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Congenital Mitral Stenosis: A Rare Case Report and Review of Literature

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ABSTRACT

Congenital mitral valve stenosis is a rare and severe disease, usually associated with other heart defects. The appropriate intervention depends on the site and mechanism of valvular obstruction and the aim is to avoid or delay valve replacement since it is associated with significant morbidity and mortality. We report a case of a 2-month male infant with a typical, severe mitral valve stenosis, mild bicuspid aortic valve stenosis, small patent foramen ovale, severe pulmonary hypertension with florid pulmonary edema.

Keywords: Congenital Mitral Stenosis, Bicuspid Aortic Valve Stenosis, Severe Pulmonary Hypertension, Patent Foramen Ovale, Transthoracic Echocardiography.

INTRODUCTION

Congenital mitral stenosis (CMS) is a rare and morphologically heterogeneous lesion that affects both the leaflets and subvalvar apparatus of the mitral valve (MV) [1]. Frequently, congenital CMS occurs in conjunction with additional left heart obstructions and/or a ventricular septal defect (VSD). [1, 2]. In severe cases, congenital MS is difficult to manage medically and entails significant morbidity and mortality [3]. Interventional therapies for medically refractory congenital MS include percutaneous transcatheter balloon mitral valvuloplasty (BMVP), surgical mitral valvuloplasty (SMVP), and mitral valve replacement (MVR). The optimal treatment in any given patient may depend on the severity of anatomical substrates of CMS, associated cardiovascular anomalies, and patient size. Although MVR can relieve left ventricular (LV) inflow obstruction, with little risk of procedure-related mitral regurgitation (MR), it carries high morbidity and mortality, particularly in infants and young children [2, 4-8].

CMS classically has been divided by Van Praagh into 4 anatomic types: typical, hypoplastic, supramitral ring, and parachute mitral valve [1].

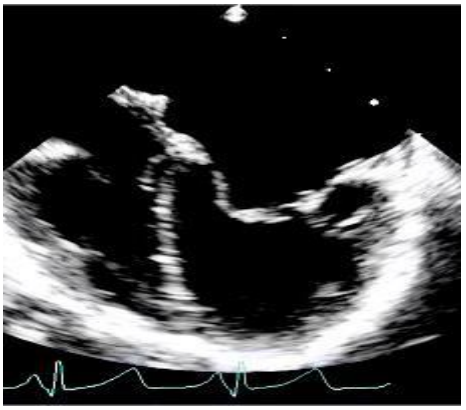
Typical Congenital MS. This form of CMS seems to correspond to the subgroup of dysplastic valve described by Anderson as the papillary muscle to commissure fusion [2]. This is the most common form of CMS and shows large phenotypic variability. Morphologically, it is characterized by thickened leaflets, short and thickened chords with obliteration of interchordal spaces, and underdeveloped papillary muscles with reduced interpapillary distance (Figure 1A). In the most severe cases, the papillary muscles come together and insert directly into the leading edge of the anterior mitral leaflet, creating a muscular arcade when the valve is viewed by the echocardiographer from the left ventricular aspect (arcade mitral valve) (Figure 1B, 1C) [3]. In other cases, the shortened and thickened chords insert directly into the

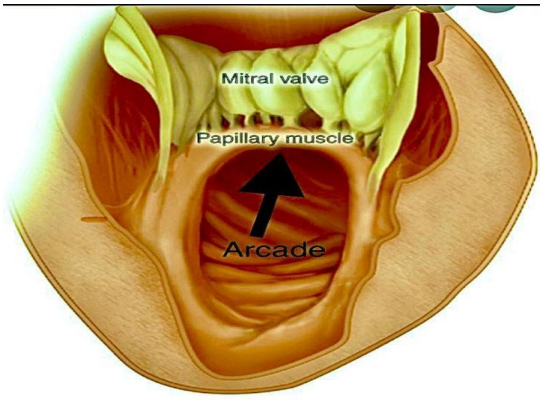
muscular mass of the posterior wall, causing the appearance of a hammock when the valve is viewed from the left atrium perspective (hammock mitral valve) [4]. This particular type of CMS is typically diagnosed early in life and is commonly associated with coarctation of the aorta and aortic stenosis.

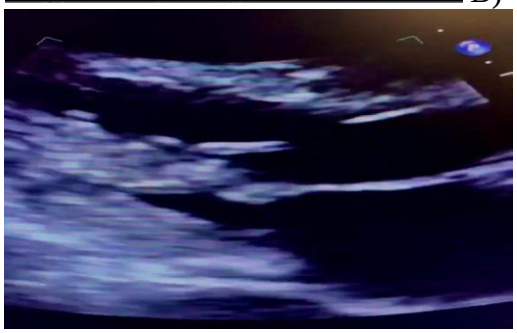
Hypoplastic MS. This is the second most common type of CMS. All the components of the hypoplastic valve are a miniature of the normal valve (Figure 1D). It virtually always is associated with severe left ventricular outflow tract abnormalities, most commonly hypoplastic left heart syndrome. It is diagnosed in infancy and never has been reported as a de novo diagnosis in adults.

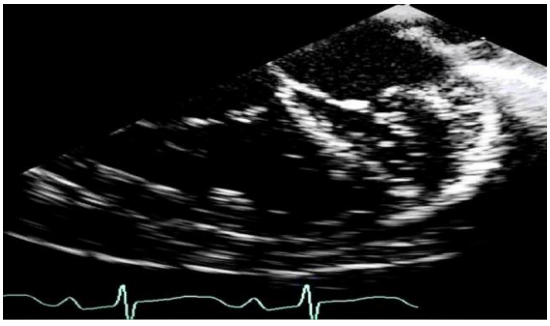
Supramitral Ring. It is a circumferential ring of connective tissue that originates from the atrial surface of the mitral valve leaflets distal to the opening of the left atrial appendage and causes various degrees of mitral inlet obstruction (Figure 1E). It frequently is associated with multilevel obstruction of the left side of the heart, as in Shone's complex (supramitral ring, parachute mitral valve, subaortic stenosis, and coarctation of the aorta) [5]. It is important to distinguish this lesion from a membrane corresponding to cor triatriatum sinister, which typically is located proximal to the left atrial appendage.

Parachute Mitral Valve. Although this is the least common form of CMS, it is discovered most frequently as a de novo diagnosis in the adult population [6-8] (Figure 1F). The chordae tendineae are thickened and shortened and insert into a single papillary muscle (typically the posteromedial), while the anterolateral is absent or hypoplastic. In parachute-like mitral valve, the hypoplastic papillary muscle is connected to an underdeveloped commissure, creating an asymmetric opening of the mitral valve.

A) 

B) 

C) 

D) 

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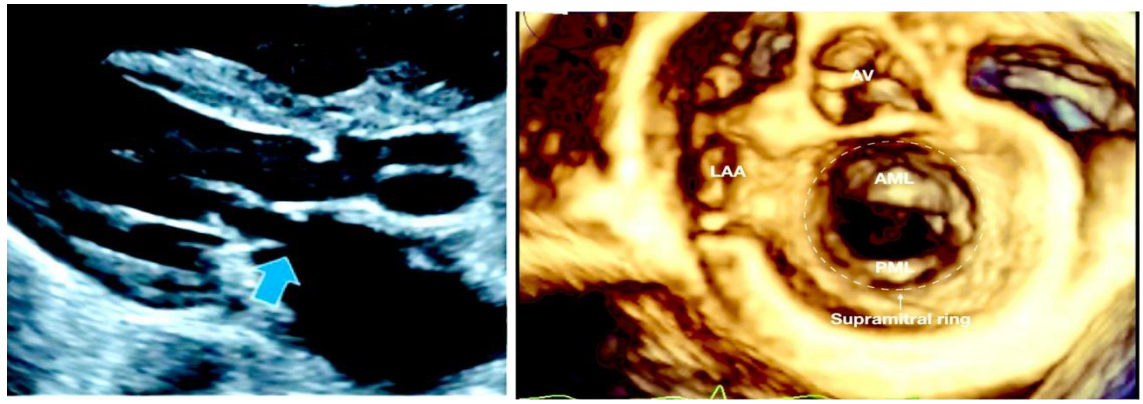

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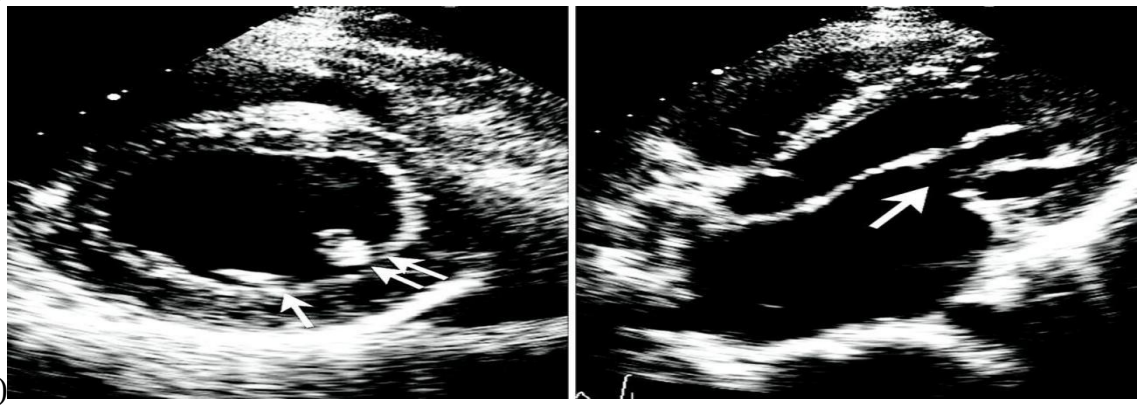


Figure 1: *A) Typical congenital mitral stenosis Transesophegeal - midesophegeal 4-chamber view showing thickened mitral valve leaflets, thickened and shortened chords, and restrictive opening of the mitral valve with diastolic doming (similar appearance to rheumatic mitral valve stenosis). B) Schematic diagram of Mitral Valve Arcade. Direct attachment of papillary muscles into mitral valve leaflets, resulting in a fibromuscular "arcade" formed by papillary muscles, short chords, and edge of anterior mitral leaflet. C), Parasternal long axis view demonstrates a characteristic "arcade" of mitral valve, causing significant stenosis. D) Hypoplastic mitral stenosis. Midesophegeal 4-chamber view showing a mildly hypoplastic left ventricle with hypoplasia of mitral valve complex (miniature mitral valve). E) Supravalvular mitral ring. Left panel - parasternal long axis view demonstrates the supravalvular mitral ring which appears as two "fangs" above the mitral valve (arrow). Right panel- 3-dimensional acquisition of the mitral valve from the left atrium perspective showing a complete supravalvular mitral ring (interrupted line). Note the location of the membrane distal to opening of LAA. AV, aortic valve, LAA, left atrial appendage, AML, anterior mitral leaflet, PML, posterior mitral leaflet. F) Parachute mitral valve. Left panel- In the short axis view a huge anterolateral papillary muscle was recognized (double arrows),while the posteromedial papillary muscle was smaller and closer to the anterolateral papillary muscle. Right panel- the shortened chordae tendineae was attached parachute like to the anterolateral papillary muscle (single arrow).*

CASE REPORT

A 2-month-old male infant presented to us in florid pulmonary edema with a history of cough, respiratory distress, chest retractions, feeding difficulties and paroxysmal nocturnal dyspnea since birth. The child was urgently managed with classical anti congestive medical management comprising of oral digitalis, furosemide, carvedilol along with oxygen inhalation. The patient responded dramatically within 3-4 hours. After the initial stabilization we were able to conduct our clinical examination even though the child was unusually irritable and incessantly crying (Figure 2).

Figure 2: Photograph of the child after decongestive management of pulmonary edema

On physical examination our patient was weighing 4kg, BP was 70/50 mmHg in the right upper limb in supine position, pulse rate was 110/min, respiratory rate was 55/min with intercostals retractions and SPO2 was 86% at room air. The pulses were normally palpable in all the four limbs. There was no apparent cyanosis, facial or pedal edema, facial dysmorphism, polydactyly or syndactyly.

On examination of the cardiovascular examination there was presence of precordial bulge, parasternal and epigastric pulsations, parasternal heave with palpable 2nd heart sound. On auscultation there was loud S1and a mid-diastolic murmur at the apex.

Respiratory system examination revealed bilateral crepitations in the interscapular and basal regions. Rest of the systemic examination was unremarkable.

Resting ECG (Figure 3) shows sinus tachycardia with a ventricular rate of 150/min, left atrial enlargement and right ventricular hypertrophy.

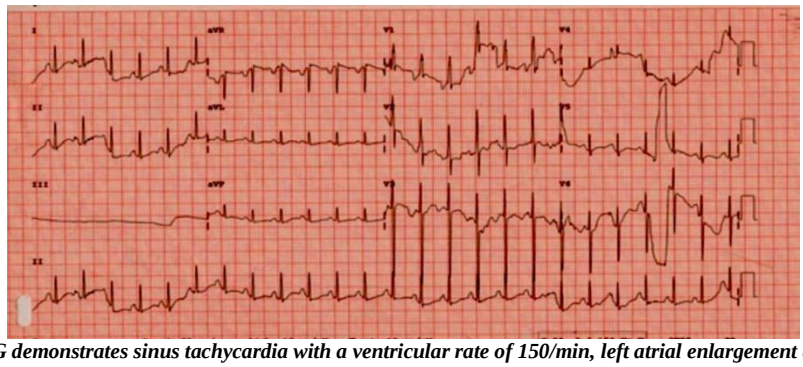


Figure 3: Resting ECG demonstrates sinus tachycardia with a ventricular rate of 150/min, left atrial enlargement and right ventricular hypertrophy.

Xray chest (AP) view (Figure 4) indicates obvious cardiomegaly with severe pulmonary venous congestion.

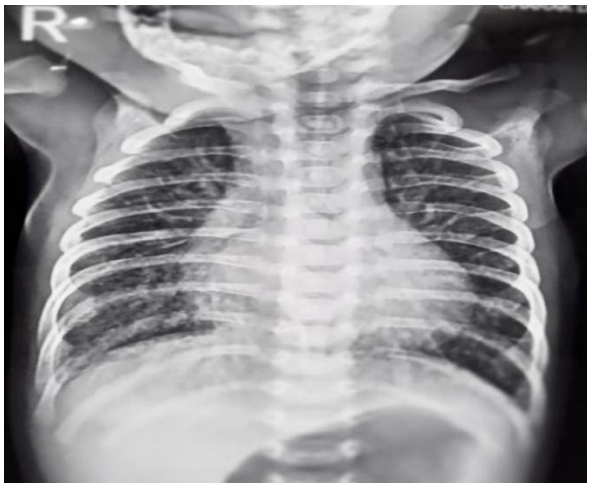


Figure 4: X-ray chest AP view indicates obvious cardiomegaly with severe pulmonary venous congestion.

Transthoracic Echocardiography

Standard transthoracic echocardiography (TTE) was performed by the author with the patient lying in the supine and left lateral decubitus position. The study was conducted in the parasternal long axis (LX), short axis (SX), four chamber (4CH), five chamber (5CH), and suprasternal views.

A detailed TTE was carried out and the findings are enumerated below:

1. Levocardia

Situs Solitus
AV Concordance
VA Concordance
Concordant D-bulboventricular loop
Normally related great arteries
Confluent pulmonary arteries
Left aortic arch.
Normal systemic & pulmonary venous drainage

2. Patent foramen ovale (small) (Figures 5A-5D)

Size 1.9 mm
Peak/mean gradient across patent foramen Ovale (PFO) was 11.4 / 6.6 mmHg
There was presence of left to right shunt

3. Congenital Mitral Stenosis (Severe)

Typical variety of congenital mitral stenosis (CMS) was recognised (Figures 6A-6D). It consisted of thickened leaflets, short and

thick cords with obliteration of interchordal spaces. Interestingly, the papillary muscles could not be delineated and the chords were directly inserting into the muscle mass of the posterior wall (Figure7). The mitral valve area by pressure half time (PHT) method was 0.41 sqcm and the peak mean gradient was 34.9/10.1 mmHg (Figure 8).

4. Bicuspid aortic valve stenosis (Mild)

In the SX view at the level of aortic valve, a bicuspid aortic valve was detected (Figure 9A). On continuous wave doppler analysis across the aortic valve showed a peak and mean gradient of 16.3/8.8 mmHg, consistent with mild aortic stenosis (Figure 9B).

5. Severe Pulmonary hypertension with consequent tricuspid regurgitation (Moderate)

In the apical 4CH view a highly turbulent tricuspid regurgitation (TR) jet was visualised in the apical 4CH view (Figure 10A). On continuous wave doppler analysis a TR velocity of 5.63 m/sec was present with a peak gradient of 126.7 mmHg (Figure 10B). The estimated right ventricular systolic pressure/pulmonary artery systolic pressure was 141.7 mmHg. Consequently, there was notable dilatation of main and branch pulmonary arteries (Figure 10C) along with moderate dilatation of right ventricle (Figure 10 D).

6. Small left ventricular cavity size with moderately dilated right ventricle was ascertained along with normal biventricular systolic function. The LVEF was normal - 80% (Figure 11 A, 11B).
7. Moreover, there was no evidence of VSD, PDA, COA, pulmonary valvular or infundibular stenosis.
8. The infant was thereafter referred to a tertiary care pediatric cardiovascular institute for suitable balloon mitral valvulotomy or MVR due to the presence of advanced pulmonary edema, severe pulmonary hypertension alongside severe CMS.

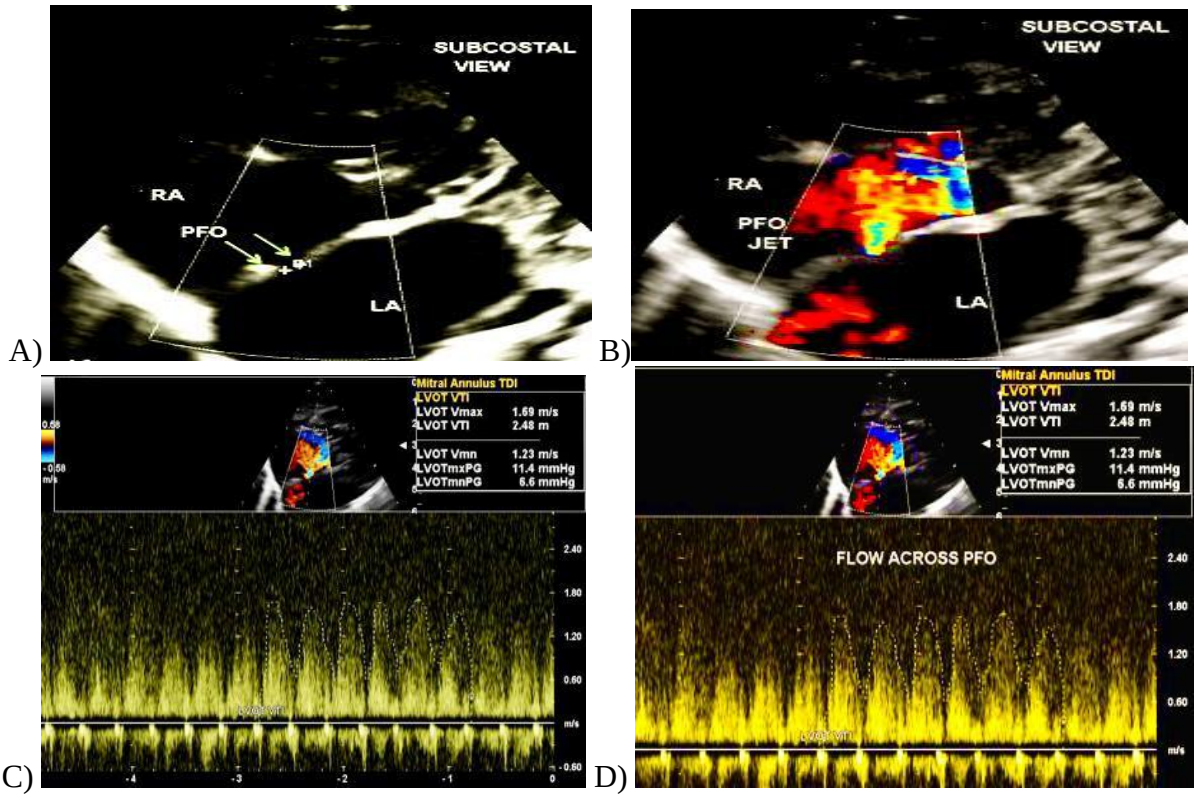
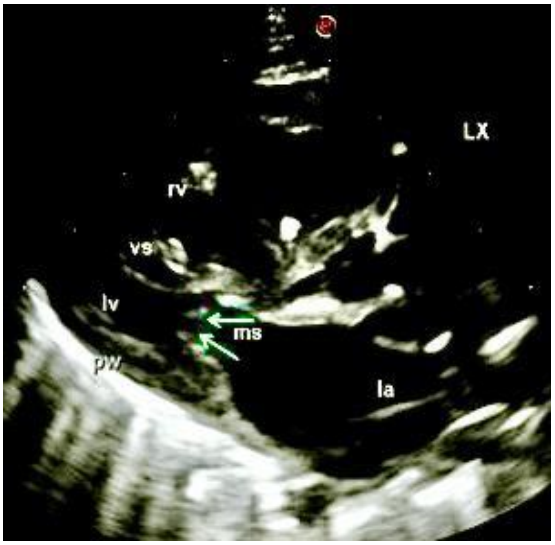


Figure 5*: Patent foramen ovale . A), In the subcostal view a small PFO is visualized. B), On color flow mapping a PFO jet is distinctly demonstrated. C), Non- highlighted flow across PFO displays a peak/mean gradient of 11.4/6.6 mmhg. D), Highlighted flow across PFO*

A)



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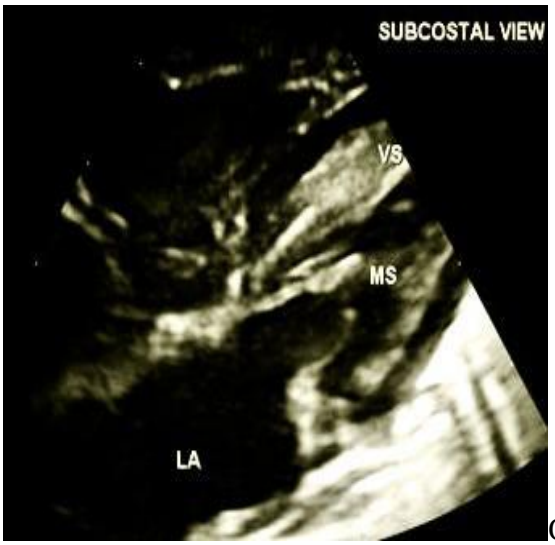

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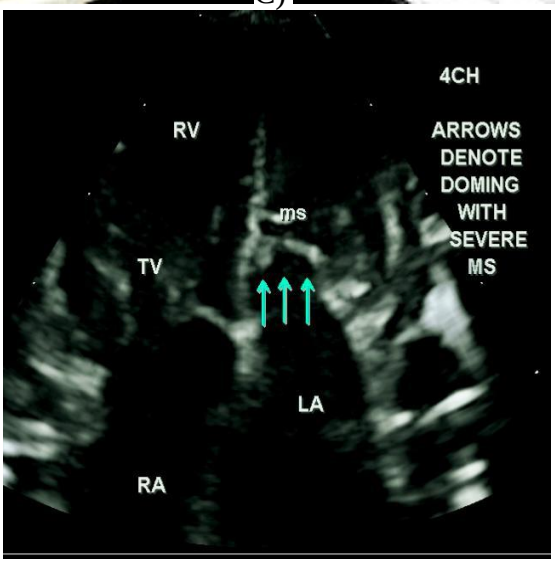
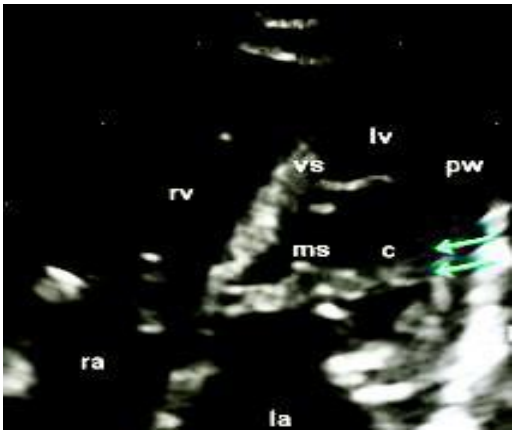


Figure 6: *Congenital mitral stenosis. A) Parasternal long axis view is showing domed and severely stenotic mitral valve. B) In the subcostal view a typical congenital mitral stenosis is identified. C) Marked restriction of mobility of anterior and posterior mitral leaflet was observed (arrows denote the non-mobility of mitral valve).D) In the apical 4CH view a severely stenotic and domed mitral valve is detected.*

A) 

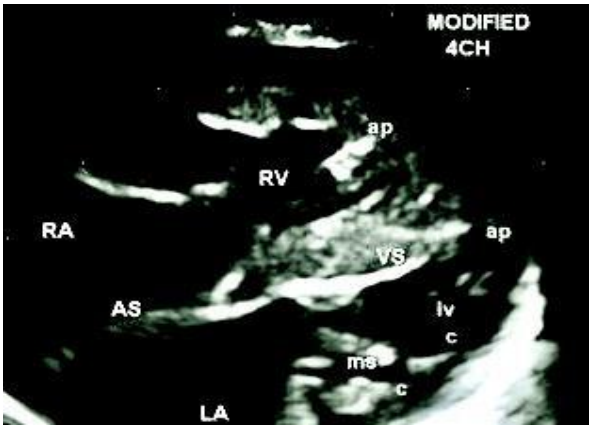
B) 

Figure 7: *A) In the apical 4CH view no papillary muscles were discerned and the chordae were directly inserting into the posterior wall. B) In the modified 4CH view similar findings were present. la, left atrium; ms, mitral stenosis; c, chordae ; pw, posterior wall; lv, left ventricle; vs, ventricular septum*

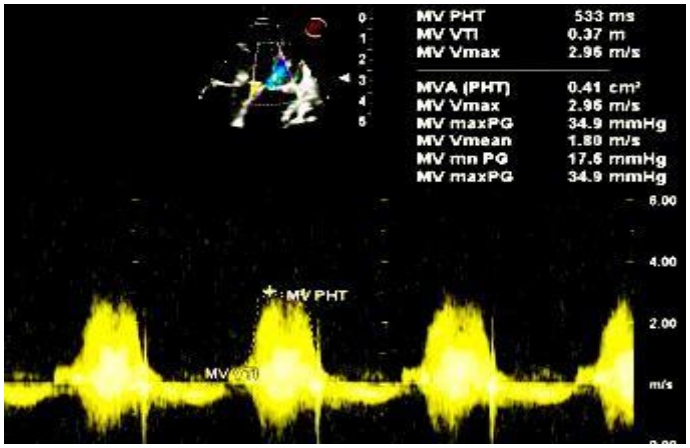
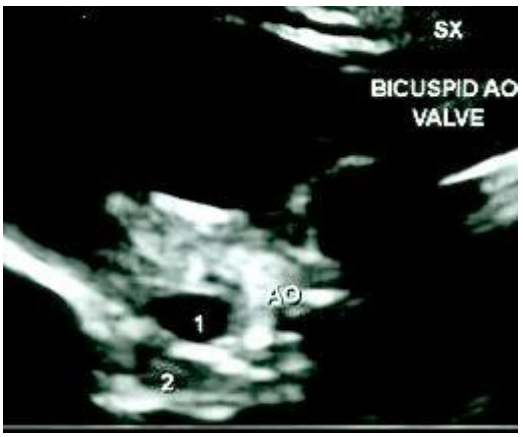


Figure 8*: Continuous wave doppler analysis across the mitral valve revealed a mitral valve area of 0.41 sqcm by pressure half time. The peak and mean gradient was 34.9/17.6 mmhg.*

A) 

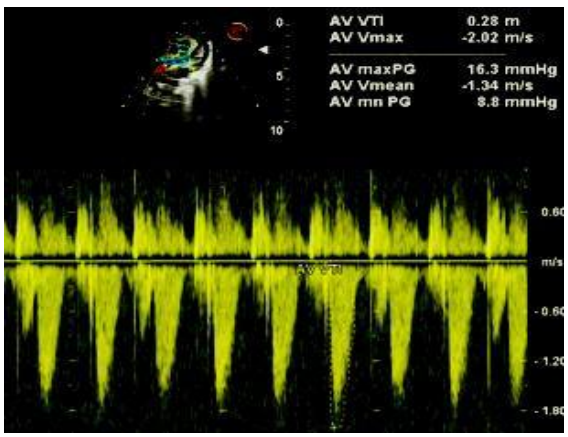
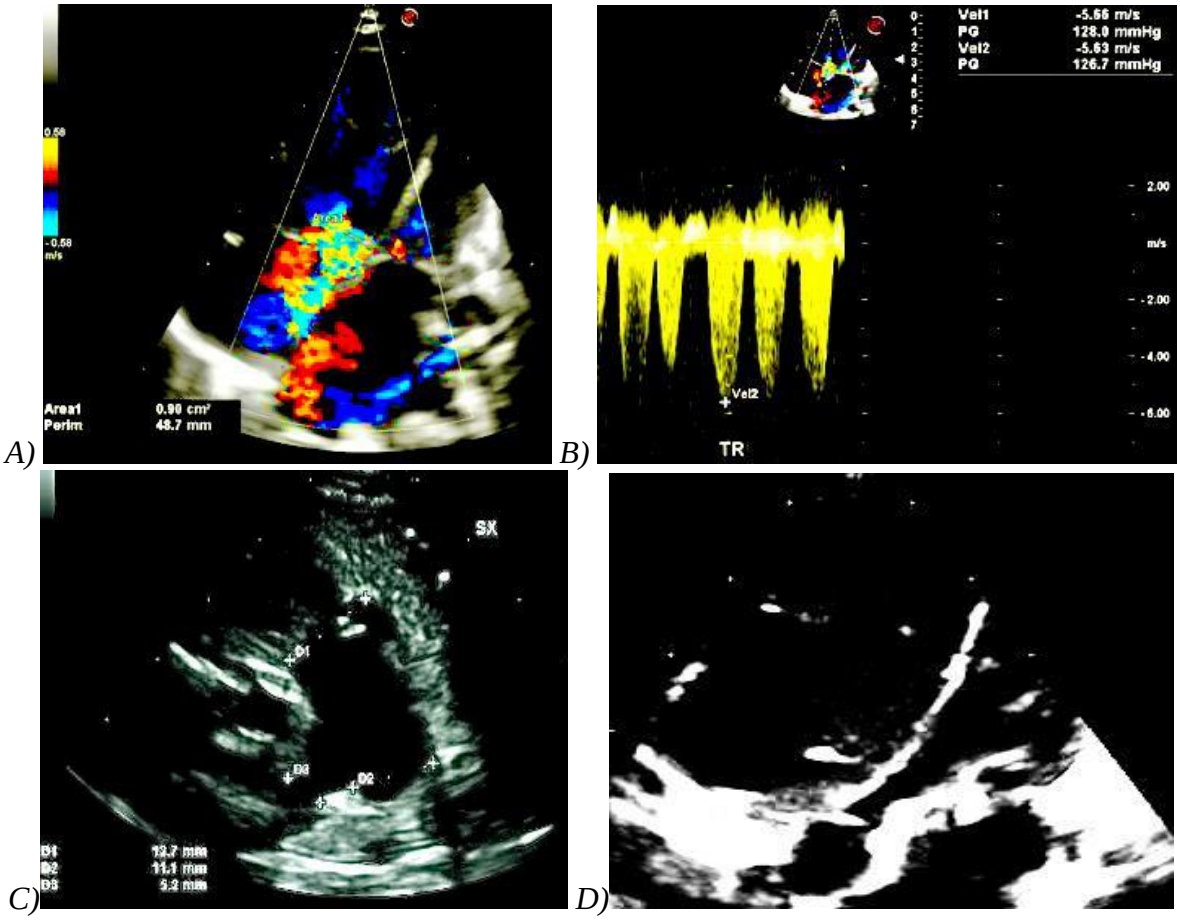
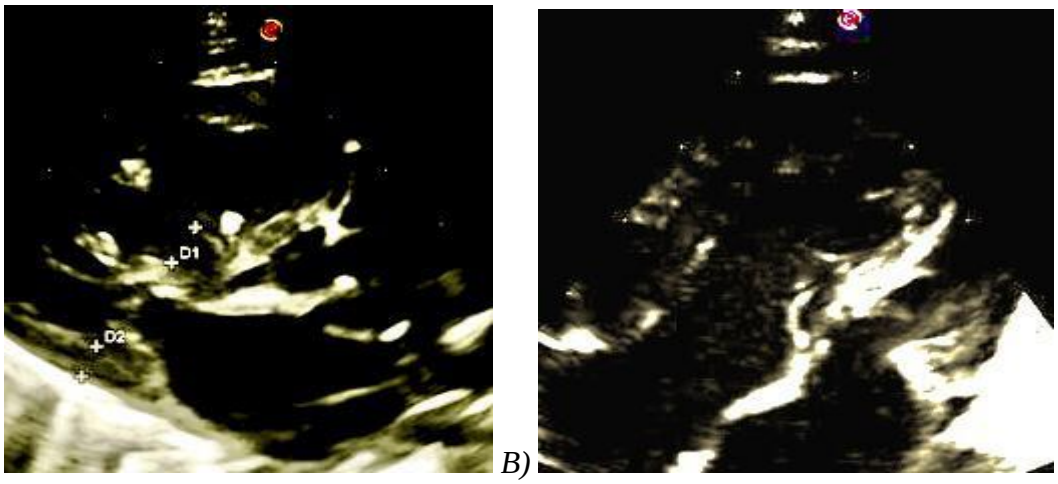
B) 

Figure 9: *Bicuspid aortic valve stenosis (mild). A) Short axis view at the level of aortic valve shows a bicuspid aortic valve. B) On continuous wave doppler analysis across the aortic valve discovered a peak and mean gradient of 16.3/8.8 mmhg , consistent with mild aortic stenosis.*



A) *B)* *C)* *D)*

Figure 10: *A) A highly turbulent tricuspid regurgitation jet was visualised in the apical 4CH view. B) On continuous wave doppler analysis a tricuspid regurgitation velocity of 5.63 m/sec was present with a peak gradient of 126.7 mmhg. C) There was notable dilation of main and branch pulmonary arteries. D) Dilated right ventricle with normal systolic function was identified.*



A) *B)*

Figure 11: *A) In parasternal LX view a small left ventricular cavity size with a moderately dilated right ventricle was ascertained. B) Similar findings were present in apical 4CH view alongwith normal biventricular systolic function.*

DISCUSSION

The existence of mitral valve apparatus (annulus, leaflets, chords and papillary muscle) along with the absence normal of supravalvular pathology (supramitral ring) and left ventricular disorder (hypoplastic ventricle) is essential for normal function.

CMS is a rare entity; reported incidence being 0.21 to 0.4% [9, 10]. Moore P et al [10] described 85 infants of CMS which

included stenotic mitral valve with symmetry of papillary muscle (52%), supravalvar mitral ring (20%). double orifice mitral valve (11%) and hypoplastic mitral valve with asymmetric papillary muscle (8%) while Mc Elhinney DB et al [11] reported typical congenital MS in 78 infants, supravalvar mitral ring in 46, parachute mitral valve in 28 and double-orifice mitral valve in 11, with multiple types in around 50% of patients of series of 108 infants with severe congenital mitral stenosis.

Isolated CMS is very rare and is found in association with left heart underdevelopment, left ventricular outflow tract obstruction and Shone complex [12].

Majority of the series demonstrated coarctation of aorta, subaortic stenosis, bicuspid aortic valve, supravalvular aortic stenosis, aortic regurgitation, ventricular septal defect, atrial septal defect, patent ductus arterious, small left ventricle, double outlet right ventricle, pulmonary stenosis and tetralogy of fallot [10, 13-15].

Clinical presentations of CMS are dependent on the anatomic variant, severity, associated lesions and age. Pulmonary hypertension and inadequacy of the left heart to support the systemic circulation are the most significant clinical consequences.

Patients with severe CMS may present with respiratory distress from pulmonary edema shortly after birth if a significant atrial septal communication does not exist.

Patients with mild to moderate CMS may present after the neonatal period with signs of low cardiac output and RV failure such as pulmonary infection, failure to gain weight, tachypnea and chronic cough [16]. Our patient was a 2-month-old infant and presented to us in florid pulmonary edema. There was history of recurrent bouts of respiratory distress, intercostal retractions, profuse sweating while feeding and paroxysmal nocturnal dyspnea, even though there was presence of small patent foramen ovale. However, after stabilizing the patient with oral medications and oxygenation, we demonstrated a severe congenital mitral stenosis and severe pulmonary hypertension which was co-existing with mild bicuspid aortic valve stenosis.

Echocardiography is the most important diagnostic tool to evaluate patients with CMS.

Functional severity of stenosis is based on the widely accepted echocardiographic definition by the American Society of Echocardiography that stratifies the mean gradient across the mitral valve to mild (mean gradient less than 5mm Hg), moderate (mean gradient between 5 and 10mm Hg), and severe (mean gradient more than 10mm Hg) [17].

Interventional therapies for medically refractory CMS depend upon specific mitral valve pathology; percutaneous valvuloplasty, surgical valvuloplasty and mitral valve replacement [18]. Surgical interventions of CMS are usually postponed until the symptoms appear as a higher surgical risk and an efficient and durable repair are somewhat difficult to achieve due to limited exposure of the valvar and subvalvar apparatus in small structured hearts. Mitral valve replacement is best avoided in infants and small children because of frequent size mismatch between the smallest mechanical valves and the hypoplastic mitral valve annulus. In addition, somatic growth in children leads to the need for subsequent mitral prosthesis replacement.

An effective option to surgical management is balloon dilatation, which is appropriate even for infants, with unsuccessful medical management but chances of restenosis reported from various studies [10]. Operative results and long-term outcome are extremely variable and highly dependent on coexisting anomalies [10, 16, 17, 19].

CONCLUSION

Clinically CMS is difficult to diagnose as coexisting intracardiac lesions may mask or unmask the disease. Echocardiography and angiocardiography are important diagnostic tools. Despite great advances in surgical

technique, surgical treatment of CMS is still challengeable particularly in neonates and small infants.

Declaration by Authors
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Conflict of Interest: The authors declare no conflict of interest.

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