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Research Letter

Congenital high airway obstruction syndrome and abnormal pulmonary situs: An extremely rare prenatal association

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Dear Editor,

A 27-year-old gravida 2 para 1 woman, at 23 weeks of gestation, was referred to Akdeniz University Perinatology Department for evaluation of fetal ascites.

Grayscale ultrasonography revealed enlarged and hyperechoic lungs, and dilated and prominent trachea that extended to the subglottic level, compressed heart and low cardiothoracic circumference ratio, inverted diaphragmatic convexity, ascites, and skin edema with polyhydramnios (Figures 1 and 2). According to these findings, congenital high airway obstruction syndrome (CHAOS) due to laryngeal atresia was considered as the diagnosis. The couple was informed about *ex utero* intrapartum treatment (EXIT) and the relatively poor prognosis of the syndrome. Finally, pregnancy termination was chosen.

Fetal autopsy revealed laryngeal atresia, intact esophagus without fistula, ventricular septal defect and hypoplastic right kidney. The lungs were bilaterally enlarged. The right and left lungs were bilobulated and trilobulated, respectively (Figures 3 and 4). The length of the right bronchus from the carina to its first branch was longer than the left side. The situs of other organs was normal. Finally, amniocentesis revealed a normal male karyotype (46,XY).

With today's high resolution ultrasound probes, it is straightforward to detect the major features of CHAOS. However, the potential higher failure rate in EXIT procedure of a fetus with a lower level obstruction ascribes further importance to recognizing CHAOS and revealing the precise level of the obstructed area. In our case, we were able to detect the level by ultrasonography alone. This gave us the opportunity to inform the couple about the EXIT procedure and emergency intrapartum tracheostomy. However, the couple chose termination of pregnancy.

During the embryonic period (3–7 weeks gestation), tracheal–bronchial tubules are formed from the pulmonary diverticulum that forms at the medial tracheal–laryngeal sulcus in the ventral wall of the foregut [1]. There are no prominent candidate genes or mouse models for laryngeal atresia to date [2]. However, in mice, normal pulmonary lobulation and situs are determined by genes (Gli-2, Lfty-1, Gdf-1) regulating lung development and left-right asymmetry [2,3]. In humans, coexistence of laryngeal atresia and abnormal pulmonary lobulation was reported in three cases. All of these cases had a bilobular right lung, whereas two cases had bilobular and one had unilobular left lungs [4–6]. In addition, the authors did not report any information with

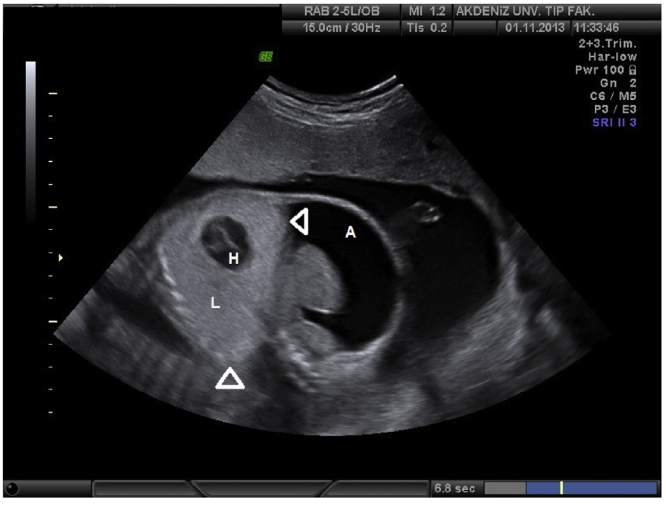

Figure 1. Sonographic view of enlarged, hyperechogenic lungs, small heart, inverted diaphragm and ascites. A = ascites; H = heart; L = lungs; arrowheads = diaphragm.

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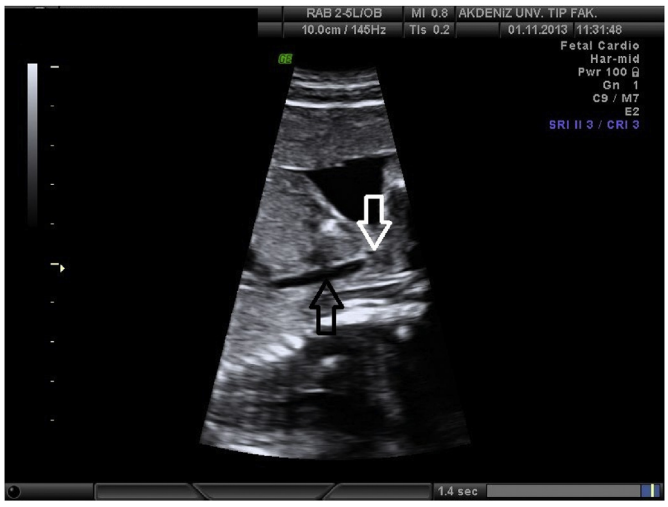

Figure 2. Sonographic view of dilated trachea (black arrow) and a sharp border (white arrow) between it and the occluded larynx.

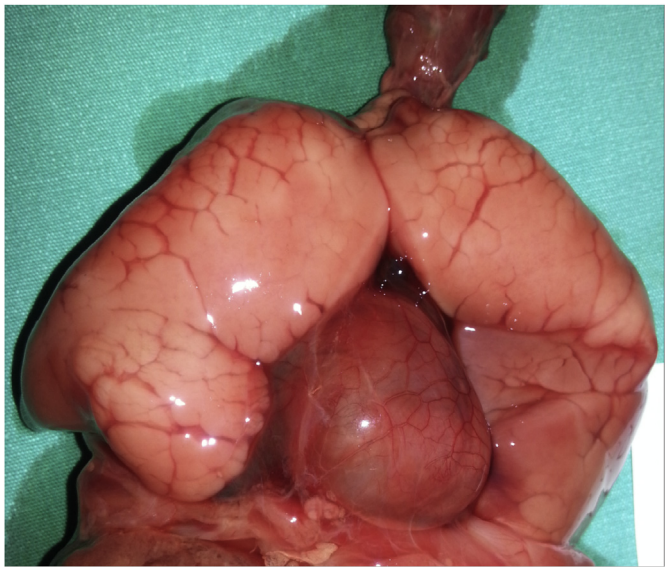

Figure 3. Note the bilobulated right lung and trilobulated left lung (anterior view).

respect to the lengths of carina to bronchus on the right and left sides, which is one of the characteristics of abnormal pulmonary situs.

The potential importance of pulmonary abnormal situs has been reported. Some of the similar pulmonary findings of our case (2 lobes in the right lung and 3 lobes in the left lung without other evidence of heterotaxy) were disclosed in a postmortem examination of a patient with achondroplasia–hypochondroplasia complex [7]. Abnormal primary pulmonary phenotype associated with

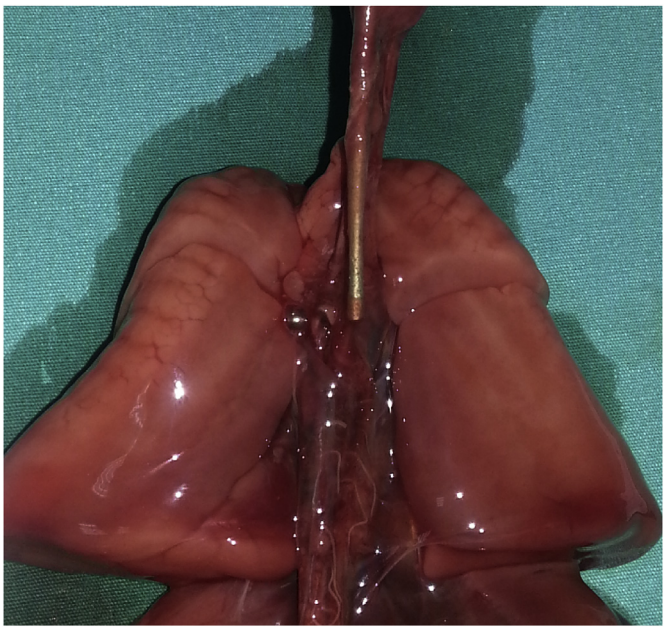

Figure 4. Note the bilobulated right lung and trilobulated left lung (posterior view, stilet in esophagus).

FGFR3-opathies was suggested as the cause for the poorer than expected respiratory status.

To the best of our knowledge, apart from abnormal pulmonary lobulation, this is the first case reporting coexistence of abnormal pulmonary situs and laryngeal atresia. Further studies should be conducted to reveal the genetic basis and clinical importance of abnormal pulmonary situs and its association with CHAOS and any other abnormality.

Conflicts of interest

The authors have no conflicts of interest relevant to this article.

References

[1] Whitsett JA, Wert SE, Trapnell BC. Genetic disorders influencing lung formation and function at birth. Hum Mol Genet 2004;13:R207–15.
[2] Sanford E, Saadai P, Lee H, Slavotinek A. Congenital high airway obstruction sequence (CHAOS): A new case and a review of phenotypic features. Am J Med Genet A 2012;158A:3126–36.
[3] Raya A, Belmonte JC. Left-right asymmetry in the vertebrate embryo: from early information to higher-level integration. Nat Rev Genet 2006;7:283–93.
[4] Rankin NE, Mendelson IR. A case of atresia of the larynx. Arch Dis Child 1956;31:324–7.
[5] Choong KK, Trudinger B, Chow C, Osborn RA. Fetal laryngeal obstruction: sonographic detection. Ultrasound Obstet Gynecol 1992;2:357–9.
[6] Minior VK, Gagner JP, Landi K, Stephenson C, Greco MA, Monteagudo A. Congenital laryngeal atresia associated with partial diaphragmatic obstruction. J Ultrasound Med 2004;23:291–6.
[7] Bober MB, Taylor M, Heinle R, Mackenzie W. Achondroplasia–hypochondroplasia complex and abnormal pulmonary anatomy. Am J Med Genet A 2012;158A:2336–41.