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Analysis of the patients diagnosed as hypoplastic left heart syndrome at prenatal period

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Abstract

Objective: The aim of this study was to analyze patients together with their demographic data who are diagnosed with hypoplastic left heart syndrome at intrauterine period.

Methods: In this study, 29 pregnant women who were referred from other centers to or followed-up in the Divisions of Perinatology of Ankara and Adana hospitals of Başkent University between June 2009 and February 2013 were analyzed in terms of their demographic data, diagnosis weeks, concomitant anomalies and the progress of their pregnancies.

Results: Among these cases, there was only one twin pregnancy. Three pregnancies were obtained by assisted reproductive technology (10.3%). When concomitant anomalies were analyzed, additional cardiac anomaly was found as 27.6% (8/29), hydrops as 13.4% (4/29) and single umbilical artery as 10% (2/29). Ten patients (34%) who were diagnosed in our hospital between 18 and 22 weeks of gestation preferred termination. Other patients were just referred to our hospital for delivery by being diagnosed at late period. While 4 (13.7%) of these babies could be in a condition to be operated after delivery, one of two surviving patients waited for second operation and the other one waited for third operation.

Conclusion: In hypoplastic left heart syndrome, to be able to have an operation and chance to survive after delivery seems still low in our country. Early prenatal diagnosis is significant in terms of proper briefing family and the decision for termination.

Keywords: Hypoplastic left heart, diagnosis, prognosis.

Özet: Prenatal dönemde hipolastik sol kalp sendromu tanısı alan hastaların değerlendirilmesi

Amaç: Bu çalışmanın amacı, intrauterin dönemde hipoplastik sol kalp sendromu tanısı almış hastaların demografik verileri ile birlikte incelenmesidir.

Yöntem: Bu çalışmada Haziran 2009 ve Şubat 2013 tarihleri arasında Başkent Üniversitesi Ankara ve Adana hastaneleri Perinatoloji Bilim dalına dışarıdan refere veya takip edilen 29 gebe demografik verileri, tanı konulma haftaları, eşlik eden anomaliler ve gebeliklerin seyri açısından değerlendirildi.

Bulgular: Bu hastalardan sadece biri ikiz gebelikti. Üç gebelik yardımcı üreme teknikleri ile elde edilmişti (%10.3). Eşlik eden anomaliler incelendiğinde; %27.6 (8/29) ek kardiyak anomali, %13.4 (4/29) hidrops ve %10 (3/29) tek umblikal arter saptandı. 18-22 haftalar arası hastanemizde tanı alan 10 hasta (%34) terminasyonu seçmişti. Diğer başvuran hastalar ise geç dönem tanı alarak sadece doğum için refere edilmişti. Bu bebeklerden doğum sonrası 4'ü (%13.7) opere olabilecek duruma gelebilirken halen yaşamını sürdüren 2 hastadan biri 2. operasyon ve diğeri ise 3. operasyon için beklemektedir.

Sonuç: Hipoplastik sol kalp sendromunda doğum sonrası operasyona gidebilme ve yaşam şansı ülkemiz şartlarında halen çok düşük görünmektedir. Erken prenatal tanı, ailenin doğru bilgilendirilmesi ve terminasyon kararı açısından önemlidir.

Anahtar sözcükler: Hipoplastik sol kalp, tanı, prognoz.

Introduction

Hypoplastic left heart syndrome (HLHS) is defined as the hypoplasia of the left side of the heart where mitral atresia, aortic atresia, aortic stenosis and coarctation of aorta may also be seen as well.[1] It constitutes 7-9% of congenital cardiac diseases.[2] It is seen in 0.016% to 0.036% of live births,[3-6] and it is responsible for 25% of cardiac-associated deaths in the first week of life.[7]

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Prenatal diagnosis is based on the fetal echocardiography to be performed between 18 and 24 weeks of gestation.[8,9] In four-chamber view of the heart, ventricle seen as a small globular or narrow gap with endocardial fibroelastosis can be detected.

Various chromosomal diseases (i.e. Turner syndrome) or syndromes (i.e. Noonan syndrome, Smith-Lemli-Opitz syndrome and Holt-Oram syndrome) may accompany the disease.[10,11] Extracardiac anomalies such as omphalocele, corpus callosum agenesis and hydrops fetalis may be observed.[12] Also, additional cardiac defects such as transposition of the great arteries, atrial isomerism, and total abnormal pulmonary venous return anomaly may be observed (7.5%).[13] Today, there are two therapy approaches which are three-step series of univentricular palliation and cardiac transplantation.[14]

In this study, we aimed to analyze retrospectively the demographic data, diagnosis weeks, concomitant anomalies and the progress of the pregnancies of the patients who were diagnosed as HLHS at intrauterine period.

Methods

In this study, 29 pregnant women who were referred from other centers to or followed-up in the Divisions of Perinatology of Ankara and Adana hospitals of Başkent University between June 2009 and February 2013 were analyzed in terms of their demographic data, diagnosis weeks, concomitant anomalies and the progress of their pregnancies. Statistical data was evaluated by SPSS 16.0 (SPSS Inc., Chicago, IL, USA).

Results

Only 10.3% of mothers were 35 years old and above, and only one of them had twin pregnancy. Three pregnancies were obtained by assisted reproductive technology (10.3%). When concomitant anomalies were analyzed, additional cardiac anomaly was found as 27.6% (8/29), hydrops as 13.4% (4/29) and single umbilical artery as 10% (2/29). Only one patient had trisomy 21. Nineteen (65.5%) patients were diagnosed between 18 and 24 weeks of gestation but only ten of them (34%) preferred termination. Other patients were just referred to our hospital for delivery by being diagnosed at late period. While 4 (13.7%) of these babies were operated after the delivery, one of two surviving patients has been waiting for second operation and the other one has been waiting for third operation.

Discussion

For hypoplastic left heart syndrome (HLHS) which responsible for 25% of the cardiac-associated deaths in the first week of life, early diagnosis is very significant to decide for maintaining pregnancy or termination.[7]

Optimal diagnosis is established by examining fetal heart ultrasonographically between 18 and 24 weeks of gestation. In four-chamber view of the heart, ventricle seen as a small globular or narrow gap with endocardial fibroelastosis can be detected (**Fig. 1**).[8,9] By the Doppler ultrasonography examination in four-chamber view, it can be seen that left ventricle cavity is not filled (**Fig. 2**), the reverse flow from foramen ovale and even the flow of mitral insufficiency in cases where mitral valve is dysplastic can be observed.

Also, additional cardiac defects such as transposition of the great arteries, atrial isomerism, and total abnormal pulmonary venous return anomaly may be observed together with HLHS (7.5%).[13]

In our study, additional cardiac anomalies were detected in 8 cases (25.7%) which were ventricular septal defect in 3 cases, tricuspid insufficiency in 2 cases, mitral stenosis in one case and abnormal pulmonary venous return anomaly.

It is significant to share the negative impact of associated chromosomal and extracardiac malformations on the prognosis with the family.[15] In a study performed by Allen et al. on 176 cases in 2005, karyotype analyses were conducted on the fetuses (22%) diagnosed. Among these cases, chromosomal anomaly was found in 42% of them.[16] In our case, trisomy 21 was found only in one case as a chromosomal anomaly concomitant to HLHS. Schulz published a case concomitant to Noonan syndrome associated with RAF1 gene mutation.[17] In our study, extracardiac anomalies (such as hydrops, single umbilical artery) were found in 17 patients (58.6%). The rate of extracardiac anomaly such as corpus callosum agenesis, diaphragmatic hernia and omphalocele etc. has been reported between 12 and 37% in the literature.[7,12,18-21] We had only four cases decided to maintain the pregnancy and have the chance for operation (13.7%). Two of them are still alive and wait for following operations. During this three-step operation process, first step is Norwood operation. The purpose in this operation is to enable systemic and pulmonary circulation in a balanced way without any obstruction.[22]

The second operation is the step for establishing cavopulmonary shunt performed at 4th-6th months. This procedure is also called as bidirectional Glenn shunt; it is a less risky operation compared to the first step and has a chance of success about 96-99%.[22] One of our patients is prepared for this operation.

The third step is the Fontan operation. It is aimed in this operation to create total cavopulmonary connection. Mortality rate after this operation is reported as about 3-4%.[23]

In a study carried out in 14 centers, 1 year survival rate reported for triple palliation and transplantation operations varies between 30-80% and 5-90%, respectively.[18] Cardiac transplantation may have a life-saver role at each step in the patients who undergone palliation.[24] The rate to reach an adult age is still below 30%.[13,25]

Neurological problems such as learning disability, low IQ level, hyperactivity and lack of attention may be seen in patients with HLHS.[26-31]

Structural and functional brain anomalies may be observed in patients with congenital cardiac disease even during birth. This is probably because of the variability of blood flow on brain and it varies according to the type of congenital cardiac disease.[32-37] Uncertainties in neurological development and long-term life prognosis affect the final decision of parents about fetus.[38]

Szwast et al. found that fetuses with hypoplastic left heart syndrome decreased cerebrovascular resistance in order to maintain cerebral blood flow at third trimester.[39] Information provided to families is very significant in terms of the expectations of families and the progress of pregnancy. Tibballs et al. stated that the rate to choose surgical treatment in patients who are established prenatal diagnosis during pregnancy is higher than the rate of those established postnatal diagnosis (94% and 47%, respectively).[32]

It is all significant in terms of termination of pregnancy, informing family about disease, termination, maintenance of pregnancy and the problems to be faced in future. Two hundred and eleven families who had fetuses with HLHS participated in a survey. Sixty-five percent of them were diagnosed at prenatal period, and 34% of them decided to terminate the pregnancy. Informing families about termination again in the subsequent interviews considerably decreased their optimistic ideas about the life of fetus. Twenty-two percent of the parents stated that they felt a pressure from physicians to terminate the gestation. Thirty-eight percent of the parents looked for a second option, and 69% of them consulted different individuals to choose a new physician.[40]

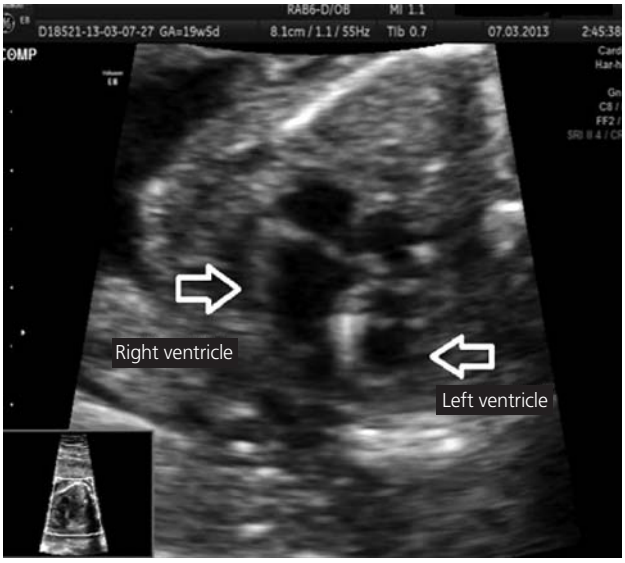

Fig. 1. 2D ultrasonographic view of left ventricle hypoplasia.

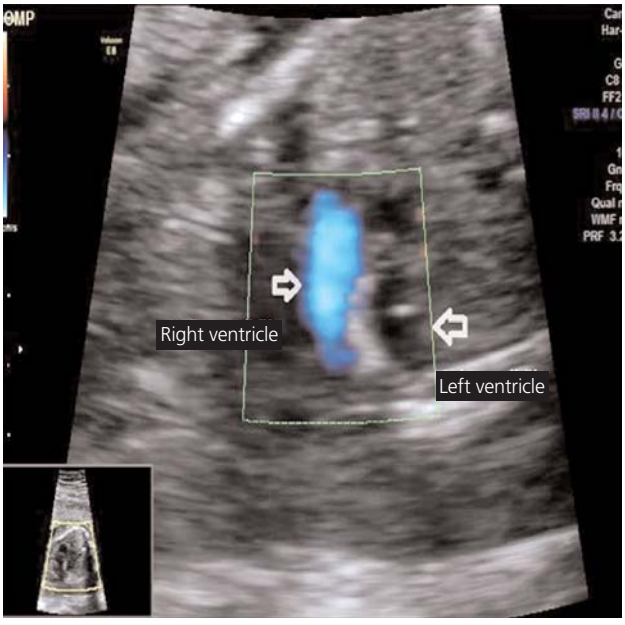

Fig. 2. While flow is seen in the right ventricle by Doppler ultrasonography, no flow is seen in the left ventricle.

Conclusion

Hypoplastic left heart syndrome can be diagnosed easily at 18-24 weeks of gestation with today's advancing technological advantages. With the opportunity for diagnosis to be presented prenatally, families will be more comfortable for making a decision about the prognosis of their fetuses compared to postnatal period. Despite the developed treatment modalities, concomitant extracardiac, additional cardiac and karyotype anomalies and operations which take long will all be effective on families to make a decision about the prognosis of fetus. This situation emphasizes the importance of early diagnosis in Turkey.

Conflicts of Interest: No conflicts declared.

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