

Isolated laryngeal atresia, prenatal diagnosis and pathology. Case report

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Abstract

Congenital high airway obstruction syndrome (CHAOS) is a rare disorder defined as any fetal abnormality that obstructs the larynx or trachea. Most cases are sporadic with an unknown incidence. We report a case that was diagnosed prenatally at 21 gestational weeks, which showed a typical CHAOS pattern with expanded hyperechogenic lungs, inverted diaphragms, dilated trachea and ascites. There were no other fetal abnormalities and the karyotype was normal (46xy). The parents opted to have an elective termination of pregnancy. Pathological examination confirmed the diagnosis, showing infraglottic atresia (type II) and complete trachea obstruction. The disease is incompatible with life, and antenatal ultrasound diagnosis is advisable. However, few cases have been reported which managed with neonatal interventions such as ex-utero intrapartum treatment.

Keywords: laryngeal atresia, prenatal diagnosis, fetus, ultrasonography

Introduction

Congenital high airway obstruction syndrome (CHAOS) is a rare condition in which the fetus has a typical triad of large highly echogenic lungs, flattened or inverted diaphragms and ascites.

The obstruction can be due to laryngeal/tracheal atresia or external compression. Prenatal diagnosis is essential because the disease is generally incompatible with life, although there are reports of some cases saved through neonatal interventions such as ex-utero intrapartum treatment (EXIT)⁽¹⁾.

We report a case of 21 week-old male fetus with isolated congenital laryngeal atresia diagnosed prenatally by ultrasound findings. The parents opted for an elective termination of pregnancy. The diagnosis was confirmed by pathology.

Case report

A 34-years-old nullipara (gravida 2) woman at 21 weeks' gestation was referred for further evaluation of abnormal sonographic findings. There was no consanguinity, and the family and medical history were noncontributory.

Her first pregnancy was finished through a missed abortion at 12 weeks of gestation.

Physical examination revealed a 22-week-sized uterus with present fetal heart movements. Sonographic findings at 21 weeks' gestation were moderated ascites, enlarged hyperechoic lungs with a dilated trachea and bronchial tree.

Color Doppler differentiated them from the adjacent pulmonary artery branches. The diaphragm was displaced downwardly (Figure 1). We found a dilated trachea of about six mm, blind-ending at the level of the fetal neck (Figure 2). The heart, apparently

structurally normal, was compressed by the echogenic lungs, with a cardio-thoracic ratio of 0.35 (Figure 3). Neither pleural nor pericardial effusion was found. The abdominal circumference was 239 mm, larger than the 95th percentile for gestational age.

No other anomalies were noticed at the ultrasound examination. The amniotic fluid volume was within the normal limits. The amniocentesis revealed a normal male karyotype, 46 XY. The diagnosis of CHAOS with laryngeal atresia was made. After counseling, the couple elected to terminate the pregnancy, and an autopsy was performed.

The aborted fetus weighed 500 g and had a distended abdomen and thorax. Postmortem examination confirmed markedly distended abdomen compatible with ascites. The increased weight was due to ascites and enlarged lungs. Internal examination showed the enlargement of right and left lungs, which weighed 32 g and 24.3 g, respectively (i.e. normal mean±standard deviation, 6.48±2.34 g).

Costal impressions were observed on the external surface of both lungs (Figure 4).

Autopsy revealed an atretic subglottic area, connected through a fibrous cord with the dilated trachea (Figure 5). Histologic section through the larynx showed absence of the cartilage in the subglottic area. The esophagus was normal, without fistula or atresia. No other abnormalities were found. The complete examination was suitable for a laryngeal atresia type II.

Discussion

Laryngeal atresia is one of the rarest congenital malformations⁽¹⁾, part of the CHAOS. Until recently, there have been fewer than 50 cases of prenatally diagnosed laryngeal atresia⁽²⁻⁴⁾.

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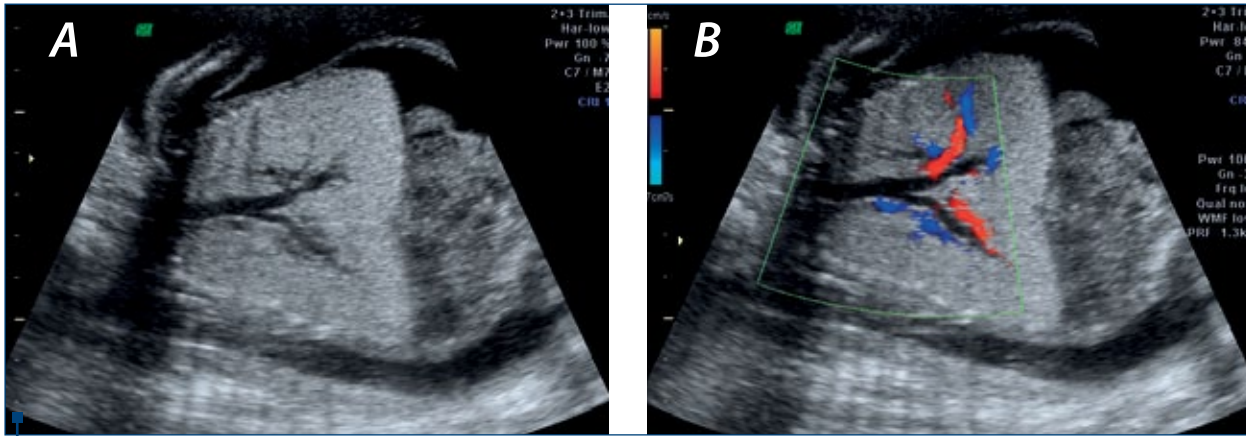


Figure 1. Laryngeal atresia at 21 weeks' gestation. **A.** Coronal image of the chest showing dilated hyperechoic lungs, dilated trachea and bronchial trees with inverted diaphragms. Also a moderately ascites can be seen. **B.** The same coronal image with color Doppler showing the adjacent pulmonary artery branches

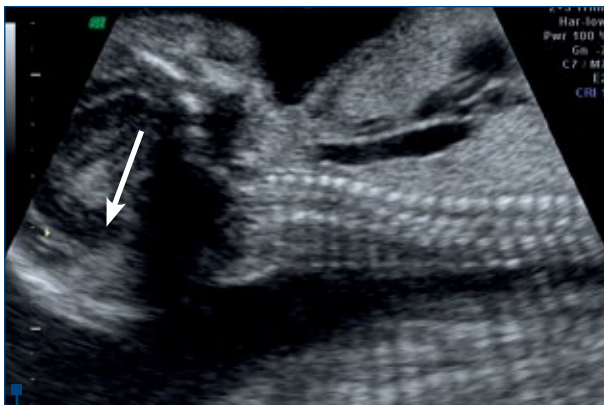


Figure 2. Sagittal image through the fetal chest shows dilated trachea blind-ending at the level of the fetal neck (arrow)



Figure 3. Axial scan of the fetal chest showed large echogenic lungs and the squeezed heart

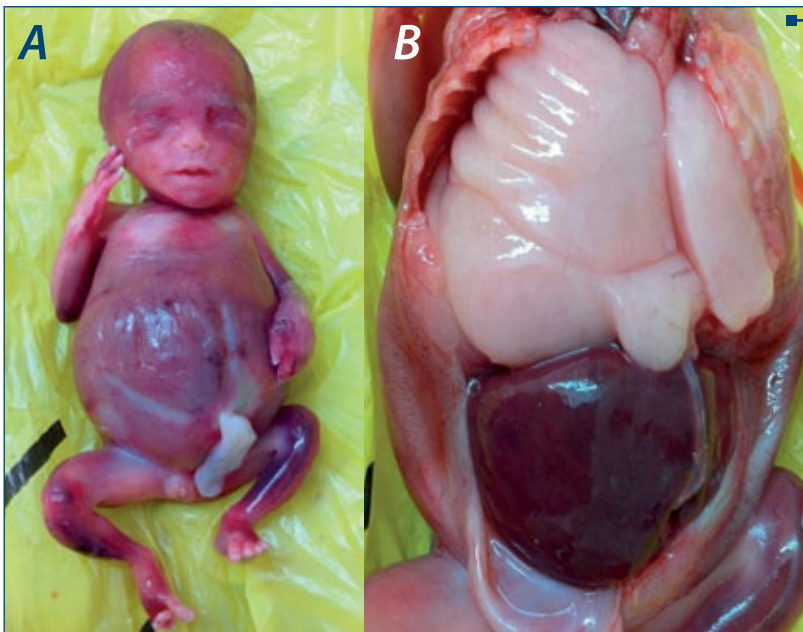


Figure 4. Postabortion. **A.** Markedly distended abdomen. **B.** Anterior view of opened thorax and abdomen showing large pale lungs obscuring the heart and displacing downwards the diaphragm



Figure 5. Bisection of the block, posterior view, demonstrating complete obstruction of the larynx. Laryngeal atresia (small arrow), the tongue (T), the epiglottis (arrowhead) and the vocal cord (large arrow) are shown

According to the Smith and Bain⁽⁵⁾, laryngeal atresia was classified under three types.

Type 1 represents complete atresia of the larynx with a combination of midline fusion of arytenoid cartilage and intrinsic muscles.

Subglottic obstruction is called type 2 and is characterized by dome-shaped cricoid cartilage obstructing the lumen.

Type 3 is the occlusion of the anterior fibrous membrane and fusion of the arytenoid cartilage at the level of the vocal processes.

An arrest of the normal development of the 6th brachial arch, at different gestational stages during embryologic life, seems to be the cause of the lack of larynx canalization⁽¹⁾. The three different types of atresia previously described are not absolute, but rather gradations of a continuous spectrum and indicate that the arrest has happened in different stages of embryonic development.

Prenatal diagnosis was possible starting with 15 gestational weeks⁽⁶⁾, aided by the secondary changes in the fetal lungs, dilated trachea and ascites.

Fetal lungs normally secrete fluid, which is retained because of laryngeal atresia, and that causes their increasing size and echogenicity, fluid-filled trachea and inverted diaphragms.

In such situations, there is an absence of flow in the trachea, evidenced by color or pulsed Doppler, during fetal breathing movements⁽⁷⁾. The level of atresia is difficult to identify prenatally⁽¹⁾. Also, there are associated

fetal cardiac failure, hydrops and polyhydramnios as secondary changes, because of an increased thoracic pressure.

In a fetus with enlarged high echogenic lungs, one should consider a differential diagnosis of a type 3 bilateral cystic adenoid malformation⁽⁸⁾.

Three possible presentations include: a) complete laryngeal atresia without an esophageal fistula; b) complete laryngeal atresia with a trachea-esophageal fistula, and c) near-complete high upper airway obstruction⁽⁹⁾. Many etiologies were proposed including laryngeal or tracheal webs, laryngeal cysts, tracheal atresia, subglottic stenosis or atresia, laryngeal or tracheal agenesis⁽²⁾. However, the most frequent cause appears to be laryngeal atresia.

The condition could be associated with other structural and genetic abnormalities. About 50% of cases reported throughout the literature had other structural malformations. They include: single umbilical artery, left persistent superior vena cava, abnormal fingers and toes, renal and diaphragmatic defects, suggesting a common embryologic origin^(1,8). In the absence of gross abnormalities, Fraser syndrome should be excluded⁽¹⁰⁾. The male-female ratio is approximately 1⁽¹⁾.

Some authors reported association of laryngeal atresia with partial trisomy 9 and 16⁽¹¹⁾, chromosome 5p⁽¹²⁾ deletion and 22q11.2 deletion⁽¹³⁾. These findings support the diagnosis of associated syndromes in order to provide the appropriate counseling for the next pregnancy. Generally, laryngeal atresia is sporadic, although its partial form might have a familial tendency, and an autosomal dominant transmission has been suggested^(10,11,14).

The vast majority of fetuses with complete larynx atresia die because the condition is not recognized prenatally and not treated immediately at birth, or because of other life-threatening anomalies. Only a few fetuses have been saved by ex-utero intrapartum treatment (EXIT)^(4,15,16) or fetoscopic decompression of the trachea⁽¹⁷⁾ while the fetus is still connected with the placenta.

The EXIT procedure allows the establishment of airway patency at birth, while utero-placental gas exchange is preserved. Thus, it has been used successfully to manage different fetal airway obstructions^(18, 19). A reduced number of cases are reported on the literature about the long-term ventilatory and neurodevelopmental outcome of CHAOS, although it seems to be poor in most cases^(20,21). In many cases, even when an EXIT procedure has been performed, the prognosis, in terms of survival and ventilation ability, may be poor, particularly if laryngeal reconstruction is impossible. Antenatal diagnosis might help patients, but ex-utero tracheostomy cannot always prevent hypoxic brain damage. The low rate of long-term survival and severe mental and growth retardation often prevented surgery^(20,22) as the patient fails to reach an age or appropriate clinical condition at which surgical correction is possible.

In our case, we observed on ultrasound at 21 weeks' gestation, enlarged bilateral echogenic lungs, inversion of the diaphragm, dilated trachea and ascites. We could follow the fluid-filled trachea until the inferior part of the larynx.

Conclusions

On the present case, there were no other major anomalies, and the amniotic fluid was normal. Although

the cryptic unbalanced translocation had not been excluded, the fetal karyotype was normal. After discussing the possible outcome with the parents, they decided to terminate the pregnancy. Inferior laryngeal stenosis, type II, without fistula was confirmed by the pathological examination. In the future, if the advance of the technology and increasing experience with EXIT procedure would save fetuses with better results, clinicians could recommend continuing pregnancies. ■

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