CONGENITAL HIGH AIRWAY OBSTRUCTION SYNDROME (CHAOS): PRENATAL DIAGNOSIS AND PERINATAL INTERVENTION – A CASE REPORT

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ABSTRACT

Congenital high airway obstruction syndrome (CHAOS) is a rare life-threatening fetal anomaly which occurs as a result of either partial or complete obstruction of the upper fetal airway. Prenatal recognition is important for multidisciplinary team management. We present a case with CHAOS diagnosed antenatal imaging and the baby was delivered via ex utero intrapartum treatment (EXIT) procedure. The airway managed to be secured via tracheostomy while maintaining the fetus on feto-placental circulation. However, oxygen saturation started to decline despite successfully securing the upper airway. Unfortunately, we lost the baby despite active resuscitation possibly due to insufficient lung function secondary to chronic obstruction prenatally.

Keywords: Airway obstruction; Ex utero intrapartum treatment; Larynx; Prenatal ultrasonography

DOI: 10.51407/mjpch.v28i1.170

CASE REPORT

MJPCH Vol. 28 (1) June 2022

The Malaysian Journal of Paediatrics and Child Health (MJPCH) | (June 2022) | Page 84 of 90

Introduction

Congenital high airway obstruction syndrome (CHAOS) is an uncommon condition, and the true incidence is still unknown. This is probably due to fetuses with CHAOS dying in utero or they die within minutes after delivery. With the evolution of fetal medicine together with the advances in antenatal ultrasonography (US) and improvement in fetal magnetic resonance imaging (MRI) images, this life-threatening fetal condition is slowly gaining attention. Kalache et al mentioned that few early cases were detected by antenatal US in 1989, 1990, and 1992 [1]. A list of structural fetal anomalies can be detected early in pregnancy, allowing for better treatment and delivery preparation, as well as counseling regarding the prognosis and outcome.

Typical US findings in a fetus with CHAOS include lung enlargement with a flat diaphragm, dilated tracheobronchial tree below the site of obstruction, and massive ascites. Spontaneous resolutions of these US findings are seen in CHAOS later at 35th - 37th week. However, there is little discussion regarding the pulmonary function when there is a resolution of the MRI findings. Pulmonary functions may not return to normal when there is long-standing chronic obstruction of the upper airway leading to lung parenchymal damage. This was observed in our patient who was delivered via ex utero intrapartum treatment (EXIT) procedure.

Case Report

A 33-year-old teacher, G5P1+3, was referred for antenatal diagnosis of CHAOS. This lady was first...
seen by the obstetrician at 20 weeks of gestation when her US revealed large fetal ascites, inverted diaphragm, and echogenic hyperinflated lungs. The trachea and the main bronchi were dilated (Figure 1). Amniocentesis was normal and Fraser syndrome was excluded. There was no renal abnormality detected by US. A repeat US at the 32nd weeks of gestation showed resolution of the ascites and reduction of the lung echogenicity. The diaphragm had returned to normal shape. A coronal view of the neck showed a narrow connection between the trachea and pharynx. A fetal MRI performed was preceded at the 38th week of gestation showed complete gap seen in fluid-filled with high signal trachea below 0.5cm below the tip of epiglottis (possible upper trachea or larynx) consistent with the airway obstruction. Airway distal to the level of obstruction remains dilated but still patent. Lung volume showed reduced signal possibility of reduced lung volume (Figure 2). Parents were counseled regarding the possible unfavorable outcome of the pregnancy. The option about EXIT procedure was offered to parents in view of anticipating difficulty in securing airway upon delivery. The elective caesarian section with EXIT procedure was carried out after discussion and preparation done with obstetrics, anesthesia, and pediatric team. After delivering the fetal head and one upper limb, direct laryngoscopy was performed to assess the airway. There was a mass in the subglottic area which obstructs the airway. The location of the mass was similar to the level predicted by antenatal MRI. Intubation with the smallest endotracheal tube (ETT) was impossible as there was no opening visualized. Tracheostomy was performed as an option to secure the airway (Figure 3). The trachea was soft and appeared translucent. Upon making an incision in the trachea, mucoid thick secretion was observed coming out from the tracheal lumen. ETT size 3 was first introduced into the trachea and the cord was clamped and cut. The newborn was transferred to the next operating table. ETT was later changed to tracheostomy. Patient oxygen saturation continued to desaturate despite continuous manual bagging. The child later became bradycardic and CPR was commenced immediately. However, the patient eventually expired despite all active resuscitation. No syndromic features were seen in this baby. A biopsy of the subglottic mass revealed fibromuscular tissue consisting of skeletal muscle bundles with a localized region of hemorrhage. Otherwise, no malignancy cells were seen. The autopsy was not done in this case as the family refused.

Figure 1. Ultrasound at 20th week of gestation showing dilated trachea and main bronchi (a), echogenic hyperinflated lungs (b), inverted diaphragm (c) and large fetal ascites (d).
Figure 2. Sagittal T2-weighted fetal MRI performed at 38th week of gestation showed a complete gap (arrow) in the fluid-filled airway with high signal airway below 0.5cm below the tip of the epiglottis (possibly upper trachea or larynx), indicating an airway blockage. The airway is dilated (arrowhead) but yet patent distal to the blockage. Lung volume revealed a lower signal, indicating that lung volume may be reduced (L).

Figure 3. Elective caesarian section with EXIT procedure was carried out. Tracheostomy was performed as an option to secure an airway while maintaining feto-placental circulation on placental support.
Discussion
Congenital high airway obstruction syndrome (CHAOS) is a rare clinical condition that was first defined in 1994. Mostly, the cases are sporadic and the true incidence is unknown [2]. CHAOS has been documented in over 100 cases in the past [3]. No true local incidence documented so far. It is detected by the antenatal US which shows total or near-total obstruction of the upper airway and usually the prognosis is poor. This syndrome is believed to occur when the upper airway fails to canalize around the 9th to 10th week of gestation thereby causing an obstruction [3]. Thus, laryngeal or tracheal atresia constitutes the most reason behind CHAOS [4]. However, rare causes of laryngeal cyst, web, and stenosis can also cause congenital obstruction of the fetal airway [2]. No author mentioned any skeletal tumor that is related to CHAOS. If the prenatal diagnosis of possible upper airway obstruction is made and could not determine the type of obstruction, the term CHAOS is used [5]. Our case fulfilled the definition of the condition whereby any condition that can cause airway obstruction during fetal development can relate to CHAOS. It should be distinguished from extrinsic causes of laryngotracheal obstruction such as lymphatic malformation and cervical teratoma. It is essential to differentiate those conditions for planning subsequent management [6].

In this case, at the 20th week of gestation, we noted the hyperinflated lungs, dilated tracheobronchial tree, inverted diaphragm, and fetal ascites. In normal fetal development, the lungs will secrete a fluid that subsequently is absorbed through the tracheobronchial tree. The obstruction prevents fluid to be cleared and leads to fluid accumulation inside the lungs. Subsequently, the trachea and bronchi will dilate at the level below the obstruction. Increase intratracheal pressure cause hyperinflated lungs and directly compress the heart and great veins, thus the heart will be displaced centrally and becomes small. Decrease venous return cause ascites and hydrops formation. The diaphragm flattens due to enlarged lungs and may invert in more severe cases. In addition, the alveolar walls thin, the number of type II pneumocytes decreases, and the amount of surfactant produced decreases proportional to the duration of the tracheal obstruction [6,7].

The antenatal US is the first-line modality for diagnostic procedures. Most diagnostic findings observed in the fetus with CHAOS are due to increased intratracheal pressure and distention of the tracheobronchial tree secondary to the accumulation of fluid within the lungs. Cardiac changes include the appearance of an elongated heart, septal shift, and small, compressed heart chambers [6]. The US findings in this fetus were the results of obstruction at the level of the subglottic area, which prevents the fluid produced by the lung parenchyma to flow out into the amniotic fluid. These subsequently result in hyperinflated lung and dilated tracheobronchial tree. However, the US has limitations in detecting the correct level of obstruction. Magnetic resonance imaging (MRI) is more effective for detecting the level of obstruction due to higher soft-tissue contrast [6].

A fetal MRI will show a gap seen in the fluid-filled, high signal airway at level of larynx with airway dilatation distal to the obstruction. The MRI findings of a dilated airway below the degree of blockage were seen in all 7 fetuses and included an increase in the lung signal, notably increased lung volumes with flattened or inverted hemidiaphragms, a centrally positioned and compressed heart, ascites, and placentomegaly [7]. Another case had been reported which showed large, hyperintense lungs causing diaphragm inversion on MRI [6]. There was also increased signal with a gap at the level of the larynx illustrated within the dilated trachea. The cardiac findings and ascites were verified, and no other abnormalities were discovered.

The level of airway obstruction must be identified to diagnose CHAOS and to distinguish it from bilateral lung masses like a congenital pulmonary airway malformation (CPAM) or other causes of extrinsic airway obstruction such a double aortic arch by demonstrating an abnormally dilated airway distal to the obstruction. Study done in Cincinnati found that two patients with past antenatal ultrasound diagnoses of bilateral CPAM were referred for further investigation. However, MRI revealed the right diagnosis of high airway blockage, which had been missed by US, and changed the prognosis and management of the fetus. At the same time, all babies with postnatal surgical evaluation were conducted and the level of airway obstruction was confirmed [7]. Bilateral congenital cystic adenomatoid malformation
(CCAM) particularly type III often misdiagnosed with CHAOS. In CCAM, ascites and flat diaphragm are absent with involvement unilateral chest and prominent systemic arterial supply [8].

Fetal MRI has also been useful in detecting these related anomalies and suspected genetic disorders. Accurate diagnosis of related syndromes aids management decisions and parental counseling. Several fetuses had been found to be associated with some abnormalities such as micrognathia, small kidney or horseshoe kidney and articular anomalies [7].

The antenatal US findings in this fetus at the 32nd week showed the lung echogenicity had improved and the diaphragm had returned to its original position. Spontaneous resolution of the US findings in CHAOS has been reported by some authors. This can be the result of spontaneous perforation of an obstructing lesion like web or presence of tracheoesophageal fistula leading to decompression of the lungs as the fluid is released into the amniotic cavity [6]. One case from total of 7 case series was the only survivor with marked lung improvement due to perforation [7]. We also witnessed a resolution of the prenatal US findings i.e. airway decompression, but without identifying any fistula or perforation. Some author obstruction suggest that an intratracheal fetoscopic laser laryngotomy may be helpful in detecting the hole and treating selected cases of fetuses with CHAOS due to the improvement of patients with spontaneous perforations of the obstruction [7].

However, resolutions of the US findings are not always associated with improvement in lung function. Significant pulmonary compromise at birth even after apparent prenatal resolution should be anticipated and must be informed to parents during a pre-EXIT counseling session. Based on the MRI findings, which showed total airway obstruction, we explained to this patient that the newborn would most likely be tracheostomized upon delivery of the head. The failure to intubate was predicted and the parents were notified. The presence of impaired pulmonary function was not highlighted in this case. The function of the lung could not be detected by MRI but due to chronic abnormalities lead to long term effects of poor development. Postnatal abnormalities in live born neonates with CHAOS include respiratory distress caused by abnormal lung development, diaphragmatic dysfunction likely caused by diaphragm stretching, tracheomalacia, and capillary leak syndrome [9].

The presence of perforation, hydrops, and other anomalies affects the severity of CHAOS and the survival rate. Associated other anomalies generally predicts more severe CHAOS [9]. Early presentations with hydrops or associated anomalies usually give rise to poor prognosis, with a high rate of fetal demise and poor survival even with the EXIT procedure [10]. However little is known or discussed by authors regarding the function of the lung after a long period of compression by fluid followed by decompression at a certain stage. Although the antenatal US findings in this child improved by 32 weeks, the MRI at 38 weeks revealed a dilated trachea, indicating that the airway had been significantly impacted. As a result, this newborn’s lung function was predicted to be poor. This was confirmed when the oxygen level did not improve despite vigorous resuscitation after delivery.

Usually, CHAOS is an isolated malformation, but it may be associated with a genetic disorder. The most common is Fraser’ Syndrome. It is autosomal recessive and characterized by laryngeal atresia, urogenital defects, syndactyly and cryptophthalmos [6]. Other syndromes that are related to CHAOS are Cri-du-Chat Syndrome, short-rib polydactyly syndrome, and velocardiofacial syndrome [6,9]. In this case, there was no family history of a syndromic child, and an amniocentesis test done revealed a normal result. Most of the cases reported normal karyotypes and did not associate with syndromic features [11,12].

The EXIT procedure is also known as ‘operation on placental support’ (OOPS), or airway management on placental support. The procedure helps to secure the neonatal airway while the fetus continues to be on placental support. This method has been used in delivering a CHAOS baby or whenever cases with difficulty to establish the airway at birth [13]. The importance key is to diagnose any structural anomalies during the prenatal period. Any delay of over 5 minutes may result in hypoxic-ischaemic encephalopathy (HIE). The upper limit for EXIT procedure is approximately one hour as fetal gas exchange can be supported by ex utero-placental circulation [2].
In 1998, a case with CHAOS has been reported to be the first successful case delivered via EXIT [14]. Generally, the procedure will be done at the 37th – 38th week of gestation unless there are specific indications [13]. Two CHAOS cases reported delivered earlier at 29th and 30th week due to rupture membrane and regular uterine contraction but both cases died after 2 hours of resuscitation [3,11]. One case was confirmed at 26th week with normal male karyotype and MRI showed no other abnormalities except for CHAOS features [11]. Another case with antenatal US at 18th weeks with addition of cleft lip and palate. No other abnormalities were seen. However, parents refused for further work up and intervention. Post delivered noted cleft lip palate, omphalocele, small genitalia with bilateral hydrocele [3]. CHAOS is associated with high mortality rated anomaly and 100% perinatal mortality if without any intervention [12]. Most of the cases reported had been terminated prenatally after a discussion with the parents regarding the unfavorable outcome.

The postnatal management of newborn neonates with CHAOS is challenging, and the prognosis of affected infants is frequently poor [12]. In 2002 and 2003, few cases were reported with the long-term survivor of CHAOS due to tracheal atresia as a result of the EXIT procedure (15,16). The cases were under follow up to 3 and 1/2 years and 5 years old. The first long-term CHAOS survivor regains diaphragmatic function recovery at 9 months of age, allowing him to be weaned off from ventilator assistance. He had a rib cartilage graft in a laryngotracheoplasty at the age of 17 months. Subsequently grow with normal development and a patent airway, and can speak at the age of five [16]. The consequences of survival baby post EXIT are long-term tracheostomy, chronic breathing problem, capillary leak syndrome, tracheobronchomalacia, diaphragmatic dysfunction, speech, swallowing problems and require multiple operations like trachea reconstruction [11,13,16].

**Conclusion**

Fetus with CHAOS carries high mortality risk. Advancement in the prenatal US predict the fetal outcome based on the structural or anatomical abnormalities present. However, the functional outcome of the vital organs is difficult to predict even with the resolution of early prenatal US findings. Therefore, thorough counseling pre-delivery and multidisciplinary approaches are mandatory.

**References**


