Shone’s syndrome diagnosed with echocardiography and confirmed at pathology

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The Shone’s complex, defined by four cardiovascular defects such as a supravalvular mitral membrane, valvular mitral stenosis by a parachute mitral valve, subaortic stenosis, and aortic coarctation, is a rare entity, which occurs most frequently in its incomplete form. We report the case of a 19-year-old female patient who presented at the emergency room for progressively worsening dyspnoea, orthopnoea, fever, and productive cough, due to bronchopneumonia. Echocardiography revealed the co-existence of aortic coarctation with bicuspid aortic valves, mitral supravalvular ring, and dysplastic mitral valves producing severe mitral stenosis and severe pulmonary hypertension. Although wide spectrum antibiotics were administered from the first day of hospitalization, the patient developed severe sepsis and died. The components of the Shone’s complex diagnosed by echocardiography were confirmed at pathology.

KEYWORDS
Shone’s complex; Supramitral ring; Mitral stenosis; Aortic coarctation; Pulmonary hypertension

Patients with Shone’s syndrome present four cardiovascular defects: a supravalvular mitral membrane, valvular mitral stenosis due to a parachute mitral valve, subaortic stenosis (membranous or muscular), and aortic coarctation.¹ When only two or three of these components are present, the incomplete form of Shone’s syndrome is diagnosed. A small series of case studies published since the syndrome was first described highlight important problems of diagnosis, treatment, and prognosis.

Case report

A 19-year-old patient presented at the Emergency Room for progressively worsening dyspnoea installed 3 days before, with orthopnoea, fever and productive cough. Six years before, mildly increased blood pressure values led to the diagnosis of aortic coarctation, with an echocardiographic peak Doppler gradient of 74 mm Hg. Severe pulmonary hypertension (HTN) was also diagnosed by echocardiography, and cardiac catheterization was performed at that time in another department. It confirmed severe pulmonary HTN (systolic pulmonary artery pressure of 145 mm Hg) with severely increased pulmonary resistance. Aortic coarctation was also confirmed. No evidence of intracardiac shunt or other abnormalities were described and a diagnosis of primary pulmonary HTN was made. There was no mention about mitral valve obstruction in the discharge summary note. Aortic coarctation was successfully treated by endoluminal balloon dilation, with a low residual gradient. Blood pressure values returned to normal, but functional capacity remained limited (New York Heart Association functional classes II–III). The patient or her family did not seek any medical advice until the current episode.

At physical examination, the patient was pale and febrile, with basal crackles over both lung fields. Cardiac auscultation revealed tachycardia (130 bpm, regular), a loud S₂ and intense diastolic murmur over the upper left parasternal border, a loud diastolic rumble at the apex, and intense systolic murmur over the tricuspid area. The liver was enlarged and the patient did not have peripheral oedema.

The ECG showed sinus tachycardia (130 bpm), biatrial enlargement, and signs of right ventricular (RV) hypertrophy. Chest X-ray showed a mildly increased cardio-thoracic index and diffuse, confluent alveolar opacities suggesting, in clinical context, the diagnosis of bronchopneumonia. Complete blood count revealed anaemia with haemoglobin value of 10.1 g/dL and leucocytosis of 22.900 per mm³. Liver enzyme values were slightly increased, and renal function tests were normal. Transthoracic echocardiography (TTE) revealed the presence of a supravalvular mitral diaphragm, with a narrow opening of 6–7 mm (Figure 1 and
Supplementary data online, Video S1), inserted very close to the mitral valves, producing severe mitral stenosis (mean transmitial gradient of 28 mmHg) (Figure 2). The patient had severe tricuspid regurgitation secondary to severe pulmonary HTN (peak RV–right atrium gradient of 139 mmHg). The enlarged and hypertrophied RV (free wall 12 mm) and the flattened interventricular septum (Supplementary data online, Video S2) confirmed the severity and long-standing history of pulmonary HTN. Transthoracic echocardiography also established the presence of bicuspid aortic valve (Supplementary data online, Video S3) and hypoplastic ascending aorta (aortic sinotubular junction 14 mm, ascending aorta 21 mm, see Supplementary data online, Video S1; aortic arch of 14 mm), with a residual peak gradient at coarctation site of 24 mm Hg. The persistence of left superior vena cava, suggested at TTE by the severely dilated coronary sinus (Supplementary data online, Video S1) was confirmed by contrast echocardiography. Transesophageal echocardiography excluded the existence of interatrial or interventricular shunts. It also delineated the narrow, flow-limiting orifice at the level of supravalvular mitral ring (Supplementary data online, Video S4).

Although wide spectrum antibiotics were administered from the first day of hospitalization, the patient remained febrile and on the fifth day developed severe oxygen desaturation requiring mechanical ventilation. Penicillin-resistant Pneumococcus was found in the sputum and bronchoalveolar lavage. After a few days, the patient developed severe sepsis and died. At pathology, the presence of aortic coarctation (Figure 3) and bicuspid aortic valves (Figure 4) was confirmed. Left atriotomy revealed the presence of a supramitral fibrous ring, delineating a 7 mm diameter orifice (Figure 5), as well as a dysplastic (thickened, myxomatous) mitral valve (Figure 6), with a narrow valvular orifice, indicating the presence of both supravalvular and valvular mitral stenosis.

Figure 1 Transthoracic echocardiography, parasternal long-axis view: end-diastolic frame showing a diaphragm on the atrial side of the mitral valve. A central orifice with a diameter of 7.2 mm can be seen (calipers). The white arrows point to the mitral leaflets.

Figure 2 Continuous-wave Doppler transmitial flow: mean gradient of 28 mm Hg.

Figure 3 Specimen of the proximal descending aorta confirming the existence of aortic coarctation (arrows).

Figure 4 Bicuspid aortic valves with a median raphe (arrow), seen from the aorta.

Figure 5 Supravalvular mitral ring visualized from the left atrium (double-headed arrow: inner diameter of ~7 mm).
Severe RV hypertrophy was confirmed (Figure 7), and histological changes consistent with long-standing pulmonary HTN were also found.

**Discussion**

The Shone’s complex is a very rare entity (four cases have been discovered among 12,520 echo studies in a series published by Zucker et al.), which is diagnosed most frequently in its incomplete form. Our patient had aortic coarctation, bicuspid aortic valves with no left ventricular (LV) outflow tract obstruction, and mitral supravalvular ring associated with a dysplastic mitral valve, causing severe mitral stenosis and severe pulmonary HTN. In addition, this is, to our knowledge, the first case study to report the association of persistent left superior vena cava to the Shone’s syndrome.

Mitral valve obstruction during early embryogenesis is considered the first pathological event in Shone’s syndrome, causing underdevelopment of the LV cavity, thus leading to various degrees of LV outflow tract obstruction and aortic coarctation. The pathological findings of this entity should be differentiated from cor triatriatum sinister since the embryological origin, morphology, and surgical implications are different in these two malformations. The embryologic origin of supramitral ring is unclear; but is considered the result of incomplete division of endocardial cushion tissue, while cor triatriatum is believed to be a result of incomplete absorption of primary pulmonary vein during the fifth embryonic week.

Patients with Shone’s syndrome have a poor long-term prognosis, with a perioperative mortality rate of 24–27%, often requiring multiple interventions at an early age. In their first description of the syndrome, Shone et al. noted that mitral valve obstruction appeared to be the most critical lesion. Other studies confirmed that the severity of mitral valve obstruction correlates with poor long-term outcome. Patients with the most severe forms of mitral valve obstruction presented with severely elevated pulmonary artery pressure and had the poorest prognosis.

In patients with aortic coarctation, the partial disappearance of elastic tissue at the level of aortic media seems to be an important feature. Aortic coarctation occurs in 20–59% of cases with mitral valve anomalies, whereas the mitral supravalvular ring is associated with other defects in almost 90% of cases. Therefore, the finding of these defects should prompt for search of other cardiac and vascular anomalies.

Our patient had previously unrecognized severe mitral valve obstruction with severe pulmonary HTN and in this context the occurrence of severe bronchopneumonia and the development of sepsis were proved to be fatal. We had no detailed data about the measurements performed during the initial examination, but it seems that unfortunately the diagnosis of mitral valve obstruction was overlooked at that time.

This case study raises the awareness about this rare syndrome and highlights the importance of a carefully performed echocardiogram particularly in patients with congenital heart disease where lesions often co-exist and can be missed in the absence of a comprehensive examination.

**Supplementary data**

Supplementary data are available at European Journal of Echocardiography online.

**References**