mitral valve disease, frequently accompanied by left heart obstructions that resulted in hemodynamic compromise. Consequently, multiple surgical interventions are often necessary for these patients. Mitral valve repair is preferred over mitral valve replacement for mitral valve abnormalities to avoid the complications associated with valve replacement. The study demonstrated favorable outcomes in surgical patients, with a total event-free survival rate of 84.8% [5].

Noonan syndrome represents a diagnostic challenge for clinicians due to its broad phenotypic presentation. The parachute like Mitral Valve is an unusual presentation of this pathology, however, a low diagnostic threshold should be kept among physicians since early recognition and prompt referral can be lifesaving for patients with congenital heart disease secondary to this syndrome.

This case report presents a unique case of an isolated parachute-like asymmetric mitral valve in a patient with Noonan syndrome. It highlights the importance of regular follow-up and monitoring of patients with Noonan syndrome to identify potential cardiac abnormalities and intervene appropriately. In addition, it underscores the challenges associated with managing patients with intellectual disabilities, particularly with respect to cardiac surgery.

Overall, increasing awareness and understanding of the diverse clinical manifestations of Noonan syndrome can help healthcare professionals provide timely and effective management, improve patient outcomes, and enhance the quality of life for affected individuals and their families.

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