

CHAOS: A fetal autopsy report

Latika Chawla, Nitika Grover, Prashant Joshi, Preeti Singh

Department of Obstetrics & Gynecology, AIIMS, Rishikesh, Uttarakhand, India

ABSTRACT

Congenital high airway obstruction syndrome (CHAOS) is a rare congenital malformation, which results from deficient recanalization of the upper airways. Laryngeal atresia is the most common cause, other etiologies being trachea atresia, laryngeal or tracheal webs, subglottic stenosis, obstructing laryngeal cysts, and laryngeal or tracheal agenesis. There is decreased clearance of the fluid produced by fetal lungs due to obstruction leading to increased intratracheal pressure and thereby secondary proliferative lung growth. The heart becomes compressed in the midline due to hyperexpansion of the lungs causing elevated intrathoracic pressure, decreased venous return, and fetal cardiac failure. This sequence causes ascites, placentomegaly, and eventually hydrops fetalis. We present a case of antenatal diagnosis of a fetus with CHAOS corroborated by fetal autopsy.

Keywords: Autopsy, Congenital high airway obstruction syndrome, fetal laryngeal atresia/stenosis, hyper-echoic lungs

Case Report

A 20-year-old primigravida presented to our hospital at 19 weeks of period of gestation for a routine anomaly scan. Nuchal scan at 11 weeks of period of gestation was reported to be normal. Ultrasound examination revealed large hyperechoic fetal lungs [Figure 1a and b] with flattened diaphragm. The heart appeared compressed [Figure 1b] between the enlarged lungs. There was minimal ascites [Figure 1c]. Amniotic fluid index was normal. No other gross anomaly was evident on detailed ultrasonography. Taking into consideration the aforementioned findings, counseling of parents was done regarding the management strategy and prognosis. Since they were still within legal gestational age limit, they requested for termination of pregnancy. A fetal autopsy [Figure 2a and b] confirmed the ultrasound findings of fetal ascites, enlarged lungs with costal markings on their external surface, and a small heart. On

gross dissection, there was a segment of laryngeal stenosis and the same was confirmed on histopathological examination [Figure 3].

Discussion

According to the characteristic ultrasound findings, diagnosis of congenital high airway obstruction syndrome (CHAOS) was made. CHAOS is a rare malformation that results from deficient recanalization of larynx or trachea around 10th week of period of gestation.^[1] This is considered to be sporadic in nature and true incidence of CHAOS is not known.^[1] It has also been reported to be inherited in an autosomal dominant manner.^[2] The first ever reported case of prenatal diagnosis of CHAOS was by Arizawa *et al.* in 1989^[3] and the term CHAOS was given by Hedrick *et al.* in 1994.^[4]

Laryngeal atresia is the most common etiology, other causes being tracheal atresia, tracheal or laryngeal webs, subglottic stenosis, cysts obstructing larynx, and laryngeal or tracheal agenesis. It may be associated with certain syndromes, commonest being Fraser syndrome.^[1] This autosomal recessive disorder is characterized by cryptophthalmos, laryngeal atresia, syndactyly, and defects of urogenital system. It is also known to occur in association

Address for correspondence: Dr. Latika Chawla,
Department of Obstetrics & Gynecology, Level 6, Academic
Block, All India Institute of Medical Sciences (AIIMS), Rishikesh,
Uttarakhand, India.
E-mail: latika.c@rediffmail.com

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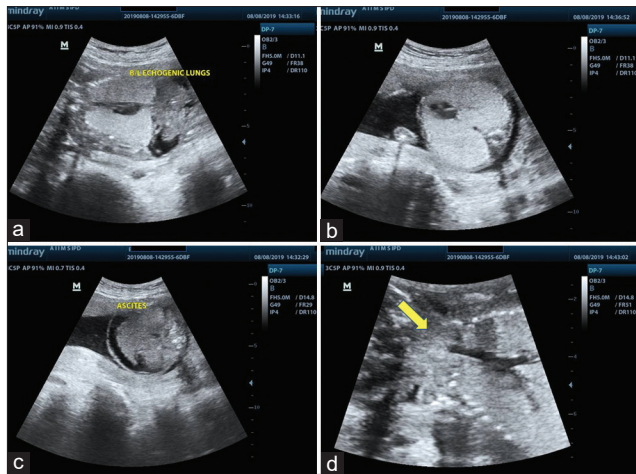


Figure 1: Antenatal 2D ultrasound images of the fetus—bilateral large hyperechoic lungs (a, b), compressed centrally placed heart (b), ascites (c), and laryngeal stenosis (d, yellow arrow)

with VACTERL (vertebral anomalies, anal atresia, cardiac anomalies, tracheoesophageal fistula, renal and limb anomalies) and TACRD (tracheal atresia/laryngeal atresia, cardiac and renal anomalies, and duodenal atresia).

Obstruction of fetal airways results in a series of events that may eventually cause intrauterine fetal demise.^[1] There is decreased absorption of fluid produced by fetal lungs due to obstruction leading to increased intratracheal pressure and thereby secondary proliferative lung growth.^[5] Hyperexpansion of lungs causes compression of heart in the midline, elevated intrathoracic pressure, leading to decrease in venous return and subsequent cardiac failure. This sequence causes ascites, placentomegaly, and hydrops fetalis. A less severe form of CHAOS has been described where a fistula formation leads to spontaneous improvement or resolution of the obstructive features in utero.^[5]

The mainstay of diagnosis is ultrasonography, from 16 to 18 weeks onward, with characteristic findings of bilateral large hyperechoic lungs,^[6] compressed and centrally placed heart, inverted or a flat diaphragm, and ascites. Esophageal compression due to dilated airways may lead to impaired fetal swallowing and polyhydramnios, although few with oligohydramnios have also been reported.^[7] MRI can be of help in localizing the exact level of obstruction and can act as a helpful adjunct if an ex utero intrapartum treatment (EXIT) is being planned.

Most important differential diagnosis of CHAOS is bilateral congenital cystic adenomatoid malformation (CCAM).^[1] Both conditions have bilateral hyperechoic lungs on ultrasonography.^[8] However, in Type III CCAM, systemic arterial supply is clearly demonstrable and site of obstruction associated with distal airway dilatation may be present in CHAOS. Other differential diagnosis include extrinsic causes of obstruction like cervical teratoma, lymphatic malformations, and vascular rings.^[1] Accurate prenatal diagnosis is of utmost importance for further management of this otherwise lethal malformation.

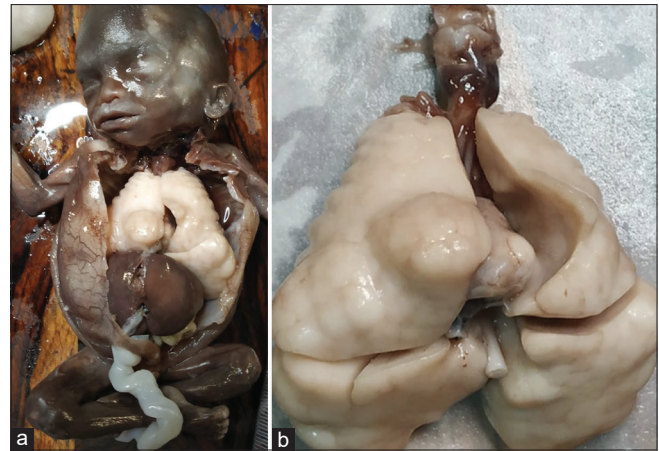


Figure 2: Fetal autopsy showing large lungs (a) with costal impressions on the outer surface and a small heart (a, b)

CHAOS was initially considered to be universally fatal but application of EXIT procedure has significantly improved survival rates. Early prenatal diagnosis of this condition helps in decision-making of whether the parents want to opt for termination, or in case of continuation of pregnancy, it allows to prepare for immediate surgical correction at birth.^[9] This procedure entails partial delivery of fetus by caesarean section whose airway is secured either by intubation or surgical interventions like tracheostomy while still on placental support. During EXIT, fetal laryngoscopy can be used to achieve decompression of laryngeal or tracheal obstruction.^[10] Postoperative complications like permanent tracheostomy, restenosis of the airway, delayed oral feeding, and speech impairment can be unnerving for parents and should be addressed during the prenatal counseling.

In India, since tertiary level fetal medicine facilities are limited, the primary physicians should be aware that fetuses with structural malformation should be investigated at birth/abortion. At least an infantogram, taking pictures of baby and preservation of fetal DNA by cord blood in heparinized vials should be done. Parents should be encouraged for fetal autopsy. This would facilitate genetic testing if required for prognostication in future pregnancies.

Key points

1. CHAOS is a rare congenital malformation characterized by classic prenatal ultrasonographical findings of bilateral large hyperechoic lungs, compressed and centrally placed heart, inverted or a flat diaphragm, and ascites.
2. Laryngeal atresia is most common etiology.
3. CHAOS was initially considered to be universally fatal but application of EXIT procedure has significantly improved survival rates.
4. The primary physicians should be aware that, even if tertiary level fetal medicine facilities are not available, fetuses with structural malformation should be investigated at birth/abortion.

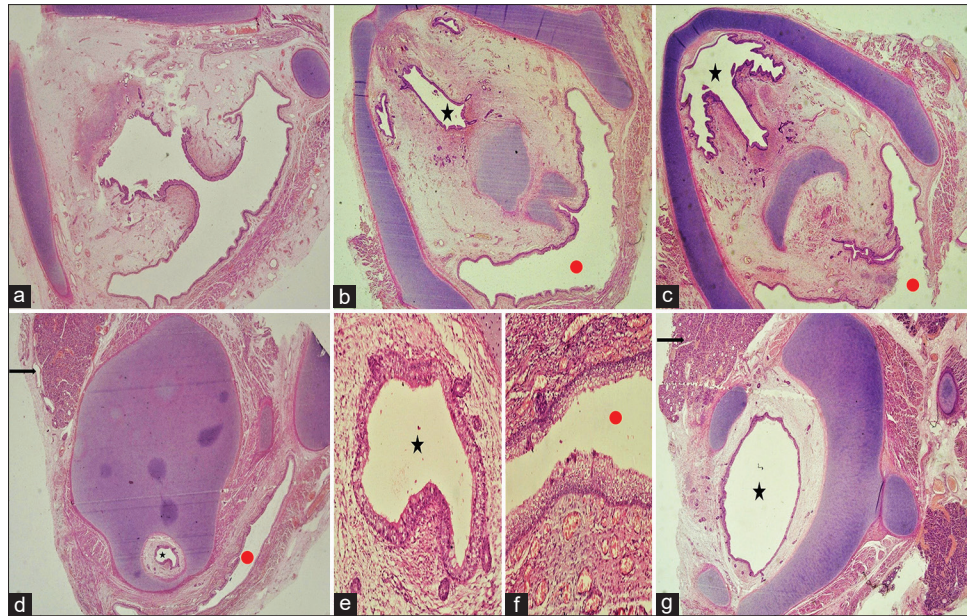


Figure 3: Cranio-caudal serial sections from larynx to trachea demonstrating the epiglottis (a–c; H&E; 20×), larynx and trachea (black star, b–g; H&E; 20× [b, c, d, g] 200× [e]), esophagus (red circle, b–g; H&E; 20× [b, c, d, g] 200× [f]), and thyroid gland (black arrow, d and g; H&E; 20×). Luminal narrowing of trachea is present at the level of thyroid gland (black arrow, d) with normal patency caudally (black arrow, g)

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Conflicts of interest

There are no conflicts of interest.

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