

# Congenital High Airway Obstruction Syndrome (CHAOS)–A Rare Fetal Malformation- Diagnosis on Antenatal Ultrasonography

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**Abstract:** Congenital high airway obstruction syndrome (CHAOS) is the obstruction of the fetal upper airways, which may be partial or complete. It is important to diagnose it antenatally as it is uniformly lethal and leads to stillbirth or death shortly after delivery. Antenatal diagnosis is made by identifying echogenic and enlarged lungs, dilated bronchi, inverted /flattened diaphragm and ascites on ultrasound. We present a rare case of congenital high airway obstruction, diagnosed on antenatal ultrasonography at 18 weeks of gestation.

**Keywords:** Congenital high airway obstruction syndrome, Laryngeal atresia, Antenatal ultrasound

## 1. Introduction

Congenital high airway obstruction syndrome is rare fetal malformation caused by obstruction of fetal airway because of laryngeal or tracheal atresia, sub glottis stenosis, laryngeal cyst or laryngeal web. This clinical condition was brought into notice firstly by Hedrick in the late 1900<sup>1</sup>. CHAOS is usually caused by atresia or stenosis of the larynx or trachea. The true incidence of CHAOS is unknown. It is important to diagnose it antenatally as it is uniformly lethal. If the syndrome is unrecognized during the prenatal period, it usually results in stillbirth or death shortly after delivery<sup>2</sup>. The prenatal ultrasound findings of CHAOS include large echogenic lungs, flattened or inverted diaphragms, dilated airways distal to the obstruction, and fetal ascites and/or hydrops<sup>3</sup>. Here we present a case of antenatally diagnosed CHAOS.

## 2. Case Report

A 22 year old primi gravida was referred for routine anomaly scan at 19 weeks of gestation. There was no history of consanguinity and the family history was unremarkable.

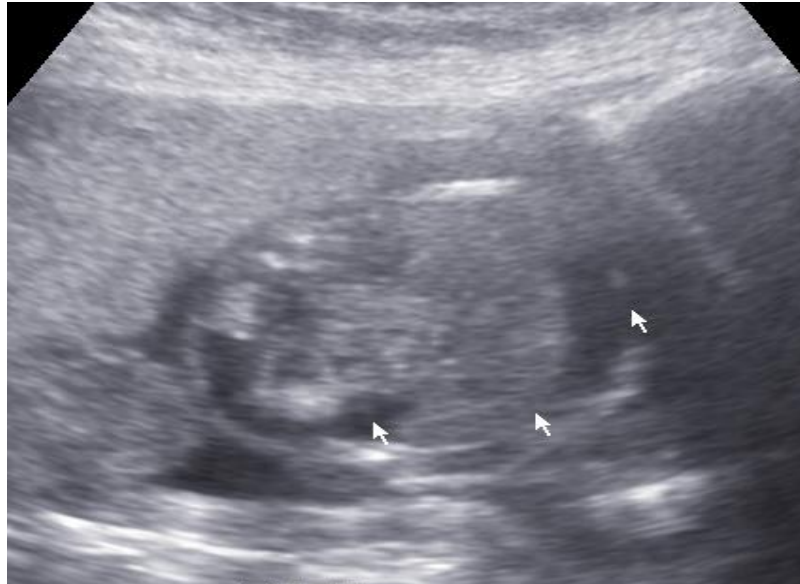
Ultrasound examination revealed a single live intrauterine fetus corresponding to 18W+2D period of gestation. Amniotic fluid was within normal limits. Bilateral lungs were echogenic and enlarged with small heart compressed in between the lungs. (Fig.1), dilated bronchi and trachea (Fig.2), mild ascites (Fig.3), and flattened diaphragm (Fig.4). Based on these findings, the diagnosis of CHAOS was made. Patient and her family members were counselled regarding the possible unfavourable outcome of the pregnancy. The patient opted for termination of pregnancy.



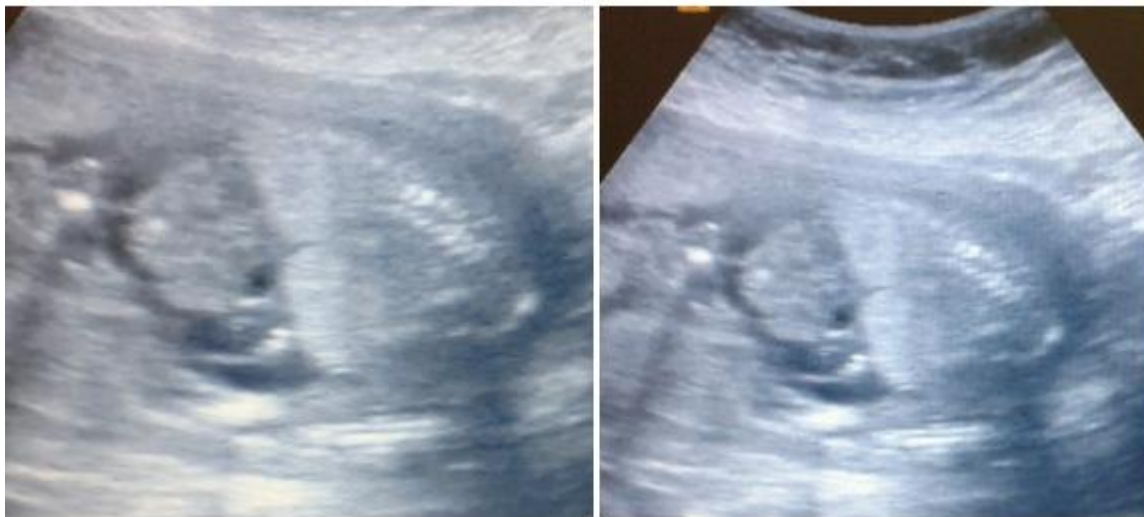
**Figure 1:** Antenatal sonography showing large hyperechoic lungs and centrally displaced heart which appears small and compressed.



**Figure 2:** Antenatal sonography showing dilated main bronchi and trachea, large hyperechoic lungs and flattened/inverted diaphragm



**Figure 3:** Antenatal sonography showing fetal ascites .



**Figure 4:** Antenatal sonography showing large hyperechoic lungs and flattened diaphragm.

### 3. Discussion

CHAOS is caused by complete obstruction or severe narrowing of the upper airway. Causes of obstruction are laryngeal atresia, subglottic stenosis, laryngeal web, a completely occluding laryngeal cyst, or, rarely, a tracheal obstruction. Laryngeal atresia is a rare congenital malformation and is usually fatal. CHAOS is mostly sporadic, and the exact incidence is not known<sup>1</sup>. The malformation is caused by non- development of 6<sup>th</sup> branchial arch during normal embryological development.

Smith and Bain<sup>3</sup> have classified laryngeal atresia into three types.

- 1) In type I there is complete atresia of larynx with midline fusion of arytenoid cartilages and intrinsic muscles.
- 2) In type II there is infraglottic obstruction that is characterized by a dome shaped cricoid cartilage obstructing the lumen.
- 3) In type III there is occlusion of anterior fibrous membrane and fusion of arytenoid cartilages at the level of vocal process.

In a healthy fetus, the fluid secreted by fetal lung is absorbed through the tracheobronchial tree. However, in case of any obstruction in the tracheobronchial tree, this fluid cannot be cleared. The accumulation of the fetal lung fluid results in gradual increase of intratracheal pressure leading to enlargement of the lungs. It is the beginning of a chain reaction: the enlarged lungs cause compression of the heart and great veins. Due to the compression, the heart replaces centrally and becomes small and dysfunctional. Decreased venous return and dysfunctional cardiovascular system end in ascites and hydrops. The diaphragm flattens or inverts according to the severity of the process<sup>2</sup>.

The early recognition of this rare malformation is important because it allows the option of termination of pregnancy to be provided earlier, with less risk of physical and psychological complications. The main diagnostic tool for prenatal diagnosis of CHAOS is sonography which has typical findings on evaluation. As a natural conclusion of the pathological process, bilateral large hyperechoic lungs, small, compressed, and centrally placed heart, flattened or inverted diaphragm, and ascites are characteristic findings on sonographic examination<sup>5</sup> as seen typically in our case.

Regarding the amniotic fluid index, compression of the esophagus by dilated airways may lead to polyhydramnios, as the fetal swallowing of the fluid is disrupted. On the other hand, impaired swallowing of the fetus may also cause oligohydramnios<sup>2</sup>. The amniotic fluid quantity thus is not a constant marker for the diagnosis, the fetus can present at either end of spectrum. The gestational age at the diagnosis may affect the amniotic fluid quantity. Polyhydramnios may not be present due to the examination in the early 2nd trimester as exemplified in our case.

The typical sonographic findings can also be recognized on MRI including voluminous lungs, centrally displaced small heart, inverted diaphragm, and ascites. Sonography is first-line diagnostic imaging tool due to its low cost and widespread use. However, especially if any fetal surgical intervention is planned, MR imaging can be used additionally by following the dilated airway up to the level of obstruction, as it is more effective for detecting the exact level of obstruction<sup>6</sup>.

There are some genetic syndromes associated with CHAOS such as Short-rib polydactyl syndrome (SRPS), Shprintzen–Goldberg Omphalocele syndrome (SGOS) and VATER/VACTERL association. In addition to these syndromes, some chromosomal abnormalities (deletions of 22q11.2, deletion of chromosome 5p<sup>7</sup>, 47, XXX, partial trisomy 9 and partial trisomy 16q) have been reported in association with CHAOS. Pierre Robin syndrome, the primary developmental defect of the mandible, may be a cause of CHAOS. The other oropharyngeal and neck anomalies are rarely responsible for the CHAOS sequence. CHAOS may occur as part of the Fraser syndrome (tracheal or laryngeal atresia, renal agenesis, microphthalmia, and syn- or polydactyly)<sup>8</sup>.

CHAOS is most often misdiagnosed as bilateral congenital cystic adenomatoid malformation (CCAM)<sup>1</sup>. CCAM (especially type III) and upper airway obstruction secondary to intrinsic causes such as tracheal or laryngeal atresia or stenosis and tracheal webs similarly have bilateral uniform hyperechogenic appearance of the fetal lungs on sonographic examination<sup>9</sup>. In order to make a differentiation between CHAOS and CCAM type III, the obstruction site with distal airway dilatation (present in CHAOS) and the systemic arterial supply (present in CCAM type III) must be clearly seen.

Congenital high airway obstruction syndrome should also be differentiated from extrinsic causes of tracheolaryngeal obstruction. Some of these extrinsic causes are lymphatic malformation, cervical teratoma, and vascular rings like double aortic arch<sup>1</sup>.

Some intrauterine and intrapartum treatment options are available to manage this condition. It is possible that earlier fetoscopic bronchoscopy may decompress the upper airway and allow an efficient functional lung development<sup>10</sup>. The only available postnatal treatment option to sustain the survival of the fetuses with CHAOS is ex utero intrapartum treatment (EXIT) with tracheal intubation and safe airway control<sup>11,12</sup>. However this treatment is still in its budding stage and most of the fetuses diagnosed with congenital high

airway obstruction are seen with stillbirth or death immediately thereafter.

#### 4. Summery

Congenital high airway obstruction syndrome is a rare clinical congenital entity presenting in the fetus with profound lethal outcome for the fetus. Therefore it is important to diagnose this clinical entity earliest antenatally to provide the mother for option of termination of pregnancy in view of virtually lethal post natal outcome.

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