Acta Scientific Clinical Case Reports

Volume 3 Issue 5 May 2022

Case Report

Shone's Syndrome: A Rare Complex Congenital Heart Disease

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Received: March 13, 2022

Published: April 08, 2022

Abstract

We present an interesting and complex congenital abnormality called Shone's syndrome, which is quite rare and demands multidisciplinary approach in order for these patients to survive to adult life as well as careful follow up and decision making. This specific case apart from its educative character, could trouble our minds and make us consider some parameters of life better, before we lead our patients to indicative treatments. Our patient had all the typical characteristics of Shone 's syndrome, which had already been diagnosed and surgically treated at early age and was hospitalized at our department due to symptoms of heart failure.

Keywords: Shone; Congenital; Complex; Mitral Stenosis; Parachute-like; Subvalvular

Abbreviations

CMS: Congenital Mitral Stenosis; MS: Mitral Stenosis; SS: Shone's Syndrome; AoS: Aortic Stenosis; SVMM: Supravalvular Mitral Membrane; LV: Left Ventricle; VSD: Ventricular Septal Defect; LVH: Left Ventricular Hypertrophy; TTE: Transthoracic Echocardiogram; TEE: Transesophageal Echocardiogram; LVOT: Left Ventricular Outflow Tract

Introduction

Shone's syndrome (SS) is a rare congenital heart disease described by Shone in 1963. Its incidence is o.6% and the population studied concerns mainly children who receive early operative treatment. The main survival is 75% in a 15-year follow up. In the complete form, four left-sided defects are present [1]:

- Supravalvular mitral membrane (SVMM)
- Parachute mitral valve
- Subaortic stenosis (membranous or muscular)
- Coarctation of the aorta

Of these four defects, supravalvular mitral membrane (SVMM) is the first to occur, and triggers the development of the other three defects. Partial complexes, have also been described. The definition is often expanded to include lesions of the left side of the heart not originally ascribed to Shone's syndrome, including mitral and aortic valvular lesions and supravalvular aortic stenosis.

In this context, SS is actually a complex [2] collection of left sided obstructive heart lesions including a wide range of malformations commonly the following eight [3]: supravalvular mitral membrane, parachute mitral valve, subaortic stenosis, coarctation of aorta, cor triatriatum, bicuspid aortic valve with small aortic valve annulus, hypoplastic (stiff) left ventricle and small aortic arch. At least three of these eight findings are needed to reach at the diagnosis.

As far as congenital mitral stenosis (CMS) is concerned, in current bibliography, there are different types described as seen above. There is the typical CMS, atypical CMS with parachute like mitral valve, double-orifice mitral valve, hypoplastic mitral valve and supravalvular fibrous ring. Figure 1 depicts the incidence of each anomaly participating in the diagnosis of Shone's complex.

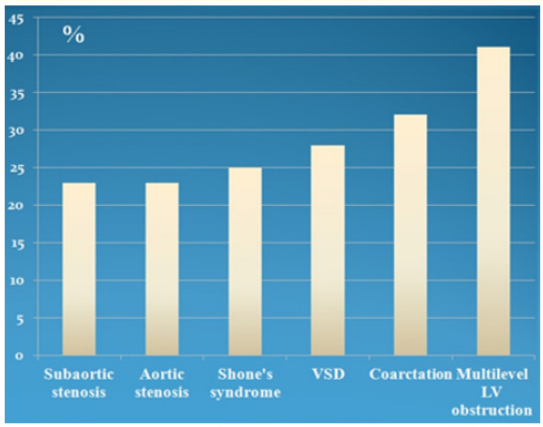


Figure 1: Incidence of each malformation participating at SS diagnosis. The main abnormality concerns a multilevel LV obstruction which enhances the diagnosis.

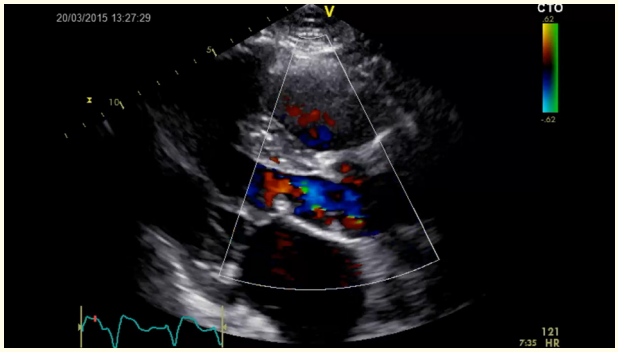
Methods and Materials

Case Presentation

A 40-year-old man was admitted at the emergency department of our hospital due to shortness of breath at the slightest effort, a symptom occurring progressively from months now. His past medical history included: persistent atrial fibrillation diagnosed 4 months ago, mitral valve valvuloplasty at the age of 3 years old accompanied by closure of atrial septal defect (ASD), redo surgery with repair of subvalvular aortic stenosis at the age of 19 years old and a known residual moderate MS and subvalvular aortic stenosis re-appearance during the past 3 years. Physical examination revealed bibasilar lung rales, dilated jugular veins bilaterally, arrhythmic S1 and S2 sounds, a holosystolic murmur heard at the second right intercostal space and a diastolic murmur at the cardiac apex. ECG showed atrial fibrillation with an LBBB-like pattern and indications of LVH.

Echocardiographic findings

A transthoracic echocardiogram (video) conducted at the emergency department depicted the presence of severe mitral stenosis as well as a significant subaortic stenosis with severe aortic regurgitation (Figures 2,3,4). Moreover, left ventricle appeared to be hypertrophic (LVH) but with preserved ejection fraction (EF) which was found 50%, whereas the right ventricle was dilated and hypokinetic. Indications of pulmonary hypertension with moderate to severe tricuspid regurgitation were also observed. Typical characteristics of Shone's syndrome were noticeable during the echocardiographic study. A transesophageal echocardiogram was also acquired for better quantification of valvulopathies and better depiction of anatomical abnormalities.



Video

As mentioned above, there are different anatomical variations of mitral valve stenosis [4] with the typical form being the most prevalent. It is characterized, not only by a valvular-level stenosis but also by a subvalvular funnel-shaped one, due to fibrotic processes occurring at the papillary muscles (Figure 6) which approach each other and chordae tendineae (Figure 7) that shorten in length or sometimes are completely absent. Multilevel mitral planimetry shows this funnel-shaped subvalvular apparatus (Figure 5). In addition, two distinct papillary muscles were seen at short axis transthoracic view, which is indicative of typical CMS versus atypical (Figure 8).

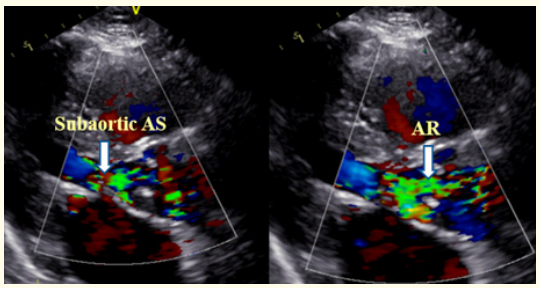


Figure 2: A long axis view at TTE revealing significant subaortic stenosis with severe aortic regurgitation.

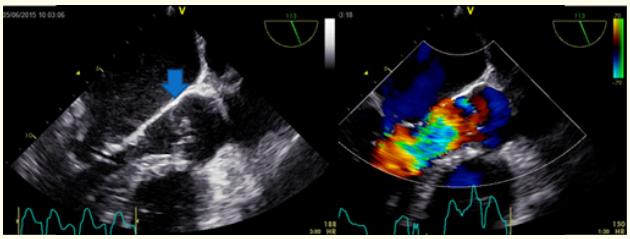


Figure 3: TEE depicting the subvalvular fibrous membrane obstructing LVOT flow.

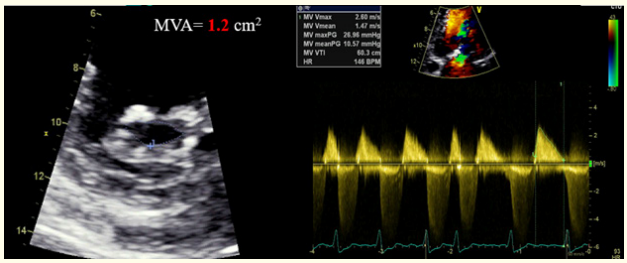


Figure 4: Multilevel mitral planimetry shows this funnel-shaped subvalvular apparatus.

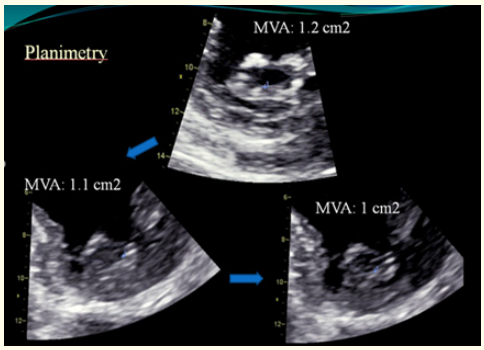


Figure 5: Significant MS needing further evaluation with multilevel planimetries as the stenosis was funnel -like and both valvular and subvalvular.

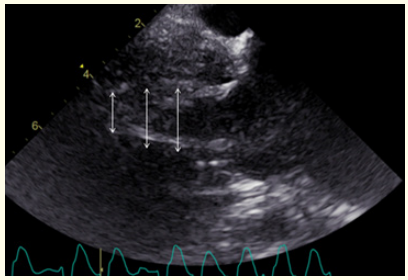


Figure 6: TEE: Papillary muscles at short distance with each other.

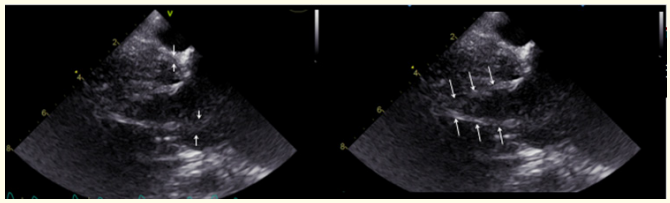


Figure 7: TEE depiction of chordae tendineae shortening on the left side and presence of fibrous lesions subvalvularly.

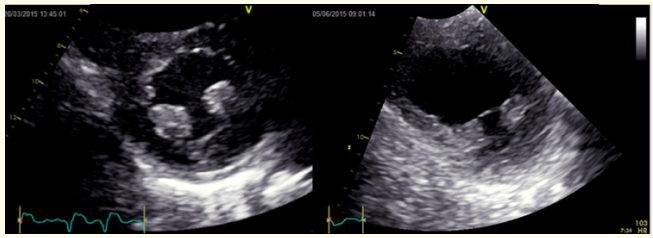


Figure 8: At the short axis view of TTE both papillary muscles are seen, isometric with no hypoplasia.

Results and Discussion

The patient was hospitalized in our clinic for about 10 days receiving at first intravenous and then per os diuretic therapy (furosemide 120mg daily) and the appropriate treatment for heart failure according to guidelines, which included ramipril 5mg once daily (o.d.), eplerenone 25mg o.d., bisoprolol 5mg o.d. plus acenocoumarol with therapeutic INR. The challenging question was the further management of this patient apart from pharmacological treatment [5].

Undoubtedly, patients with congenital anomalies, especially complex and rare ones, should frequently be evaluated for their risk of sudden death. The major factor of prevention in sudden death is early surgical treatment. An early operation leads to decongestion from volume and pressure overload conditions, avoidance of myocardial damage as well as re-scarring and arrhythmias, considering of course, the appropriate surgical technique and the expertise of the surgeon [6]. It is recommended that patients with complex congenital heart disease should be managed by an expert surgical team for this matter.

Concerning the risk stratification after surgery, it is advised that patients should be closely monitored and classified according to specific proposed characteristics to high risk for adverse events and low risk. High risk features include the following: presence of atrial fibrillation, symptoms of heart failure with pulmonary hypertension, QRS interval>130 ms, high rate of progression of LV and RV dysfunction and the level of LVOT obstruction [7].

Indications for primary prevention are better studied in patients with more common congenital anomalies and are consistent of LVEF ≤ 35% and NYHA II-III symptoms for Fallot's Tetralogy plus prolonged QRS duration equal or above 14oms or inducible VT on EPS [8].

Unfortunately, our patient suffered sudden cardiac death a month after his discharge, after having already programmed his surgical operation by an expert team [9], Even though, he was classified as high risk patient, he delayed the upcoming surgery for personal reasons and that costed him his life.

Conclusions

In conclusion, clinicians and especially those who are occupied with adult congenital disease, should be alert and recognize early the complications in disease progression in patients with congenital heart anomalies. Especially patients with rare and complex syndromes, which have not been studied thoroughly, should receive individualized care according to symptoms, echocardiographic findings and arrhythmiological risk. Moreover, other parameters including the accessibility to specialized health care should be considered and that presupposes the creation of a network of cooperation between central hospitals and rural healthcare units.

Conflict of Interest

There is no conflict of interest to declare.

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