



Progressive Fetal Diaphragmatic Hernia: A Case Report

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Abstract

Objective: We herein aimed to present a case of progressive diaphragmatic hernia in which fetal gastric herniation occurred in the end of the second trimester and this case revealed no major sign except cardiac dextroposition in previous pregnancy weeks.

Case: Cardiac dextroposition was detected in fetal ultrasonography which was carried out in a woman with 16 week pregnancy for her first child. Irregular hyperechoic sites were observed in lower left part of fetal thorax. There was no finding to consider the intestinal peristaltism in this area. Fetal heart and the other fetal anatomy were evaluated and admitted as normal. Primarily, diaphragmatic hernia was considered. Information about the possible diagnosis and prognosis was given to the family. The same ultrasonographic findings were also detected in the 19th pregnancy week. The patient did not accept the amniocentesis for the karyotype identification. When she was controlled at the 28th pregnancy week, it was observed that there was a gastric herniation supporting the diagnosis of diaphragmatic hernia. Hernia reparation was made for the baby who was operated postpartum 3rd day following the birth by cesarean in 39th pregnancy week. The newborn was dependent on the ventilator for about 16 days and was discharged with recovery in postoperative 30th day.

Conclusion: In fetal diaphragmatic hernia, a hypoechoic intestinal ans with peristaltism or the hypoechoic appearance of the stomach which is observed at the lateral side of the heart may not be always seen ultrasonographically. These findings may emerge in later pregnancy weeks. Cardiac dextroposition may be the only cautionary major finding in early pregnancy weeks.

Keywords: Fetal diaphragmatic hernia, cardiac dextroposition, prenatal diagnosis.

Progresif fetal diafragmatik herni: Olgu sunumu

Amaç: Fetal mide herniasyonu ikinci trimesterin sonunda gerçekleşen ve daha önceki gebelik haftalarında kardiyak dekstropozisyon dışında major bir belirti vermeyen progresif bir diafragmatik herni olgusunu sunmayı amaçladık.

Olgu: Birinci çocuğuna 16 haftalık gebede yapılan fetal ultrasonografide kardiyak dekstropozisyon saptandı. Fetal toraks sol alt lokalizasyonunda düzensiz hiperekoik alanlar izlenmekteydi. Bu alanda barsak peristaltizmini düşündürecek bulgu mevcut değildi. Fetal kalp ve diğer fetal anatomi normal olarak değerlendirildi. Öncelikli olarak diafragmatik herni düşünüldü. Aileye olası tanı ve prognoz hakkında bilgilendirme yapıldı. 19. gebelik haftasında da aynı ultrasonografik bulgular saptandı. Karyotip tayini amaçlı amniosentezi kabul etmeyen hasta daha sonra 28. gebelik haftasında başvurduğunda diafragma hernisi tanısını destekleyen mide herniasyonunun gerçekleştiği izlendi. 39. gebelik haftasında sezaryen ile doğumu takiben postpartum 3. gün opere edilen bebeğe herni reparasyonu uygulandı. 16 gün ventilatörde kalan yenidoğan postoperatif 30. gün şifa ile taburcu edildi.

Sonuç: Fetal diafragmatik hernide ultrasonografik olarak her zaman kalbin lateralinde mideye ait hipoekoik görünüm veya peristaltizmin izlendiği hipoekoik barsak ansları görülmeyebilir ve bu bulgular daha geç gebelik haftalarında ortaya çıkabilir. Kardiyak dekstropozisyon erken gebelik haftalarında uyarıcı tek major bulgu olabilir.

Anahtar Sözcükler: Fetal diafragmatik herni, kardiyak dekstropozisyon, prenatal tanı.

Introduction

Congenital diaphragmatic hernia (CDH) is a common major malformation. Its incidence is 1/3,000–1/4,000 births.^[1,2] CHD's are classified according to the place of the defect in diaphragma; 80–85% of the defects are at the posterolateral side (Bochdalek type), 10–15% are at the right anterior (Morgagni), 3–4% are bilateral.^[2,3] Although the mortality of CDH is decreasing recently, it is reported to be 30–50% and this rate may be higher in prenatally diagnosed cases.^[4] In a current study evaluating 51 mutual center data in the world, the general mortality rate before discharge was found as 31% by "Congenital Diaphragmatic Hernia Study Group".^[5] In this case report, We discussed the case of diaphragmatic hernia showing intrauterin progressive course because cardiac dextroposition and non diagnostic hyperechoic focuses in the sub-section of the left thorax were only observed until the end of the second trimester in ultrasonography, and specific diaphragmatic hernia Picture emerged at the onset of the third trimester.

Case Report

G:1, P:0 patient at the age of 30 got pregnant as a result of 3rd IVF experimentation due to the azospermia. In her history, she used metformin 850 mg 2x1 till the 9th pregnancy week and 5 mg folic acid daily in the first trimester because of the polycystic over and insuline resistance diagnosis. Her male spouse was a cystic fibrosis carrier. The

cystic fibrosis scanning which contains 36 mutations was found as normal when this was performed to the patient before the pregnancy. Even though information was given, the patient did not accept the detailed cystic fibrosis mutation scanning. The complete blood count, urine analysis and the routine biochemical examinations in the first trimester were found within the normal limits. She was recorded as HbS Ag(-), Anti HbS Ag(-), Anti HCV(-), HIV(-). The nuchal translucency was 1.2 mm in 12th pregnancy week. The first trimester Down Syndrome scanning test was within the normal limits. Fetal cardiac dextroposition was detected in the ultrasonographic examination which was performed in the 16th pregnancy week. Irregular hyperechoic sites on the lower left part of fetal thorax were observed (Figure 1a). There was no finding to consider the intestinal peristalsism in this area. In fetal echocardiography, the cardiac structures were evaluated as normal. The fetal anatomy other than the heart was also normal. Primarily, diaphragmatic hernia was considered. In the differential diagnosis, other masses invading the lung such as congenital cystic adenomatoid malformation and pulmonary sequestration were considered. Information about the possible diagnosis and prognosis was given to the family. Genetic amniocentesis was recommended. But the family stated that they did not think about the pregnancy termination and they did not accept the amniocentesis. The same ultrasonographic findings were detected in the 19th pregnancy week (Figure 1b).



Figure (a-b). (a) Cardiac dextroposition, irregular hyperechoic sites related to herniated intestinals in the place where the heart should exist in the lower part of the left thorax in the 16th pregnancy week (arrowheads). (b) Cardiac dextroposition and irregular hyperechoic sites related to the herniated intestinals which are more specific in the lower part of the left thorax in the 19th pregnancy week (arrows)

Information about chromosomal abnormalities and prognosis was given to the family once more and amniocentesis was recommended. But the family did not accept again. The patient did not come for the follow up in the date which was offered in the 19th pregnancy week. But the patient came in the 28th pregnancy week. It was observed that the gastric herniation supporting the diagnosis of diaphragmatic hernia occurred (Figure 2). There was no change in ultrasonographic findings in the later pregnancy weeks. She delivered a male baby, 2,650 g in weight in another center by cesarean section in the 39th pregnancy week. The baby was operated postpartum 3rd day following the birth and the hernia reparation was performed. The newborn was dependent on the ventilator for about 16 days and was discharged with recovery on the postoperative 30th day.



Figure 2. It was seen that the gastric herniation occurred in the 28th pregnancy week.

Discussion

Fetal diaphragmatic hernia is on the left side of 80%-90% cases. The most important finding in the diagnosis is the existence of cystic mass on the left side of thorax and the stomach is unable to be seen. The observation of peristalsis in the cystic mass is pathognomonic.^[2] The other findings are the deviation of the heart to the right and polyhydramnios. The diagnosis of small CHDs which do not have the gastric herniation is difficult. The abnormal heart axis may be the only finding.^[2,3,6] In our case, there was no gastric herniation in the first 2 trimesters and the only important finding was

cardiac dextroposition. There were irregular hyperechoic sites in the lower part of the left thorax in ultrasonography. We considered that this finding might be the herniated small intestines. Therefore, in cases which the cardiac malposition was only seen especially in early pregnancy, the careful examination of the lower part of thorax may help the diagnosis for the possible small intestinal hernia. Although there is a liver herniation at the rates of 85% on the left side diaphragmatic hernia, the diagnosis of the liver herniation is difficult because the echogenities of the liver and the lung are similar to each other.^[2] The pushing of the stomach posteriorly supports the diagnosis. But the examination of portal venous system by Doppler is important in the diagnosis.^[2,6] In our case, a finding related to the liver herniation was not detected during all pregnancy period. The most important cause of the perinatal mortality in CHD is pulmonary hypoplasia. The diagnosis of CHD may be performed from the first trimester onwards. The airway and vascular development of the lung are deteriorated after the herniation occurs. It causes the development of a progressive complex pulmonary pathology.^[7] Decreased thorax capacity avoiding the development of the lungs causes the numerical decrease in airways, alveolus and arteries. And thickness occurs in the media layers of the artery walls. It causes the existence of the muscular layer in small pre-asiner arteries in the peripheral side. These changes account for persistant fetal circulation and pulmonary hypertension which are observed after the postpartum hernia reparation.^[7] In our case, the perinatal result is fairly good. The newborn was discharged with recovery following the postpartum hernia reparation. Gastric herniation occurred in the end of the second trimester and at the onset of the third trimester, that is, in the latest pregnancy period. This caused minimal effect on the lungs and led to good perinatal results.

Conclusion

Consequently, the existence of the cystic mass on the left side of the thorax and the normal localization of the stomach can not rule out the diaphragmatic hernia in cardiac dextroposition cases. The existence of irregular hyperechoic sites which may be the sign of the small intestinal her-

niation in the lower part of the left thorax supports the diagnosis of diaphragmatic hernia in these cases. In cases which produce the diagnostic difficulties and progress with only cardiac dextroposition, the serial ultrasonographic follow ups are important during the pregnancy in order to detect the organ herniations such as stomach and liver.

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