

Prenatal Diagnosis of a Rare Congenital Heart Disease: Shone's Complex: Case Report

Nadir Bir Konjenital Kalp Hastalığının Prenatal Tanısı: Shone Kompleksi

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ABSTRACT Congenital heart diseases are one of the most common congenital malformations identified in prenatal period and cause considerable anxiety for both parents and clinicians. Shone's complex is a rare congenital heart disease first described by Shone et al. The syndrome consists of four defects; a supra-valvar mitral membrane, parachute mitral valve, subaortic stenosis and coarctation of aorta. An incomplete form has been described when only two or three of the components are recognized. In this report we present (with the informed consent of the patient) a prenatally diagnosed incomplete form of Shone's complex. Caring for these families would require input from a multidisciplinary team involving obstetricians, neonatologists, pediatric cardiologists and cardiovascular surgeons, which is also known as hybrid approach. For the well being of the patient this approach is thought to be essential.

Key Words: Heart defects, congenital; prenatal diagnosis; ultrasonography

ÖZET Konjenital kalp hastalıkları prenatal dönemde en sık tespit edilen konjenital malformasyonlardan biri olup hem aile hem de hekimlerde endişeye neden olmaktadır. Shone kompleksi nadir görülen bir konjenital kalp hastalığı olup ilk defa Shone ve ark. tarafından tanımlanmıştır. Sendrom dört kusurdan meydana gelir; supra-valvüler mitral membran, paraşüt mitral kapak, subaortik darlık ve aort koarktasyonu. Sendromun inkomplet formu, sendrom bileşenlerinin sadece iki ya da üç tanesinin bulunması ile tanımlanır. Burada, (hastanın bilgilendirilmiş onamı ile birlikte) prenatal dönemde tanı alan Shone kompleksin inkomplet formu sunulmaktadır. Bu ailelerin takibi; obstetrisyen, neonatolog, pediatrik kardiyolog ve kardiyovasküler cerrahlardan oluşan hibrid yaklaşım olarak da bilinen multidisipliner bir ekip çalışmasını gerektirmektedir. Hastanın iyiliği için bu yaklaşımın gerekli olduğu düşünülmektedir.

Anahtar Kelimeler: Kalp kusurları, doğumsal; prenatal tanı; ultrasonografi

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Shone's complex is a rare congenital heart disease first described by Shone et al. in 1963.¹ The syndrome consists of four defects; a supra-valvar mitral membrane, parachute mitral valve, subaortic stenosis and coarctation of aorta.¹ An incomplete form has been described when only two or three of the components are recognized.¹ Only four cases of Shone's complex have been reported in the literature, mostly reporting the surgical outcomes. In this study, we describe a prenatally diagnosed incomplete form of Shone's complex, antenatal history, echocardiographic findings and postnatal outcome.

CASE REPORT

A 30-year-old multiparous pregnant woman with postpartum death 1 and living 2 was referred to our hospital after detection of abnormal cardiac findings at 30th weeks of gestation. In her antenatal history, 2nd trimester aneuploidy screening, morphology screening were normal. Her second baby died at the 24th day of life due to an unidentified etiology, the other two living children had no medical problems. History was negative for any drug consumption, tobacco or ethanol. Family history was also negative for anomalies. She didn't have consanguinity with her husband.

The fetal echocardiography revealed incomplete form of Shone's complex with mitral stenosis (Figure 1a and 1b), aortic stenosis, left ventricular endocardial fibroelastosis (Figure 2), restricted mitral valve motions with shortness of chordae. Mitral annulus was 9 mm, left ventricle length was 17 mm. Velocity of flow at aorta was 2.8 m/sec.

Termination of pregnancy was not considered due to advanced gestational age. The patient was consulted with neonatologists, cardiovascular surgeons and they agreed to follow up the patient until delivery.

At the 39th week patient admitted to delivery room due to increased uterine contractions. She delivered a male baby through vaginal route weighting 3810 g with 9 and 10 Apgar scores at 1st and 5th minutes, respectively. His cord blood pH was 7.41. The baby's physical examination revealed perioral cyanosis and 2-3/6 degree systolic murmur. The postnatal echocardiography showed severe aortic stenosis, mitral stenosis, bicuspid aortic valve, fibrotic aortic valves, rudimentary papillary muscles, short and thickened chordae and patent ductus arteriosus with reverse flow. Aortic annulus 6 mm, interatrial septal defect with a diameter of 3-4 mm was observed. Interventricular septum was intact. At postnatal echocardiography with the absence of aortic coarctation the diagnosis of incomplete Shone's syndrome was confirmed. The neonate received prostaglandine infusion. Balloon valvuloplasty was planned by the cardiovascular surgeons. However family refused the intervention because

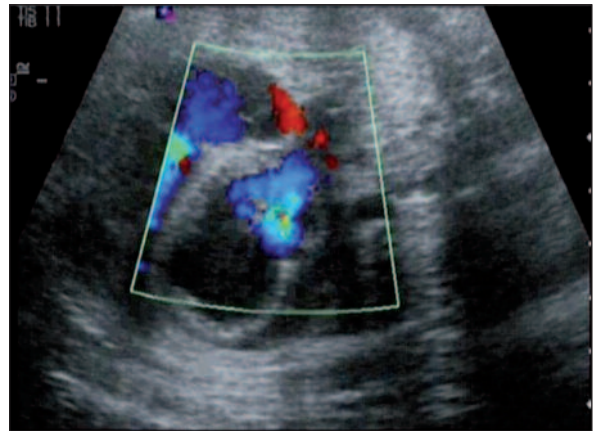


FIGURE 1A: Fetal echocardiography showing mitral stenosis with insufficient influx during ventricular filling phase.
(See for colored form <http://jinekoloji.turkiyeklinikleri.com/>)

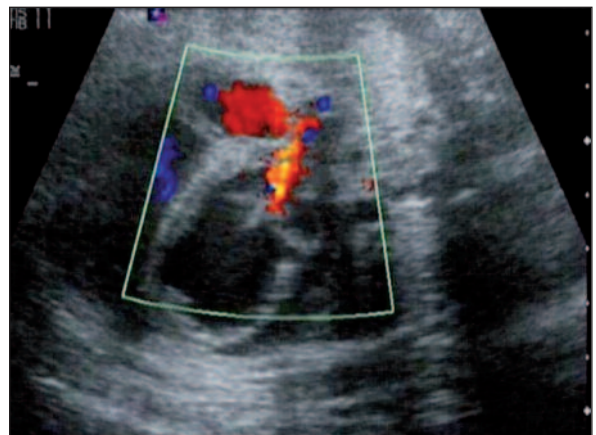


FIGURE 1B: Fetal echocardiography showing efflux during ventricular systole due to mitral valve insufficiency accompanying mitral stenosis.
(See for colored form <http://jinekoloji.turkiyeklinikleri.com/>)



FIGURE 2: Fetal echocardiography showing left ventricular endocardial fibroelastosis (arrow).

of their religious beliefs and the baby died at postpartum 17th day.

DISCUSSION

The Shone's complex is a very rare congenital heart disease in medical literature only four cases diagnosed among 12.520 fetal echocardiographies in a study by Zucker et al.² The syndrome is mostly seen in its incomplete form. The main pathology *in-utero* is thought to be the obstruction of mitral valve causing obstruction of left ventricular outflow and underdevelopment of the left ventricle.² In the study by Shone et al. it has been shown that degree of mitral valve obstruction is the most important prognostic factor for long term outcome.¹ Since our patient had moderate-severe degree of mitral valve obstruction, the long term outcome was poor. Despite that poor prognosis, the pregnancy had continued due to the advanced gestational age at initial diagnosis.

Our patient had incomplete form with mitral stenosis, parachute mitral valve, aortic stenosis and no coarctation of aorta. Endocardial fibroelastosis which is considered as a poor prognostic factor was also present. Neonatal echocardiogram also showed that the left ventricle with a size smaller than 5th percentile which was also detected in all cases in the study of Zucker et al.² Additional diagnosis included in postnatal echocardiography: interatrial septal defect, bicuspid aortic valve, systolic dysfunction and patent ductus arteriosus and reverse flow. Bicuspid aortic valve which was also detected in all Shone's complex patients in a study by Zucker et al.²

Patients with Shone's syndrome have a wide spectrum of outcome. In study by Zucker et al., two babies developed symptoms during the first week of life due to coarctation of the aorta and underwent coarctation repair.² The third patient underwent coarctation repair later in life.² The fourth

one was asymptomatic.² All patients were alive after a mean follow-up of 7.8 (range, 3.3–10.5) years.² In our study the patient was symptomatic but parents refused the intervention because of their religious beliefs, the result was the death of the baby on the seventieth day of life. 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.^{3,4} This variability in outcome causes difficulty in predicting neonatal outcome and counseling with parents.

It is hard to prognosticate accurately when counseling parents and only postnatal echocardiography and long-lasting clinical and echocardiographic follow-up eventually determine the correct nature of this lesion.²

Late outcome in Shone's anomaly seems to correlate with the predominance of mitral valve involvement and the degree of pulmonary hypertension. Valve repair is indicated whenever feasible and should be considered before the occurrence of pulmonary hypertension as it is shown in the studies of Brauner et al.⁴ In addition, the accrued surgical experience with this disease is very limited, and consists mainly of case reports.^{5,6}

To conclude, hybrid approach for congenital cardiac defects would be appropriate for those patients in which congenital cardiac surgeon and pediatric cardiologist having the will to co-operate simultaneously and collaborate to each other as a single team for the well being of the patient.⁷ Despite high surgical risk and considerable early mortality, late outcome in the majority of patients is favorable. We believe that an aggressive reconstructive approach to both left ventricular inflow and outflow lesions may further extend event-free survival in these critically ill patients.

REFERENCES

1. Shone JD, Sellers RD, Anderson RC, Adams P Jr, Lillehei CW, Edwards JE. The developmental complex of "parachute mitral valve," supralvalvular ring of left atrium, subaortic stenosis, and coarctation of aorta. *Am J Cardiol* 1963;11:714-25.
2. Zucker N, Levitas A, Zalstein E. Prenatal diagnosis of Shone's syndrome: parental counseling and clinical outcome. *Ultrasound Obstet Gynecol* 2004;24(6):629-32.
3. Bolling SF, Iannettoni MD, Dick M 2nd, Rosenthal A, Bove EL. Shone's anomaly: operative results and late outcome. *Ann Thorac Surg* 1990;49(6):887-93.
4. Brauner RA, Laks H, Drinkwater DC Jr, Scholl F, McCaffery S. Multiple left heart obstructions (Shone's anomaly) with mitral valve involvement: long-term surgical outcome. *Ann Thorac Surg* 1997;64(3):721-9.
5. Banerjee A, Kohl T, Silverman NH. Echocardiographic evaluation of congenital mitral valve anomalies in children. *Am J Cardiol* 1995; 76(17):1284-91.
6. Celano V, Pieroni DR, Morera JA, Roland JM, Gingell RL. Two-dimensional echocardiographic examination of mitral valve abnormalities associated with coarctation of the aorta. *Circulation* 1984;69(5):924-32.
7. Aydemir NA, Erdem A. [Hybrid approach in congenital cardiac diseases]. *Turkiye Klinikleri J Cardiovasc Surg-Special Topics* 2011; 3(1):71-7.