



OLGU SUNUMU / CASE REPORT

Prenatal diagnosis of rare co-occurrence of congenital diaphragmatic hernia and aortic coarctation

Konjenital diafragmatik herni ve aortik koarktasyon nadir birlikteliğinin prenatal tanısı

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Abstract

Congenital diaphragmatic hernia is a common thoracic abnormality with increased risk of associated abnormalities. The etiology of congenital diaphragmatic hernia is not exactly known and the plausible reason is the defect of the amuscular component of the primordial diaphragm. Although, fetal heart abnormalities are the most common associated abnormalities in fetuses with congenital heart defect, aortic coarctation is occurred rarely. We present a case with the prenatal diagnosis of the co-occurrence of congenital diaphragmatic hernia and aortic coarctation.

Key words: Aortic coarctation, congenital diaphragmatic hernia, prenatal diagnosis.

Öz

Konjenital diafragmatik herni eşlik edebilecek diğer organ anomalileri riskinin arttığı, sık görülen bir toraks anomalisidir. Kesin etyolojisi bilinmemekle beraber, olası nedeni primordiyal diafragmanın amuskuler parçasındaki kusurdur. Konjenital diafragmatik hernili fetuslarda en sık birliktelik gösteren anomalisi kalp anomalisi olmakla beraber, aortik koarktasyon nadir görülmektedir. Bu olguda konjenital diafragma hernisi ve aortik koarktasyonu nadir birlikteliğinin prenatal tanısı sunulmuştur.

Anahtar kelimeler: Aort koarktasyonu, konjenital diafram hernisi, prenatal tanı.

INTRODUCTION

Fetal congenital diaphragmatic hernia (CDH) is a life-threatening anomaly with an incidence of around 1/2500 live births¹. CDH has been observed to be associated with smoking, vitamin A deficiency, alcohol and anticonvulsant exposure². The most common associated chromosomal anomaly is trisomy 21 followed by trisomy 18 and 13². CDH may be isolated or associated with malformations that includes cardiovascular (52%), urogenital (23%), gastrointestinal (14%), and central nervous (10%) systems³. Here, we report the prenatal diagnosis of a rare co-occurrence of CDH and left ventricular outflow tract obstruction. Although the combination of these 2 anomalies has been reported before, authors were not able to diagnose the aortic

coarctation prenatally^{4,5} thus, we emphasize in the present case the diagnostic difficulties and management options such as pregnancy termination.

CASE

A 27-year-old primigravida was referred for evaluation of fluid filled mass in the fetal thorax that was seen on routine second trimester ultrasonography (US) at 21 weeks' gestation. Her obstetric history was unremarkable. Detailed US of the fetus (Voluson 730; General Electric, Tiefenbach, Austria) showed fluid filled stomach and liver herniated to left chest cavity. Fetal hepatic vessels were visualised by colour Doppler examination to confirm liver herniation (Figure 1a). Moreover, fetal heart was identified in the right

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chest cavity. The pre-diagnosis was left CDH that was confirmed by fetal magnetic resonance imaging (MRI) (Figure 1b, 1c) (Siemens, Erlangen, Germany). Fetal echocardiogram was performed for accompanying heart anomalies. A right axis deviation and slightly decreased size of the left ventricle was present in the four-chamber view (4CV). Doppler US also showed decreased blood flow through the left ventricle (Figure 1d). In the three vessels and trachea view (a transverse view of upper mediastinum demonstrating the main pulmonary trunk in direct communication with the ductus arteriosus, a transverse section of the aortic arch and the superior vena cava) the diameter of the aorta was smaller (ratio, 0.6) compared to the main pulmonary artery (Figure 1e) with Z-scores (standard deviation scores used to measure growth of cardiac structures) of -2.59 and 0.53, respectively. The observed/expected lung-to-head ratio (O/E LHR) and fetal MRI were used to define the

estimated lung volumes. The O/E LHR of the right lung was less than 25% (Figure 1f), indicating increased risk of subsequent pulmonary hypoplasia. Fetal karyotyping was done after amniocentesis and demonstrated a 46, XY normal male pattern. Additional microarray analysis of the amniotic fluid did not detect any abnormalities. Subsequently, we offered counseling to consider management options of CDH with LVOT obstruction and explained the risks and benefits of pregnancy continuation in an unbiased and non-directive fashion. The couple opted for termination of the pregnancy. We performed feticide, using ultrasound-guided intracardiac KCl injection followed by vaginal misoprostol administration. Fetal autopsy revealed displaced fetal abdominal organs to thorax via a large left-sided defect and diffuse segment narrowing of the aortic lumen at the insertion of the arterial duct. There were no additional abnormalities.

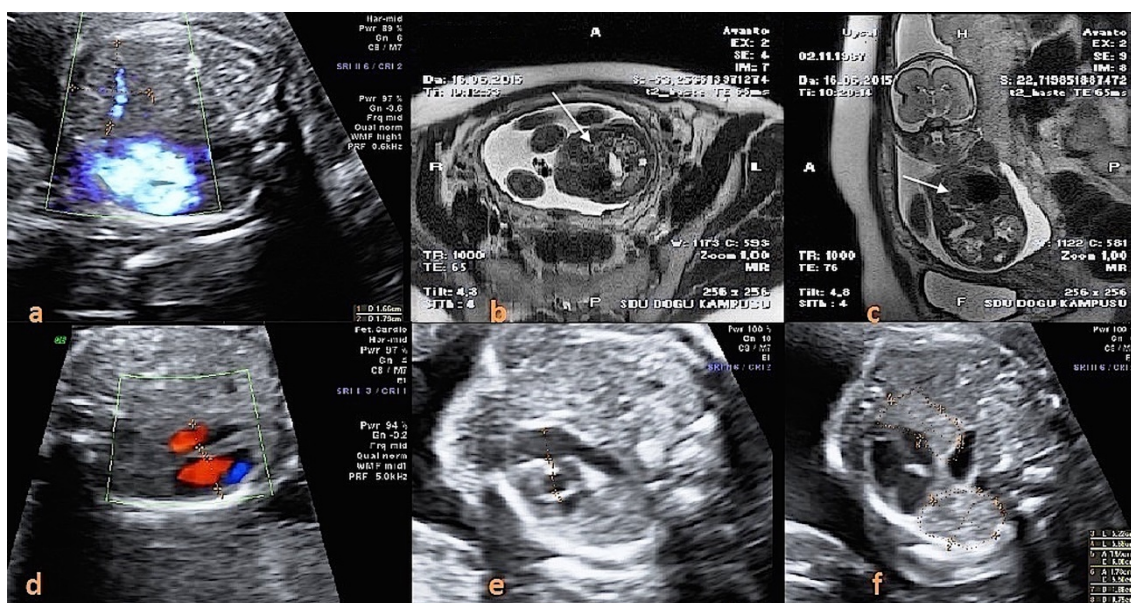


Figure-1a. Colour Doppler sonography shows hepatic vessels of herniated liver, Figure-1b. Magnetic resonance imaging (MRI) shows bowels in thorax cavity (arrow), Figure-1c. Arrow shows displaced bowels next to fetal heart in MRI. Figure-1d. Colour Doppler sonography shows decreased blood flow through the left ventricle. Figure-1e. The three vessels and trachea view shows the narrowed aorta, Figure-1f. Determination of the estimated lung volume by the tracing method.

DISCUSSION

The diaphragm forms between the 7th and 10th weeks of gestation. CDH is occurred due to the

defect of the amuscular component of the primordial diaphragm⁶. The defect causes visceral herniation to fetal thorax and may influence the normal development of the lungs, the bronchi and

pulmonary arteries to result in potential pulmonary hypoplasia.

The outcome of fetuses prenatally diagnosed with CDH is improved with proper counseling and management. CDH can be diagnosed in the first trimester by improvement of imaging techniques. However, the diagnosis of CDH is generally suggested after 20 weeks of gestation by detecting intestinal peristalsis in the fetal thorax. Most of the CDHs are left sided (85%) with the remaining being right-sided (13%) or bilateral (<2%)⁷. Although the diagnosis of right-sided CDH is more challenging, left shift of the heart due to herniated liver can be a significant finding during ultrasound screening. The CDH should be distinguished from other pulmonary abnormalities including bronchopulmonary sequestration, cystic masses, and bronchial atresia. Peristalsis of bowel in the fetal thorax and displacement of intra-abdominal organs usually help to distinguish CDH from other conditions. Therefore, appropriate transverse sections of fetal thorax during ultrasonography should be obtained and the fluid-filled stomach visualized properly in fetal abdomen to rule out a CHD.

Approximately, 10 to 15% of CDHs are associated with a congenital heart anomaly (CHA). The most common associated cardiac malformations are ventricular and atrial septal defects. Others include Fallot tetralogy, hypoplastic left heart syndrome, dextrocardia, and aortic arch abnormalities⁸. CDH accompanied by major CHAs are significantly associated with decreased survival rate at 1 year compared to isolated CDH (40.0% versus 77.1%)⁹. Furthermore, severity of CHA is the main predictive factor for perinatal mortality and morbidity in cases of CDH¹⁰. Therefore, detailed cardiac evaluation is an important component of appropriate prenatal work-up of antenatally diagnosed CDH.

A plausible aetiology of outflow tract and aortic arch abnormalities associated with CDH may be a disorder of neural crest cell migration to the aortic arches¹¹. Left-sided CDH and liver herniation are also associated with functional left heart hypoplasia, possibly due to altered ductus venous flow and external compression by herniated abdominal organs¹². Due to similar features in fetal echocardiogram isolated CDH may be overdiagnosed as CDH accompanied aortic coarctation. In the case described here, the severity of left ventricle outflow tract obstruction at fetal

echocardiogram increased within a week. This may suggest an anatomic defect of the fetal left heart accompanied by progressive functional deterioration. Therefore, the progressive nature of fetal heart defects during antenatal follow-up should be emphasized.

In summary, CDH may be accompanied by congenital heart defects including aortic coarctation, which is a relatively rare association. Therefore, the disproportion in favour of right ventricle in 4CV and larger pulmonary artery according to Z-scores should alert physician about the co-occurrence. Major CHA, liver herniation, and decreased lung volume in fetuses with CDH are poor prognostic factors. Thus, genetic tests, fetal echocardiogram, and determination of the estimated fetal lung volume should be the mainstays of prenatal work-up. These will aid for proper counseling and perinatal management.

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