Perinatal death due to laryngeal atresia: case report

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Abstract

Laryngeal atresia is a congenital malformation of the airway, which occurs due to the lack of recanalization of the laryngeal lumen during embryogenesis. The detection of characteristic sonographic features during pregnancy is required to consider this diagnosis. In this article we present the case of a 39.2-week gestation with poor prenatal controls and a single late gestational ultrasound that did not report abnormalities. At birth, the fetus was non-vigorous and hypotonic, with insufficient respiratory effort, oral cyanosis, and Apgar scores of 4-4-0, therefore resuscitation maneuvers were performed with great difficulty to achieve ventilation and multiple failed endotracheal intubation attempts, although the medical staff indicated that despite identifying the vocal cords, it was impossible for them to pass the tube. Finally, the newborn died. Suspecting an upper airway malformation, the pathology service was asked to perform a clinical autopsy to confirm the cause of death.

Key words: Larynx, laryngeal diseases, airway obstruction, perinatal death, congenital abnormalities, autopsy.

Resumen

La atresia laringea es una malformación congénita de la vía aérea, que se presenta por la falta de recanalización de la luz laringea durante la embriogénesis. Se requiere de la detección de hallazgos ecográficos característicos durante la gestación para considerar este diagnóstico. En este artículo, se presenta el caso de una gestación de 39,2 semanas con pobres controles...
prenatales y única ecografía gestacional tardía que no reportaba anomalías. Al nacimiento, el feto estaba no vigoroso e hipotónico, con insuficiente esfuerzo respiratorio, cianosis bucal y puntajes de Apgar 4-4-0, por lo que realizaron maniobras de reanimación con gran dificultad para lograr ventilación y múltiples intentos de intubación endotraqueal fallidos, aunque el personal médico indicó que a pesar de identificar las cuerdas vocales les fue imposible el paso del tubo. Finalmente, el neonato fallece. Ante la sospecha de malformación en la vía respiratoria superior, solicitan al servicio de patología realizar autopsia clínica para confirmar causa de la muerte.

**Palabras clave:** Laringe, enfermedades de la laringe, obstrucción de las vías aéreas, muerte perinatal, anomalías congénitas, autopsia.

**Introduction**

Congenital airway anomalies are rare but potentially fatal conditions (1,2). Laryngeal atresia is characterized by complete or almost complete intrinsic obstruction of the lumen, secondary to the lack of recanalization of the bi-tubular cavity derived from the laryngeal primordium or laryngotracheal groove, which occurs during the tenth week of gestation (3), resulting in a firm membrane that completely obliterates the lumen of the glottis (1). It is classified into three types according to Gosavi et al. (4): Type I - complete atresia with fusion of the midline of the arytenoid cartilages, Type II - infraglottic obstruction with dome-shaped cricoid cartilage obstructing the lumen, and Type III - anterior fibrous membrane and fusion of the arytenoid cartilages at the level of the vocal processes. At birth, it presents with severe respiratory distress despite respiratory effort (2), aphonia, cyanosis and death if not treated at delivery (1).

Laryngeal atresia is the most common cause of congenital upper airway obstruction syndrome (hereafter CHAOS) (5), but most cases of laryngeal atresia resulting in CHAOS are isolated and sporadic, with no known risk of recurrence (6). Since the specific type of congenital anomaly is rarely diagnosed prenatally, the term CHAOS is used when the characteristic sonographic findings are identified, since it is more appropriate to refer to the clinical syndrome (5). Therefore, this is the condition that will be discussed in this article, since it has specific diagnostic criteria and management plan to reduce perinatal mortality.

The following is the case of a 39.2-week gestation, with poor prenatal controls and data, which did not allow adequate preparation for delivery,
and which ended in perinatal death despite medical efforts to secure the airway. This case occurred due to a very unusual cause, which was detected in the perinatal autopsy.

**Case report**

A 30-year-old woman (pregnant 4, deliveries 2, miscarriages 1), with no reported pathologic history, with a gestation of 39.2 weeks at the date of last menstrual period. She was evaluated at a second level hospital, where she was admitted in the active phase of labor with 8 cm dilatation, 90% effacement and spontaneous amniorrhea. During pregnancy, the patient had only two prenatal check-ups and a late gestational ultrasound that reported no abnormalities, therefore, limited information on gestation was available at the time of delivery.

Vaginal delivery was performed, with a fetus in cephalic presentation and double circular neck, not vigorous and hypotonic, with poor respiratory effort, oral cyanosis, and Apgar scores 4-4-0, so they started resuscitation maneuvers with great difficulty to achieve ventilation and multiple failed attempts of endotracheal intubation by the neonatology and anesthesiology team, so the baby finally died. A pathological study was requested, as it was suspected to be a possible congenital malformation of the larynx, given that despite observing the vocal cords, it was never possible to pass the endotracheal tube.

At perinatal autopsy, a male fetus was identified with weight and size appropriate for gestational age. The macroscopic examination showed atresia of the larynx in the subglottic region, due to the presence of an abnormal cricoid cartilage in the shape of a dome, which was 4 mm thick when cut (Figure 1). The rest of the bronchial tree showed no other abnormalities and abundant mucous secretions were present. The lungs were expanded, pungent and weighed 105 grams, and the diaphragm was of normal configuration. The rest of the organs showed no abnormalities, no fistulas or ascites were found, and the placenta was not sent for pathological study.
Figure 1. Macroscopic findings: epiglottis (arrow), vocal cords (dots) and atresia area (square), formed by abnormal cricoid cartilage that obstructed the laryngeal lumen and was 4 mm thick.

Microscopic examination showed that the abnormal cricoid cartilage causing the obstruction of the laryngeal lumen consisted of hyaline cartilaginous tissue of the usual characteristics (Figure 2), without other microscopic findings of importance. The pulmonary parenchyma presented multiple dilated air spaces. No other associated anomalies were recognized, and no genetic studies were performed in this case.
Discussion

CHAOS is a very rare and usually fatal entity. Most cases are associated with laryngeal atresia, but other etiologies include laryngeal or tracheal webs, laryngeal cysts, subglottic stenosis, laryngeal or tracheal agenesis and tracheal atresia (5).

It is characterized by a complete or almost complete obstruction of the upper fetal airways (5,7), either intrinsic or by extrinsic compression (8). The diagnosis is usually made during pregnancy by prenatal ultrasound when identifying characteristic findings (9) such as: enlarged echogenic lungs, dilated airways distal to the obstruction, flattened or inverted diaphragm, fetal ascites (7,10), small and compressed heart in midline position, and abnormal abrupt respiratory movements (4). Although so far, no author defines how many ultrasound findings are necessary for the diagnosis of CHAOS, the identification of three of the above mentioned findings suggest a probable diagnosis (10).

The actual incidence of CHAOS is unknown, but it is described to affect 1 per 50,000 newborns (8), its nature is sporadic (4) and most cases die in utero (6).
Obstruction occurs at any level of the respiratory tract and makes fluid exchange between the lungs and the amniotic cavity impossible; therefore, as gestation progresses, the fluid trapped in the bronchial tree dilates the airways and expands the lungs, with consequent compression of the surrounding structures such as the heart, blood and lymphatic vessels, and the diaphragm. Thus, increased mediastinal pressure affects venous return, causing progressive cardiac dysfunction, ascites, edema, hydrops and placentomegaly (11). In addition, esophageal compression due to dilated trachea may result in polyhydramnios (9), although oligohydramnios may be present due to impaired fetal renal function in cases of hydrops (12).

CHAOS has three possible presentations which include: Type I - Complete laryngeal atresia without esophageal fistula, Type II - Complete laryngeal atresia with tracheoesophageal fistula and Type III - Near complete upper airway obstruction (12). In case the syndrome is not recognized during the prenatal stage, it generally results in fetal or perinatal death (8), cases in which the diagnosis requires a careful post mortem macroscopic dissection and histological evaluation, which allows finding the findings associated to the syndrome (7).

Prenatal diagnosis of the congenital abnormality in the present case could not be made due to the lack of prenatal controls and ultrasound scans, which did not allow detection and diagnosis of the laryngeal atresia. It is important to mention and clarify that, if a prenatal diagnosis had been performed, it would have been possible to adequately plan and schedule the time of delivery together with the necessary interventions for the management of the anomaly. The post mortem study evidenced a Type II laryngeal atresia, with dome-shaped cricoid cartilage that completely obstructed the laryngeal lumen and was 4 mm thick. Therefore, typical postnatal interventions such as endotracheal intubation were useless, due to the lack of connection between the oropharynx and the distal trachea (11), and the fatal outcome was inevitable.

Therefore, survival depends on prenatal diagnosis; consequently, ultrasound plays a vital role (9), since the detection of anomalies is fundamental for the planning of a therapeutic strategy, which allows programming an adequate ex-utero intrapartum treatment (7). If the fetus is diagnosed prenatally and survives to delivery, the standard therapeutic approach is an ex-utero intrapartum procedure to secure the airway through a tracheostomy
while on placental support (7,11), which will allow aeration of the distal airways; however, not all facilities are equipped or prepared to perform this intervention in the delivery room (11).

In this case, during the macroscopic study only laryngeal atresia was evidenced, but no dilatation of the airway distal to the obstruction, flattening of the diaphragm, ascites, hydrops, or other anomalies that would allow concluding a diagnosis of CHAOS were identified, although laryngeal atresia is the most frequent cause of the syndrome and according to the literature no case of laryngeal atresia in the absence of CHAOS has been reported (3). Thus, the information resulting from the perinatal autopsy was important to clarify the cause of death and to exonerate the medical team from responsibility. Finally, we emphasize the importance of adequate prenatal monitoring for the detection of this and other anomalies during gestation.

**Conclusion**

Laryngeal atresia is the most frequent cause of CHAOS syndrome, but this diagnosis requires ultrasonographic detection of characteristic findings. Although, according to the literature, no case of laryngeal atresia in the absence of CHAOS has been reported, in this case, no other findings were found during the perinatal autopsy that would allow concluding this diagnosis.

**Ethical considerations**

This article was written after obtaining the patient’s informed consent.

**Conflict of interest**

The authors declare no conflict of interest.

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**References**


