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Case Report

A rare case that can be diagnosed with prenatal ultrasound: Fryns syndrome

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Abstract. Fryns syndrome is a syndrome that is accompanied by multiple congenital anomalies, with extremely high mortality, showing autosomal recessive inheritance and often leaves severe mental retardation on those who survive. Abnormal facial appearance, distant faded nipples, small rib cage, distal extremity and nail hypoplasia, pulmonary hypoplasia and diaphragmatic hernia are among major criteria for the diagnosis. Later, cardiovascular, genitourinary, central nervous system and skeletal system anomalies have been identified. Here, we presented Fryns syndrome case which has been diagnosed by prenatal ultrasound, and of which the diagnosis has been confirmed by physical examination and ultrasound at the postnatal period.

Key words: Diagnosis, prenatal, ultrasound, Fryns syndrome

1. Introduction

Fryns syndrome is a rare pathology which is lost shortly after birth due to pulmonary hypoplasia, in which the rate of survival is very small and showing autosomal recessive inheritance accompanied by mental retardation at varying levels (1, 2). It was described in two sisters' cases by Fryns et al in 1979 for the first time. Although, at the beginning, the major criteria for the diagnosis were reported as abnormal facial features, distant faded nipples, small rib cage, diaphragmatic hernia, distal extremity and nail hypoplasia, later, it was stated that it may be accompanied by cardiovascular, gastrointestinal, genitourinary, central nervous system and skeletal system abnormalities. In the general population the incidence is noted as one in 10,000 live births (2). Today, Fryns syndrome can be diagnosed by after-birth physical examination findings and genetic features. In this article, we aimed to present the Fryns syndrome case that is rare and is diagnosed by prenatal ultrasound and prenatal and postnatal sonographic features of this case.

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2. Case report

A 38-year-old female patient with no kinship between spouses and with gravida 8, parity 7 applied to our clinic for the routine antenatal sonography.

Biometric measurements of the patient who did not know the date of the last menstrual were consistent with 35 weeks. At the obstetrical ultrasonography, the amniotic fluid index was measured to have increased. Also, 40 x 30 mm in size cystic dilatation and cerebellar vermis defective appearance at the fetal brain case posterior fossa localization which is interpreted as Dandy-Walker anomaly (Figure 1), left heart hypoplasia that is seconder to the defective appearance of the diaphragm and the translocation of the stomach from this defect towards the intrathoracic heart level, displacement of the heart to the right and the hernia compression were observed (Figure 2). The right kidney of the fetus was viewed as lightly malrotated and pelvicalyceal system as dilated. At the sonographic examination, four parallel lines sign that was created by labia major and minor belonging to the female gender were revealed. 50 x 26 mm sized cystic lesion with lobulated contour and containing internal echoes and septa was observed at the lower abdominal section of the fetus.

The baby was born 2800 gr with spontaneous vaginal delivery at the 40th week. The baby with cyanosis and bradycardia, who did not cry after

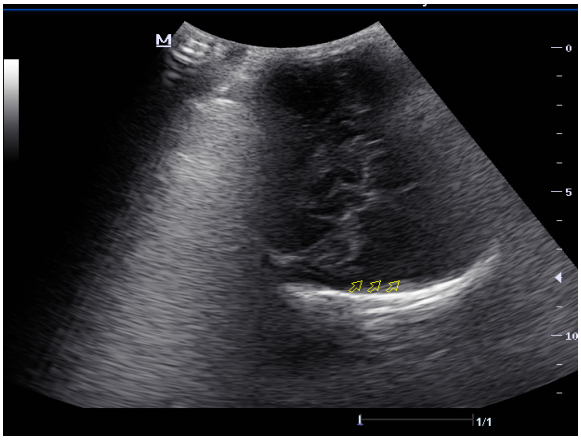

Fig. 1. Prenatal sonography cystic lesion and cerebellar vermis defect in fetal brain posterior fossa.

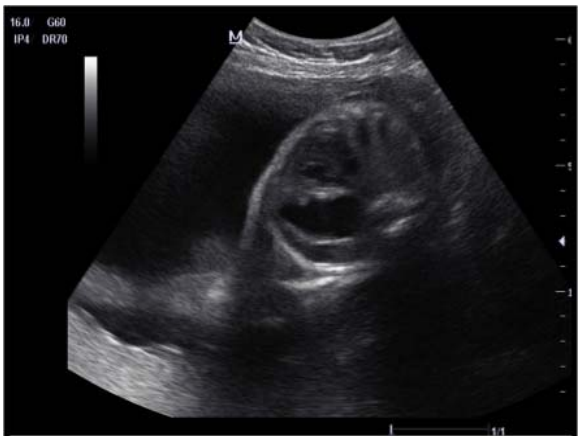

Fig. 2. Diaphragmatic hernia is monitored on the left side of the heart on fetal gastric pouch thoracic level at prenatal sonography.

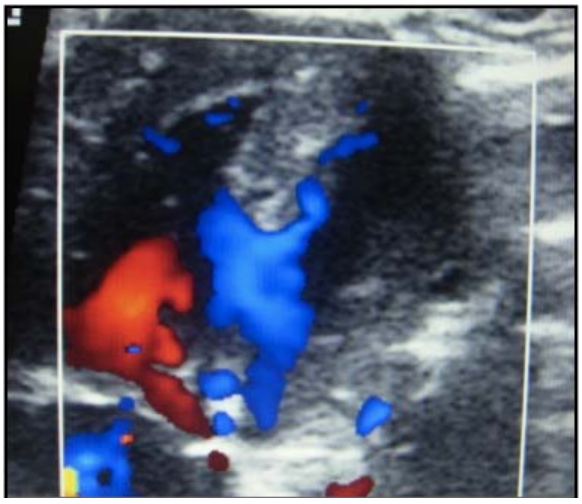

Fig. 3. Ventricular septal defect.

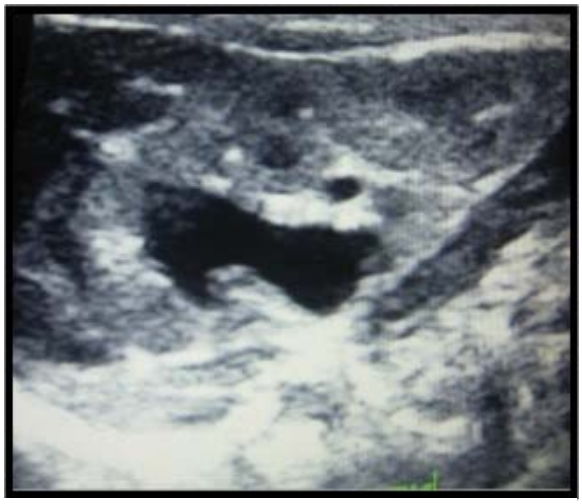

Fig. 4. Pelvicaliectasy.

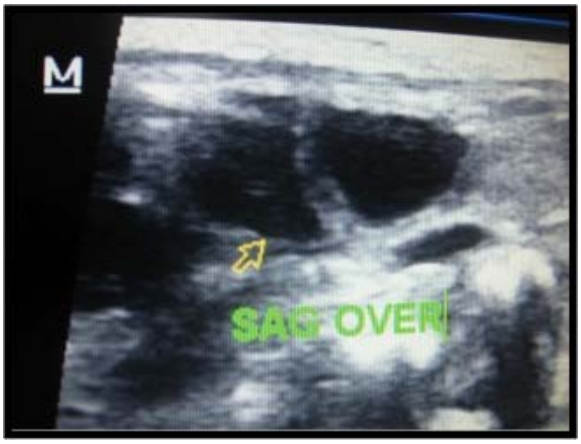

Fig. 5. Ovarian cysts.

the birth was intubated and was transferred to the neonatal intensive care unit. The baby who was monitored mechanical ventilation mounted at the intensive care unit was treated with surfactant. On chest radiography; it was viewed that the stomach gas was in the neighborhood of the heart at the left hemithorax and the heart pushed to the right, and the left lung volume decreased.

In the ultrasonography performed in the neonatal intensive care unit in the postnatal period, left heart hypoplastic and ventrikuloseptal defect with the intrathoracic location at the stomach-heart neighborhood, and the heart displaced to the right, were detected (Figure 3). A cystic mass lesion with internal echo, in the appearance of dilated pelvicalyceal system at the malrotated right kidney (Figure 4) and showing dimensional reduction at the right over region according to the prenatal sonography were detected (Figure 5).

In the transfontanel sonography performed in the postnatal period, cerebellar vermis defective

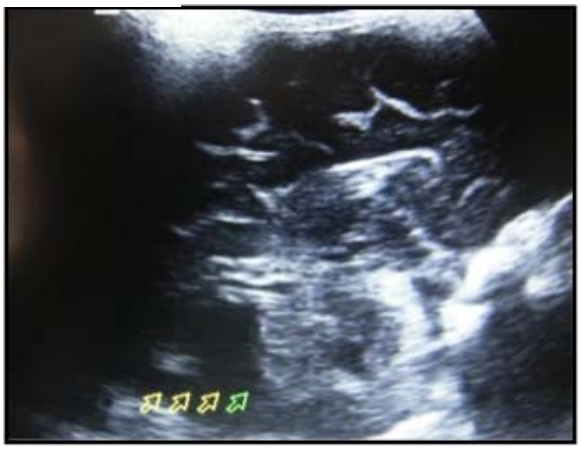

Fig. 6. Postnatal sonography cystic lesion and cerebellar vermis defect in fetal brain posterior fossa.

Fig. 7. Low hypoplastic ear.

appearance consistent with the sonographic findings in the prenatal period and cystic lesion associated with the fourth ventricle in the posterior fossa were detected (Figure 6).

At the physical examination, there were right ear hypoplastic and helix deformity, left microphthalmia and high palate (Figure 7). Skeletal abnormalities were not detected. With these findings, the case was diagnosed as Fryns syndrome. Diaphragm hernia operation was planned for the patient. However, the patient was lost 24 hours after the birth due to the rapid decline in oxygen saturation and bradycardia development.

3. Discussion

Fryns syndrome (X-pass mental retardation with marfanoid habitus syndrome) is a usually fatal disease showing autosomal recessive inheritage together with severe mental retardation in the patients surviving (3). In the general population, the incidence is noted as one in 10,000 live births (1, 2). Fryns syndrome is similar to the Marfan syndrome with its physical properties such as tall stature, kyphoscoliosis, long cylindrical limbs, arachnodactyly, joint hyperextensibility, long face, small chin, and hypotonia. Although there were physical properties such as arachnodactyly, long face, small chin in our case, skeletal abnormalities were not observed (2-5). Findings such as FBN1 mutations, autosomal dominant inheritance and lens subluxation that can be detected in Marfan syndrome do not exist in Fryns syndrome. Observing autosomal recessive inheritance, severe mental retardation and behavioral problems in Fryns syndrome are distinctive features from Marfan syndrome.

Majority of the patients with Fryns syndrome are lost as a result of pulmonary pathology (2-5). However, 15% of patients live due to the phenotypic differences (4). Our patient has been lost due to hypoxia and bradycardia in a similar way. In these cases, there are usually family histories. Therefore, genetic counseling is required for the families of these patients and the next pregnancies should be monitored for Fryns syndrome. The most common anomaly which is among the major diagnostic criteria in Fryns syndrome cases described first is diaphragmatic hernia (1-4). In our case, diaphragmatic hernia was also found. Pulmonary hypoplasia cases without diaphragmatic hernia have been reported in the literature. In these, there usually exists central nervous system abnormalities, it is asserted that the lack in the diaphragm innervation associated to this anomaly and pulmonary hypoplasia associated to this may involve (5, 6). In our case, diaphragmatic hernia as well as central nervous system anomaly was present, and we believe that both of them are in charge of formation of pulmonary symptoms.

In recent publications, central nervous system such as corpus callosum aganez, the Dandy-Walker anomaly, heterotopia and cardiac anomalities such as isolated ventriculoseptal defect, atrial septal defect, bicuspid aortic valve are mentioned among Fryns syndrome diagnostic criteria (7, 8).

As a result, Fryns syndrome is a pathology which is a one of the major causes of congenital diaphragmatic hernia and has a quite high mortality due to pulmonary hypoplasia. Survival can be increased by repair operations to the diaphragm, which will be detected and

implemented on time in the early period by fetal ultrasound scans.

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