Congenital unilateral pulmonary hypoplasia presenting as cardiac dextroposition on prenatal ultrasonography


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Summary
Objective: To report a case of congenital right pulmonary hypoplasia presenting as cardiac dextroposition on prenatal ultrasonography. Case Report: A 29-year-old woman with fetal dextroposition visited this institution at 24 weeks of gestation. Prenatal fetal echocardiography and detailed ultrasonography could not detect fetal anomaly except for fetal dextroposition. However, congenital pulmonary hypoplasia was diagnosed postnatally. Conclusion: Primary unilateral pulmonary hypoplasia should be suspected in cases with abnormal heart position of an unknown origin and fetal magnetic resonance imaging (MRI) could be an additional option for prenatal diagnosis of primary pulmonary hypoplasia.

Key words: Cardiac dextroposition; Congenital unilateral pulmonary hypoplasia; Perinatal ultrasonography.

Introduction
Unilateral primary pulmonary hypoplasia or agenesis is extremely rare but may be a cause of neonatal respiratory distress. Most cases of pulmonary hypoplasia are usually associated with reduced intrathoracic space, including diaphragmatic hernia, pleural effusion, or thoracic skeletal dysplasia. The present authors encountered a case of primary unilateral pulmonary hypoplasia that presented as dextroposition on prenatal ultrasonography. Here, they report their case with a review of literature.

Case Report
A 29-year-old woman (gravida 1, para 0) with a singleton gestation visited this institution because of fetal heart anomaly at 24 weeks’ gestation. A previous ultrasonography scan at 22 weeks’ gestation showed dextrocardia with situs solitus in other abdominal organs. She did not have any underlying and hereditary diseases. Her previous laboratory study had normal results, so amniocentesis was not performed.

Fetal echocardiography performed in this institution at 24 weeks’ gestation revealed a parallel cardiac axis to the interventricular septum and dextroposition (Figure 1). However, no abnormal findings were obtained from the cardiac chambers, and atrioventricular and ventriculoarterial connection. In addition, the authors identified drainage of the pulmonary veins into the left atrium and bifurcation of the pulmonary artery.

At the follow-up examination at 28+4 weeks’ gestational age, the authors could not find any intracardiac abnormalities. However, ultrasonography revealed an increase in the amniotic fluid index (AFI; 22–24). Therefore, they carefully re-examined the fetal face and stomach to detect cleft lip or palate and gastrointestinal obstruction. In addition, microtia in the right ear was suspected on three-dimensional ultrasonography, but it was not a definitive finding. Her AFI increased up to 25–28 at 37 weeks of gestation, and she complained of decreased fetal movement. Ultrasonographic examination revealed greater increase in AFI (36) at 38+2 weeks’ gestation. Therefore, the authors decided to deliver the fetus by emergency cesarean section.

The female fetus was delivered and weighed 2,950 grams, with one- and five-minute Apgar scores of 5 and 6, respectively. The umbilical arterial gas analysis after cord clamping revealed the following values: pH 7.355, PCO2 45.6 mm Hg, PO2 33.1 mm Hg, and base excess –0.9.

Immediately after delivery, the neonate had no initial crying and showed whole-body cyanosis. Therefore, she was admitted to the neonatal intensive care unit for further evaluation and management.

Figure 1. — Heart in dextroposition at 24 weeks 5 days of gestational age.
A first chest radiograph was obtained shortly after birth (Figure 2) and showed a homogenous right chest opacity without air bronchogram. In addition, the cervical hemivertebrae spine was shown in the anteroposterior view (Figure 2). Additional chest computed tomography (CT) showed complete absence of the lung, bronchus, and pulmonary vascular structures in the right thoracic cavity (Figure 3). Vertebral segmentation and formation failure anomalies were found coexisting in the lower cervical and upper thoracic spine, causing scoliosis. Postnatal echocardiography revealed agenesis of the right pulmonary artery and right pulmonary vein without accompanying intracardiac abnormality. Moreover, temporal CT and three-dimensional head CT revealed facial anomalies, including complete absence of the mandibular ramus, atresia of the extra-auditory canal, hypoplasia of the right middle ear, and displaced lobular remnant with absent auricle on the right face. The neonate was suspected of having several genetic syndromes (e.g., Goldenhar syndrome); therefore, the authors recommended a gene analysis. However, the parents refused further evaluation.

Discussion

Dextroposition of the heart is a condition in which the heart is in the right chest and the cardiac apex points medially to the left, as in this case, and its incidence is known to be one in 12,019 neonates [1]. Fetal cardiac malposition is of two main types; one is caused by intrinsic congenital heart diseases, and the other is caused by extracardiac malformation. After excluding intrinsic heart diseases, dextroposition is usually associated with space-occupying lesions, including lung mass (especially congenital pulmonary airway malformation), congenital diaphragmatic hernia, or pleural effusion. However, right lung agenesis or hypoplasia may be the cause of dextroposition, and congenital unilateral pulmonary hypoplasia may present as an abnormal heart axis as in the present case.

Primary unilateral pulmonary hypoplasia is extremely rare and may be caused by embryological defects of the lung or vascular accident, idiopathic deficiencies in certain transcription or growth factors, or other syndromes, fetal akinesia-hypokinesia sequence, Scimitar syndrome, and trisomy 21 [2]. Despite the improvement of the prenatal ultrasonography finding, prenatal detection of pulmonary hypoplasia may only be possible in secondary hypoplasia on prenatal ultrasonography [3]. Previous studies showed that reduced thoracic circumference or lower thoracic-to-abdominal circumference ratio may be associated with congenital pulmonary hypoplasia [4-6]. In addition, fetal magnetic resonance imaging (MRI) can be used to evaluate lung volume during a pregnancy in suspected cases [7]. The present authors could not suspect the possibility of unilateral right pulmonary hypoplasia in the prenatal period; therefore, they did not perform fetal MRI. As the clinical manifestation of neonates or infants with congenital pulmonary hypoplasia may be diverse depending on the severity of diseases, which range from completely asymptomatic to severe respiratory failure, they suggest that fetal MRI could be considered in cases with dextroposition of an unknown origin during prenatal ultrasonography. Meanwhile, pulmonary hypoplasia could be accompanied with other congenital anomalies, including cardiac malformation, skeletal or vertebral anomalies, and trachea-esophageal fistula [7]. In this case, cervical and upper thoracic hemivertebrae, right micrognathia, and right microtia coexisted. The authors carefully performed prenatal ultrasonography to determine the cause of the dextroposition and polyhydramnios, but they did not find the cause during the prenatal period.

The prognosis of primary pulmonary hypoplasia depends on the degree of lung underdevelopment. Previous case reports about primary pulmonary hypoplasia showed high mortality rates (75–100%), but unilateral primary hypoplasia showed favorable outcomes [7-10]. However, respiratory function must be closely followed up because
obstructive pulmonary diseases associated with tracheobronchomalacia, reactive airway disease, and bronchial compression by surrounding vessels often develop [11]. In some cases, surgical resection of the hypoplastic lung is recommended to prevent recurrent infection [7].

In conclusion, the authors had a case of unilateral primary pulmonary hypoplasia that presented as dextroposition on prenatal ultrasonography. While secondary pulmonary hypoplasia is suspected easily, primary pulmonary hypoplasia is not usually suspected in the prenatal period. On the basis of this case, they suggest that primary unilateral pulmonary hypoplasia should be suspected in cases with abnormal heart position of an unknown origin and fetal MRI could be an additional option for prenatal diagnosis of primary pulmonary hypoplasia.

References


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