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# Shone's complex: An extremely rare congenital cardiac anomaly in a preterm newborn

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## Abstract

Shone's Anomaly (SA) is a rare congenital cardiac malformation that consists of four obstructive lesions of the left heart and the aortic arch. We report on a preterm infant with Respiratory Distress Syndrome (RDS) diagnosed with SA after cardiac auscultation revealed a loud second heart sound. A 2D echocardiogram study revealed a large ventricular septal defect and a variant of SA consisting of sub-valvular aortic stenosis, hypoplastic aortic arch, coarctation of the aorta, and dysplastic mitral valve along. The infant underwent a successful Hybrid procedure at two months of age consisting of pulmonary artery banding, stenting of the patent ductus arteriosus, and placement of an atrial stent. Although SA can present in infancy, we have not found a separate report of a preterm infant with SA in the literature. This case presentation raises awareness about this anomaly, sheds light on the importance of careful cardiac examination in newborns, and emphasizes the importance of an echocardiographic exam in evaluating preterm infants with an unusual course of RDS.

***Keywords:*** Premature; Preterm; Congenital heart disease; Infant; Shone's anomaly; Dysplastic mitral valve; Aortic stenosis; Subvalvular aortic stenosis; Hypoplastic aortic arch; Coarctation; Mitral valve dysplasia; Hybrid procedure.

## Introduction

Shone's Anomaly (SA) is a rare Congenital Heart Disease (CHD) initially described by John Shone et al. in 1963 [1]. The incidence of this cardiac complex is 0.6% of all CHD cases [2]. Complete SA comprises four left-sided heart defects, including supra-valvular mitral ring, "parachute" mitral valve, membranous or muscular subaortic stenosis, and coarctation of the aorta (CoA) [1]. Partial forms of SA have also been described where less than four of the above pathological anomalies are present [1-4]. The prognosis of this anomaly depends on the complexity and the severity of the different obstructive lesions [1,5,6].

Although few reports of this anomaly in newborns, SA is traditionally diagnosed in infants and children and very rarely in young adults where the clinical presentation and post-surgical prognosis may have distinctive features [2,3,5-10].

In this report, we present the case of a preterm infant with respiratory distress syndrome, who was diagnosed with SA at two weeks of age when found to have abnormal second heart sound on cardiac auscultation.

## Case report

A male infant, the third of a trichorionic triamniotic pregnancy, born at 32 weeks gestational age to a 34-year-old cau-

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casian mother who had class A1 gestational diabetes and preeclampsia, conceived via in-vitro pregnancy using frozen sperm technique. Antenatal laboratory screening and fetal ultrasound scans were unremarkable. Then the baby was born via an emergency cesarean section for maternal preeclampsia with elevated liver enzymes. The baby was born vigorous, Apgar scores were 9 and 9 at 1 and 5 minutes, respectively. Birth weight was 1.29 kg (8th percentile), head circumference 25 cm (<5th percentile), and length 35 cm (>fifth percentile). The baby immediately developed signs of respiratory distress requiring Continuous Positive Airways Pressure (CPAP) en route to the neonatal intensive care unit. The placental histopathological examination was grossly unremarkable.

Physical examination upon admission to the NICU revealed tachypnea and subcostal retraction; air entry was equal bilaterally. The rest of the clinical examination was unremarkable. Chest X-ray revealed a moderate ground-glass appearance consistent with Respiratory Distress Syndrome (RDS).

At 12 hours of age, the baby was weaned to 2 liters of nasal cannula. However, he was shifted back to CPAP on the fourth day of life due to tachypnea and intercostal retractions. On day 6 of life, he developed tachycardia, hypoactivity, mottled skin, and acidosis. Capillary blood gas analysis showed pH 7.19, $pCO_2$ 78, and BE - 1.5. Therefore, the baby was intubated and started on mechanical ventilation. A chest x-ray demonstrated moderate perihilar and diffuse interstitial congestion without focal opacification. The baby underwent complete evaluation for sepsis and started on antibiotics which were discontinued after five days. On the following day, he was extubated to Nasal Intermittent Ventilation (NIV).

The baby continued on NIV, gradually weaned to nasal cannula, $FiO_2$ 0.21 to 0.25, and thriving well on orogastric feeding. On day 18 of life, cardiac auscultation revealed a loud second heart sound. Two-dimensional echocardiography (2D echo) study (Figure 1) demonstrated dilated Right Atrium (RA) and Right Ventricle (RV), dysplastic Mitral Valve (MV) with restrictive opening of the mitral orifice, large low apical muscular Ventricular Septal Defect (VSD) with bidirectional shunting, dilated main pulmonary artery and branch PAs, normal pulmonary veins, marked flattening of the interventricular septum, smallish left heart with left ventricular diastolic dysfunction, a subaortic membrane with no significant Left Ventricular (LV) outflow tract obstruction, aortic annulus size of 6 mm, hypoplastic aortic arch, common branching of brachiocephalic, transverse aortic arch 3 mm, aortic isthmus 2 mm (Figure 2), and moderate Patent Ductus Arteriosus (PDA) with bidirectional shunting.

The echo findings were consistent with Shone's complex with Pulmonary Hypertension (PH), indicating cardiac catheterization and surgical repair. The baby was started on furosemide and a low dose of Alprostadil infusion to maintain ductus patency. However, the procedure was deferred until the child gains more weight.

Additional investigation revealed left ectopic kidney and normal chromosomal microarray and newborn metabolic screening results. The baby had developed complications of Alprostadil infusion, including episodes of apnea and fever, which required repeated screening for sepsis. He also developed an attack of Supraventricular Tachycardia (SVT) at four weeks of age, which was aborted after a single dose of adenosine. Subsequent attacks of SVT were brief, infrequent, and self-resolving. Repeat echo studies did not show significant cardiac anatomic changes.

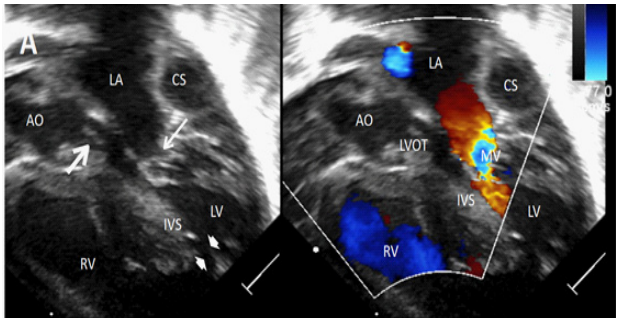


**Figure 1:** (A) Apical 5 chamber view in two-dimensional echocardiography (left) with color-flow Doppler (right) depicting stenotic MV (thin arrow), LVOT obstruction (thick arrow), large apical VSD (arrow heads) and dilated CS.

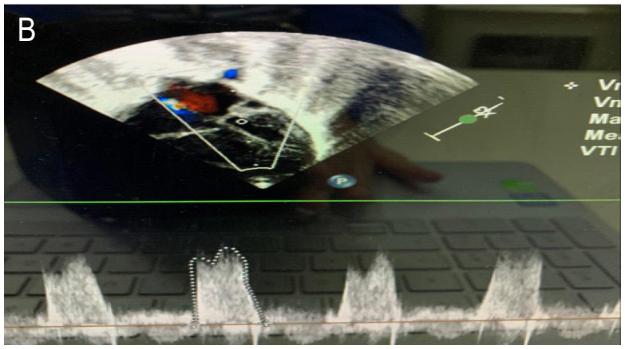


**Figure 2:** (B) Apical 5 chamber view in two-dimensional echocardiography depicts an abnormal peak velocity blood flow through mitral valve and abnormal E/A ratio , a marker of left ventricular diastolic dysfunction.

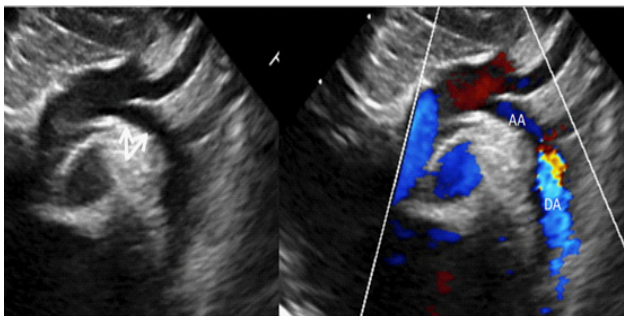


**Figure 3:** Suprasternal view in two-dimensional echocardiography (left) with color-flow Doppler (right) depicting hypoplastic aortic arch (thick arrow). MV: Mitral Valve; LVOT: Left Ventricular Outflow Tract; VSD: Ventricular Septal Defect; CS: Coronary Sinus; AO: Ascending Aorta; IVS: Interventricular Septum; RV: Right Ventricle, LA; left atrium; LV: Left Ventricle; AA: Aortic Arch; DO: Ascending Aorta.

At two months of age, cardiac catheterization and angiography demonstrated a PDA size of 4 mm, larger than the descending aorta, hypoplastic transverse aortic arch, and a borderline small left ventricle. The child underwent a hybrid procedure that comprises bilateral PA banding with stenting of the PDA and atrial septal defect stenting. The chest wall was closed after five days of surgery, and the baby was extubated to CPAP then

to a microflow nasal cannula within a few days.

A follow-up echo at four months of age (Figure 2) demonstrated an atrial septal stent in place with laminar flow across, a PDA stent with a bidirectional flow, a left PA band with a peak gradient of 50 mmHg, a right PA band peak gradient of 40 mmHg, hypoplastic MV and aortic valve, peak gradient of 37 mmHg across LVOT, and normal biventricular systolic function.

## Discussion

SA is a very rare anomaly diagnosed most frequently in its incomplete form, which is consistent with the cardiac findings in our patient [7]. The primary lesion in SA is congenital anatomic MV stenosis of different forms, in addition to at least one of the following: Subvalvular aortic stenosis, valvular aortic stenosis, supra-valvular aortic stenosis, aortic arch hypoplasia, or CoA [1,7].

Our patient had dysplastic MV, sub-aortic stenosis, and CoA; in addition to a large VSD, he also has a left ectopic kidney with no other associated congenital anomaly and normal genetic evaluation. It is not surprising that the MV lesion in our case did not include parachute deformity. A study of 45 patients with SA showed MV parachute in only less than 20% of the MV lesions [5].

Although PH was found in both echo studies, treatment with selective pulmonary vasodilators was not considered since its role in these patients is still unclear. In addition, data from smaller published series indicate that PH improves following the resolution of MV disease [11].

Our patient underwent the first step of surgical correction through the Hybrid procedure, which included bilateral pulmonary artery banding, stenting of the PDA, and placement of an atrial stent, therefore, deferring correction of other anomalies to the next step. In line with this approach, St. one study reported a large surgical series of 28 children with SA and showed that deferring MV repair does not worsen the patient's prognosis [12]. Similarly, Ilkemba CM et al. [9] indicated that in the management of SA, MV repair in patients with parachute MV and subaortic stenosis is increasingly required in the absence of CoA. However, others favor an aggressive approach to MV pathology once the transmitral gradient reaches more than five mmHg [5].

In our case, the first two echo studies demonstrated a small LV while hoping this patient gains more LV growth for a better outcome; it is undetermined if he will be subjected to two-ventricle physiology or single ventricle palliation. Some studies indicated that smaller MV and smaller LV are associated with the failure of two-ventricle physiology circulation [7,13]. In addition, an LV and RV ventricle diastolic longitudinal ratio >0.75 predicted a successful biventricular repair in one study involving 19 patients, even in cases with a small MV [14].

The operative mortality in patients with SA is adversely affected by the severity of MV disease, the degree of LV hypoplasia and outflow obstruction, and the need for multiple operative procedures [6,9,15]. It's also worth noting that postoperatively, LV dysfunction might persist, with high LV end-diastolic pressure and PH following repair of obstructive lesions in SA [6,16].

Survival in patients with SA is now favorable. In one study involving 45 children ranging from 2 months to 16 years, the 10-year transplant-free survival was 76% [5], a similar study involving 121 patients of the same age group showed a survival rate of 86% [7]. In the study done by St. Louis JD et al., he reported an overall survival rate of 93% with an average follow-up of 6.3 years (range, 1 to 16 years). All 24 surviving patients were in class I or II congestive heart failure [12].

Morbidities and potentially fatal complications in these patients include PH, cardiac failure, renal failure, permanent heart block, pneumonia, growth failure, stroke, and gastrointestinal bleeding [5,7,8,16]. In addition, the presence of CoA in our patient increases the likelihood of further intervention. Furthermore, premature birth is considered an independent risk factor for mortality in infants with complex CHD [17].

## Conclusions

This case report raises awareness about this rare syndrome. It highlights the importance of repeated cardiac examination in premature newborns, especially with an unusual neonatal course inordinate to lung disease. The use of echo might have led to an early diagnosis of this condition in such patients.

### Authors' contributions

Dr. Alkhazzam and Dr. Gad reviewed the literature and drafted the first manuscript; Dr. Rahmath provided the echo images and reviewed the manuscript. All authors approved the last version of the manuscript.

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