Adult patient with Shone's syndrome and patent ductus arteriosus: a case report

Shone sendromu ve duktus arteriyoz açıklığı olan erişkin hasta: Olgu sunumu

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Summary– Shone's syndrome is a very rare cardiac malformation composed of four obstructive congenital lesions, which include the parachute mitral valve (PMV), supravalvular ring, subaortic stenosis, and coarctation of aorta. Diagnosis requires a comprehensive examination including a carefully performed echocardiography. It is extremely unusual for a patient to remain undiagnosed during adulthood. Here we report a 26-year-old male patient with Shone's syndrome and patent ductus arteriosus (PDA). The patient reported that he had been suffering from exercise intolerance and aggravating dyspnea. Two years ago, he was operated on in another hospital based on the wrong diagnosis of mitral valve prolapse and subaortic membrane. Transthoracic echocardiography revealed the existence of a PMV, which led to severe mitral stenosis through a previously implanted mitral annular ring. Other components of Shone's syndrome, subaortic membrane and aortic coarctation, together with PDA, were also observed. Another operation was suggested, but the patient refused. During the two year follow-up period, in spite of an atrial fibrillation attack and deteriorating dyspnea, the patient still did not want another operation. Once any of the four components of the Shone's complex is detected, clinicians must look for the presence of other lesions.

Özet– Shone sendromu paraşüt mitral kapak (PMV), supravalvüler halka, subaortik darlık ve aort koarktasyonu gibi dört tıkayıcı doğumsal lezyondan oluşan çok nadir bir kardiyak malformasyondur. Tanı, titizlikle yapılacak kapsamlı bir ekokardiyografik inceleme gerektirir. Bir hastanın, erişkin yaşamda halen tanınmamış olması oldukça ender rastlanan bir durumdur. Bu yazıda, Shone sendromu ve patent duktus arteriyozusu (PDA) olan 26 yaşında erkek hasta sunuldu. Hasta egzersiz intoleransı ve şiddetlenen nefes darlığından yakınmaktaydı. İki yıl önce başka bir merkezde tanısının atlandığı ve mitral kapak prolapsusu ve subaortik membran gibi yanlış tanılara dayanılarak ameliyat edildiği öğrenildi. Transtorasik ekokardiyografide daha önce yerleştirilen mitral anüler halka nedeniyle ciddi mitral darlığa yol açan PMV saptandı. Shone sendromunun diğer bileşenleri olan subaortik membran, aort koarktasyonu ve ek olarak PDA gözlendi. Tekrar ameliyat önerildi, ancak hasta kabul etmedi. İki yıllık izlem sırasında, atriyal fibrilasyon atağı ve kötüleşen nefes darlığına rağmen hasta tekrar ameliyat olmayı yine kabul etmedi. Shone sendromuna ait herhangi bir unsur saptandığında, klinisyen diğer unsurların varlığını mutlaka araştırmalıdır.

Shone's syndrome is a very rare congenital cardiac malformation defined by four cardiovascular defects leading to left side heart obstruction at multiple levels. It is composed of the supravalvular mitral ring, valvular mitral stenosis (MS) due to parachute mitral valve (PMV), subaortic membrane, and aortic coarctation.[1] Few cases have been reported since this syndrome was first defined, and very few of these have been adult patients.

Abbreviations:

CHD	*Congenital heart disease*
MS	*Mitral stenosis*
PDA	*Patent ductus arteriosus*
PMV	*Parachute mitral valve*

The diagnosis of Shone's complex should be considered and further exploration is required when any of the four pathologies is observed during the echocardiographic study.

Here we report an undiagnosed Shone's syndrome and patent ductus arteriosus (PDA) in a 26-year-old

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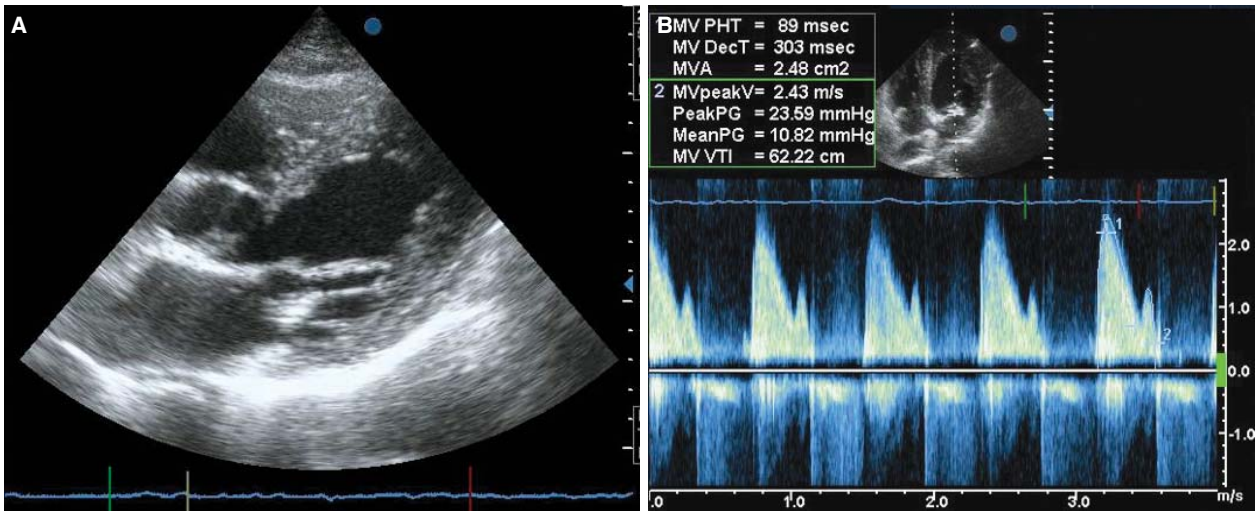

Figure 1. **(A)** Image demonstrating the parachute like mitral valve and the single papillary muscle (apical four chamber window). **(B)** Mitral valve area calculated via pressure half time method indicating moderate mitral valve stenosis. The diastolic gradient through the mitral valve was also significant (see supplementary video file 1).

patient who was operated on two years ago due to mitral valve prolapse and subaortic membrane.

CASE REPORT

A 26-year-old man presented to our hospital due to ongoing exercise intolerance after surgery. He was operated on two years ago in another center due to mitral valve prolapse and severe mitral regurgitation and associated subaortic membrane. The patient's discharge summary note from that center showed that the implantation of a mitral annular ring and subaortic membrane resection were performed, but no other pathologies were defined.

Upon examination, the patient's pulse was weak in both lower limbs, while his pulse was strong in both upper extremities. His blood pressure was 160/110 mmHg in the right upper extremity. Cardiac auscultation revealed that the first heart sound was loud, and there was a long ejection systolic murmur (grade 3/6) at the aortic and mitral areas. There was also a continuous murmur near the scapula. There was no evidence of right heart failure. Electrocardiography showed signs of right and left ventricular hypertrophy.

Transthoracic echocardiography revealed the existence of a PMV leading to MS through the mitral annular ring (Fig. 1a, b, see supplementary video file 1). The mean gradient between the left atrium and the left ventricle was 11 mmHg. Mild left ventricular outflow obstruction via recurrent subaortic membrane with a 35 mmHg peak gradient and a mean gradient of 16 mmHg was also observed (Fig. 2, see supplementary video file 2). The patient's pulmonary artery pressure was calculated to be 45 mmHg from the Doppler study of tricuspid regurgitation. His left ventricular systolic function and ejection fraction was normal. The parasternal short axis view revealed reverse flow at the pulmonary artery, which is indicative of PDA (Fig. 3, see supplementary video file 3). The suprasternal view of the aortic arc revealed aortic coarctation (Fig. 4a, b, see supplementary video file 4). The peak Doppler gradient of aortic coarcta-

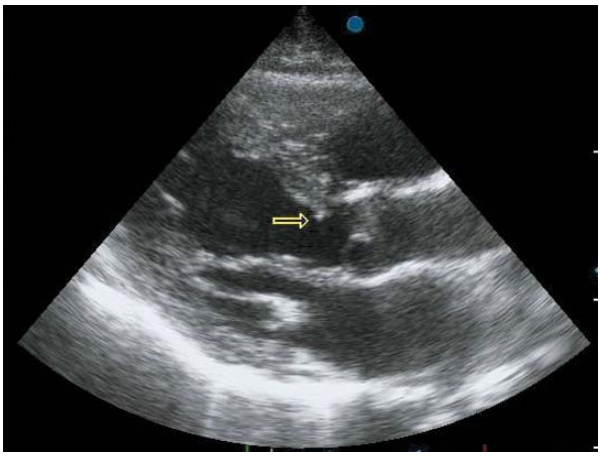

Figure 2. Parasternal long-axis view of the left ventricular outflow tract shows a remnant of the previously excised subaortic membrane (arrow) (apical five chamber view). (see supplementary video file 2).

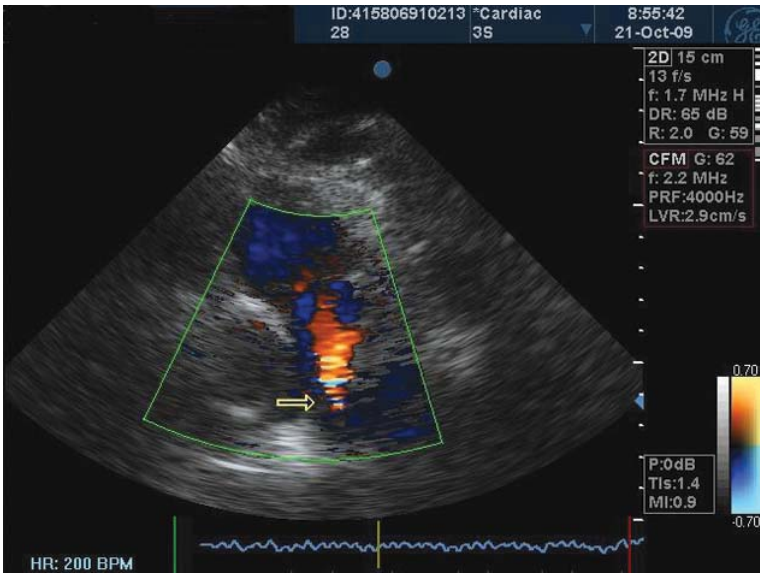

Figure 3. The parasternal short axis view showing the retrograde color Doppler flow in the pulmonary artery concordant with patent ductus arteriousus. (see supplementary video file 3).

tion was calculated to be 82 mmHg. It was decided that cardiac surgery was necessary, but the patient refused the surgery. During the two year follow-up period, he was admitted once with a new onset of atrial fibrillation, and medical cardioversion was successful with amiodaron. After a few months, his diuretic therapy was augmented because of congestive symptoms. Again, operation was suggested, but he refused.

DISCUSSION

Shone's syndrome, a very rare congenital heart disease (CHD), was first described by Shone et al in 1963.[1] The incidence of Shone's syndrome is 0,6% of all CHDs.[2] It is characterized by four obstructive lesions on the left side of the heart, which include the parachute-like mitral valve, supravalvular mitral ring, subaortic stenosis, and coarctation of the aorta.

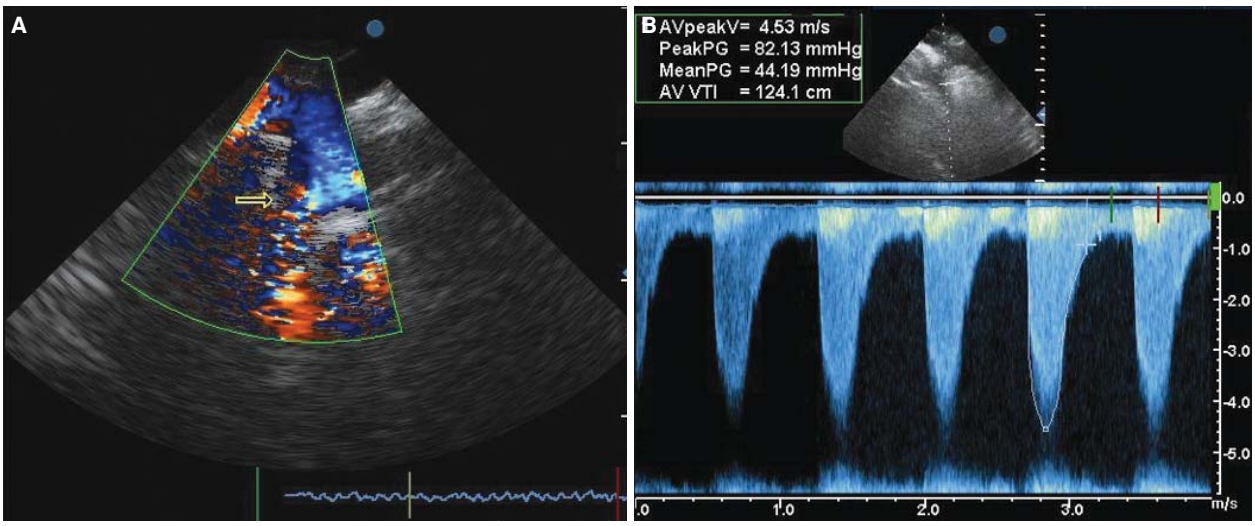

Figure 4. **(A)** The color Doppler image showing the aortic coarctation in the descending aorta (suprasternal view). **(B)** The continuous wave Doppler study demonstrating the significant gradient between the proximal and the distal part of the aortic coarctation in the descending aorta (suprasternal view). (see supplementary video file 4).

Among these, the severity of MS is the most significant indicator of survival and long term prognosis.[2]

During the early phase of embryogenesis, mitral valve obstruction is considered to be the first pathological event in Shone's syndrome.[2] Mitral valve obstruction is thought to cause impairment of left ventricular cavity development, which in turn leads to the other pathological features of left ventricular outflow obstructions and aortic coarctation.

In the literature, less than 100 cases have been reported, and while a few cases of adult patients have been described, most of the reports were among the pediatric population. Shone's syndrome is typically diagnosed during childhood. As the patient ages, it becomes symptomatic. Symptoms and signs like dyspnea, tachypnea, exercise intolerance, fatigue, and nocturnal cough may be experienced. During childhood, a failure to thrive and recurrent episodes of respiratory tract infections may be some indicators for this kind of CHD.

Patients with Shone's syndrome have a poor long term prognosis. It is well established that the severity of the mitral valve obstruction correlates with elevated pulmonary artery pressure and poor long term outcome.[3,4] Patients have a perioperative mortality rate of 24-27%, and they often require multiple interventions at an early age.[5]

It is unusual for a patient to remain undiagnosed throughout childhood, and it is extremely unusual to remain undiagnosed in adulthood after cardiologic evaluation and cardiac surgery. Unfortunately for our patient, the diagnosis of parachute like mitral valve, PDA, and aortic coarctation was overlooked at the time of surgery. Since it was not mentioned in the discharge note, we do not know whether the patient had a congenital supravalvular ring or a membrane before the surgery. However, it seems that the Shone's complex was iatrogenically completed after the mitral annular ring implantation surgery, which caused severe MS due to a PMV.

This case study highlights the importance of a careful echocardiographic evaluation, including all echocardiographic windows, particularly in patients with CHDs. Diagnosis of any of the four components of a Shone's complex should prompt a clinician to investigate the patient in detail, and co-existing pathologies should be questioned. Awareness of this syndrome and comprehensive examinations of patients with CHDs should lower the probability of missing the diagnosis of co-existing pathologies.

Supplementary video files associated with this article can be found in the online version.

Conflict-of-interest issues regarding the authorship or article: None declared

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Key words: Abnormalities, multiple; aortic coarctation; heart defects, congenital; mitral valve/abnormalities; mitral valve stenosis/congenital; mitral valve stenosis; parachute mitral valve; patent ductus arteriosus.

Anahtar sözcükler: Anomaliler, çoklu; aort koarktasyonu; kalp defekti, doğuştan; mitral kapak/anormallik; mitral kapak darlığı/doğuştan; mitral kapak darlığı; paraşüt mitral kapak; duktus arteriyozus açıklığı.