

Medical Case Report

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Congenital high airway obstruction syndrome: prenatal ultrasound diagnosis and literature review

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Abstract

Congenital high airway obstruction syndrome (CHAOS) is a rare congenital anomaly involving the upper airways (trachea, larynx). It is a life-threatening condition whose true incidence is unknown. The obstruction may be due to tracheal/laryngeal atresia, stenosis, or the presence of a mass lesion. Prognosis is poor, generally resulting in stillbirth or intrauterine fetal demise. Ex utero intrapartum treatment (EXIT) is possible if the condition is detected early. We present a case of CHAOS diagnosed during a second-trimester anomaly scan with postmortem confirmation and literature review.

Keywords: Congenital high airway obstruction, ultrasound, echogenic lungs, tracheal stenosis

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INTRODUCTION

Congenital High Airway Obstruction syndrome (CHAOS) is a rare, life-threatening anomaly defined as upper airway atresia or aplasia with distal airway dilatation, expanded lungs, fetal hydrops, and ascites [1]. This anomaly may also be due to intrinsic or extrinsic compression of the airway by cervical tumours [2]. The clinical condition was first coined by Hedrick in 1994 [3], and is usually caused by atresia of the trachea or larynx. Laryngeal atresia, however, remains the commonest cause [4]. The clinical outcome of this condition is poor and most cases result in stillbirth [1]. Due to the advances in prenatal imaging, namely Magnetic Resonance Imaging and improved ultrasonography equipment resolution [5], prenatal diagnosis is possible, making management options such as ultrasound-guided intrauterine management, ex utero intrapartum treatment, and medical termination of pregnancy possible. Sonographic features include enlarged hyperechogenic lungs, inverted diaphragm, fetal ascites, and fetal hydrops [6]. We report a case of CHAOS diagnosed during a routine anomaly scan. Though rare,

more than 100 case reports have been reported in the literature [7]. To the best of our knowledge, this is the first case report in Ghana and our literature search did not yield any results for West Africa.

CASE PRESENTATION

A 29-yr. old female with Gravida 3 para 1 and 1 spontaneous abortion (G3P1+1SA) at 21 wk presented to the Obstetric Emergency Unit with complaints of reduced perception of fetal movement and rib pain for about a month. There was neither bleeding per vaginum nor urinary symptoms. The booking visit was at 11 wk and 5 days and an ultrasound scan done showed a single live intrauterine gestation. She had had three antenatal visits and the pregnancy had been uneventful until the complaint. She had been taking the pregnancy supplement Pregnacare® (Vitabiotics, UK), and had received one dose of sulphadoxine-pyrimethamine for malaria prophylaxis. The patient disclosed no use of herbal medication, no drug allergies, no alcohol use, cigarette smoking, or recreational drug use. There was no significant personal or family history of congenital disorders, diabetes, hypertension, or other medical conditions. Her first pregnancy was 6 yr. ago and uneventful. The baby was delivered by spontaneous vaginal delivery at 38 wk, and the child is alive and well.

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The second pregnancy was 1 yr. ago at 8 wk, culminating in spontaneous incomplete abortion managed by an evacuation of the uterus that had no post-procedure complications. Products of conception were not sent for histopathological assessment and the cause for the pregnancy loss was not ascertained. On examination, the patient was stable and not in obvious pain; vitals on presentation were within the normal range. Symphysiofundal height was 19 cm – which was compatible with gestational age – and fetal heart rate was 124 bpm. Urine dipstick was unremarkable. An anomaly scan was requested. The ultrasound showed a single live

fetus with an average estimated gestational age of 21 wk with bilaterally enlarged echogenic lungs that compressed the heart and displaced it centrally. The four heart chambers were visualized and appeared normal (Plate 1). Both hemidiaphragms were inverted with mild tracheal dilatation and massive fetal ascites (Plate 2a). Color Doppler revealed absent flow in the dilated trachea (Plate 2b). Also noted were subcutaneous oedema and increased nuchal fold thickness consistent with fetal hydrops. The amniotic fluid was significantly reduced with an index of 5.2 cm and below the 5th percentile for the corresponding gestational age. The cerebellum, upper lip, spine, gastric bubble, both

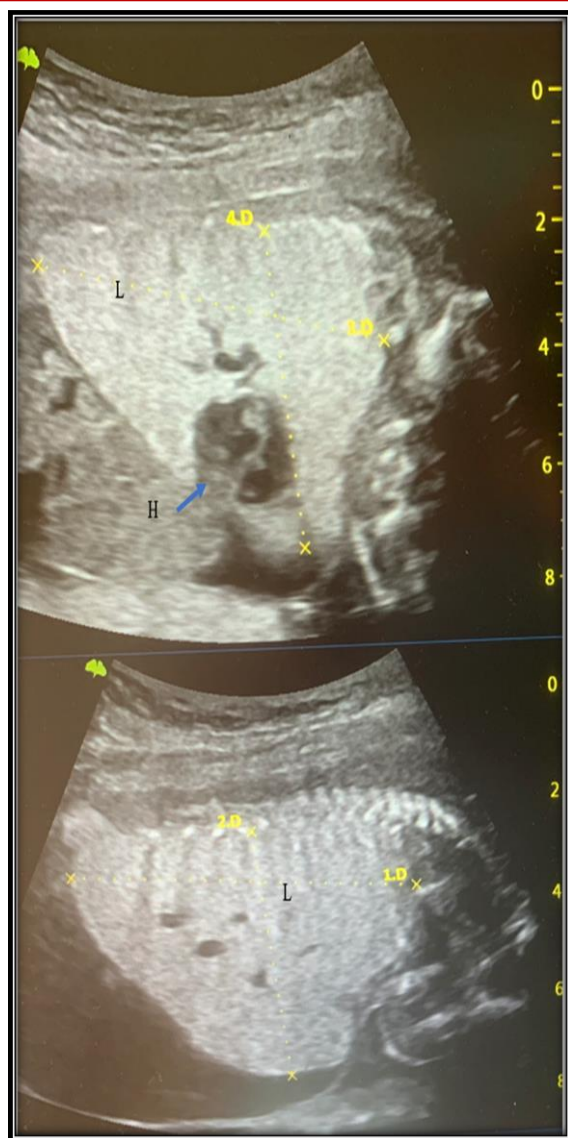


Plate 1: Sagittal fetal sonogram showing bilateral enlarged echogenic lungs 'L', compressing, and displacing the heart 'H' centrally. The left ventricle and left atrium are showing (x 2 magnification)

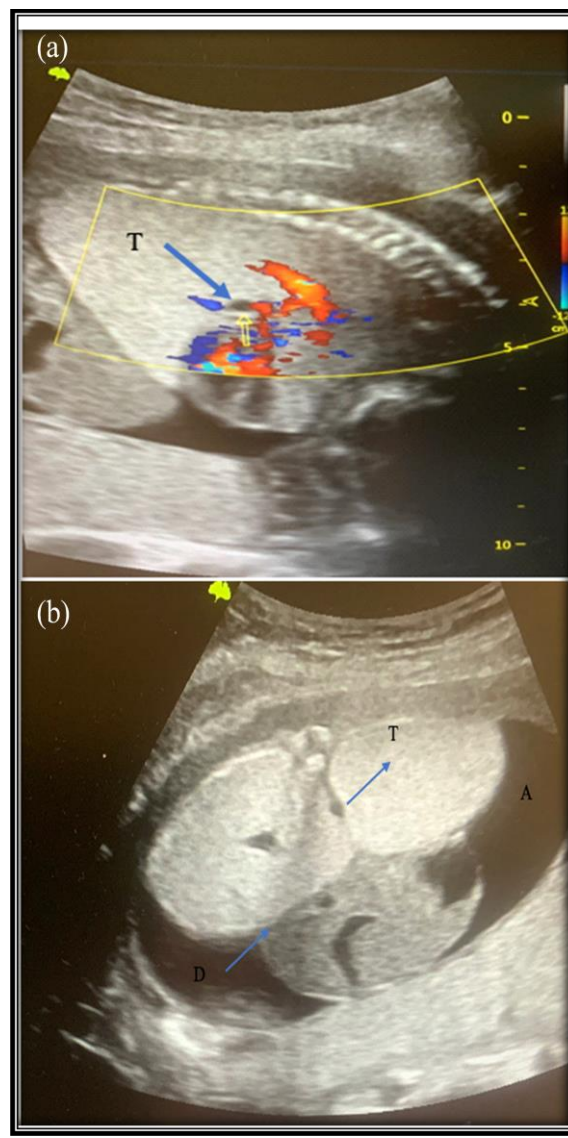


Plate 2: (a) Transverse sonogram of both hemidiaphragms 'D' are inverted with mild tracheal dilatation 'T' and massive fetal ascites (b) Colour Doppler revealed absent flow in the dilated trachea. The Colour Doppler sonogram also shows absent flow in the dilated trachea 'T'. Sonograms at x 2 magnification

kidneys, urinary bladder and the limbs were normal. The placenta was in the upper posterior location and appeared normal. Abdominal circumference was not measured due to the presence of fetal ascites. Based on the above ultrasound findings, the diagnosis of CHAOS was made. The couple were counselled in detail regarding the diagnosis, prognosis (likelihood of intrauterine fetal demise and poor post-delivery outcomes), and available management options. The couple opted for an elective medical termination of the pregnancy to be performed. Medical termination of

pregnancy was done using the International Federation of Gynecology and Obstetrics recommended misoprostol only regimen appropriate for the gestational age. A female fetus was delivered weighing 800 g with generalized oedema of the skin and gross abdominal distension (Plate 3). At postmortem, a well-preserved, pink, non-syndromic female fetus 800 g was seen with generalized skin and subcutaneous tissue oedema as well as an abdomen distended by fluid (Plate 3). Internal findings revealed clear serous ascites with scanty pericardial and pleural effusions.



Plate 3: Gross image of a well-preserved, pink, non-syndromic female fetus 800 g with generalized skin and subcutaneous tissue edema as well as distended abdomen by fluid



Plate 4: Gross image of the lungs show enlarged lungs with surface rib markings

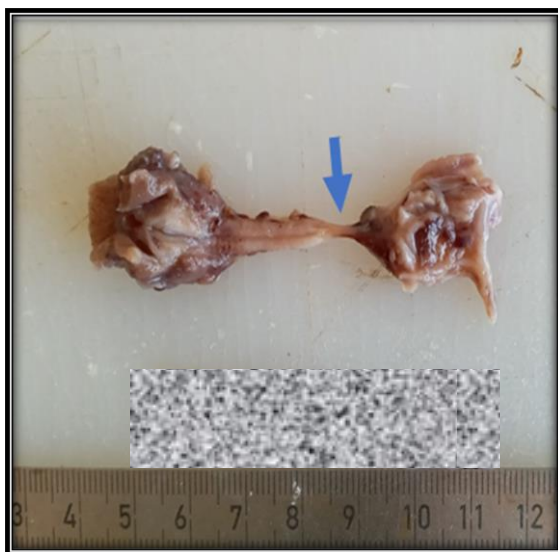


Plate 5: Gross image of the trachea showing stenosis of the distal trachea and lacking the usual cartilaginous rings in this segment, making it difficult to probe through the bifurcation into the bronchi

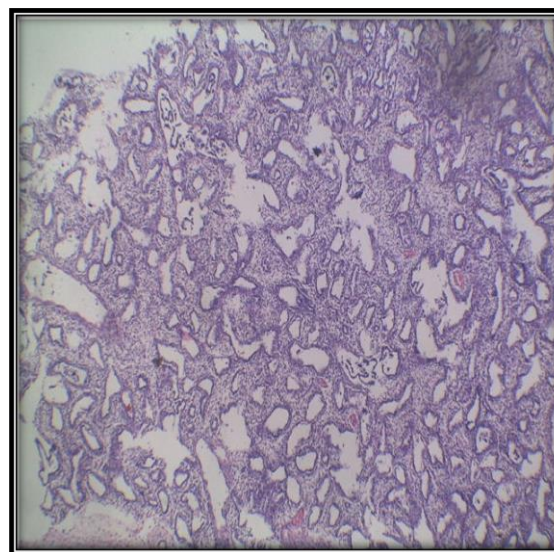


Plate 6: Haematoxylin and eosin-stained photomicrograph (x 100 magnification) showing increase in the number and size of airspaces

The lungs were enlarged, showed surface rib markings (Plate 4) and splinted the diaphragm with the central placement of the heart. The oesophagus was patent and no fistulous connection with trachea was seen. The distal end of the trachea was stenosed (Plate 5) and lacked the usual cartilaginous rings in this segment, making it difficult to probe through the bifurcation into the bronchi. Photomicrograph showed an increase in the number and size of airspaces (Plate 6). These findings were consistent with high airway obstruction secondary to tracheal stenosis with non-immune hydrops fetalis.

DISCUSSION

Congenital high airway obstruction was first reported in 1989 by Arizawa et al. [8]. The term was subsequently coined in 1994 by Hendrick [9]. This rare condition often has a lethal outcome and is mostly caused by laryngeal atresia [9]. Other causes include subglottic stenosis, laryngeal masses (tumours and cysts), tracheal atresia or stenosis (as observed in the index case), and tracheal or laryngeal webs [2]. Due to the upper airway obstruction, there is compensatory hyperplasia of the lung tissues with resultant overgrowth of the lungs [2]. Prenatal ultrasound plays a vital role in the diagnosis of CHAOS. The constellation of features are enlarged hyperechogenic lungs, inverted diaphragms, compression and midline displacement of the heart, and fetal ascites [1-5,9] similar to the features observed in our index case and hitherto reported represent the commonest form of the disease [1,2,10]. Liquor volume abnormalities may also occur depending on the gestational age and change in the physiological mechanism of fetal swallowing. The index case had oligohydramnios. Although polyhydramnios is commonly seen, later in the pregnancy, because there is compression of the oesophagus and thus reduced fetal swallowing, some researchers have reported oligohydramnios [11]. The compensatory hyperplasia of lung tissue is responsible for the sonographic lung appearance. The enlarged lung may also compress the superior and inferior vena cavae, compromising venous return and resulting in fetal ascites and hydrops fetalis [2,10]. Sonographically, it is difficult to accurately predict or detect the exact cause, however, Kalache et al. [10] demonstrated that the use of Color Doppler during respiratory movement may be helpful as a dilated trachea will not demonstrate flow on Doppler. It is known that fetal magnetic resonance imaging provides a better delineation of the location, level, and length of obstruction [11]: this was not done, however, as no fetal surgical intervention was planned.

The CHAOS may be associated with genetic disorders, which usually indicate a more severe form of the disease and all cases, therefore, require detailed evaluation because of the possibility of inheritance in future pregnancies. Common syndromes include Fraser syndrome, Chromosome 5p deletion (cri-du-chat syndrome), velocardiofacial syndrome, short rib polydactyly, and

partial trisomy 5 and Chromosome 16q [12]. The index case had no additional gross anomalies such as urogenital defects, syndactyly, or cryptophthalmos, thus excluding a syndromic association. The genetic karyotype, however, could not be determined due to restrictions created by the COVID-19 pandemic. Often, a CHAOS must be differentiated from other extrinsic causes of tracheal or laryngeal obstruction. These were ruled out during the autopsy of the index case. A variant of CHAOS has been reported in which there is some decompression of the respiratory tract and resolution of the underlying pathology due to a minor pharyngo- or laryngo-tracheal communication [2,13,14]. This, however, does not equal resolution of the underlying pathology. Majority of reported cases have a total or near-total obstruction that often leads to fatal outcomes, which usually occur 20 - 23 wk, which is usually when the pregnancies are electively terminated [9,10]. The prognosis for CHAOS depends on the gestational age, presence or absence of hydrops, location of the blockage, type of malformation causing the airway obstruction, presence of other birth defects [15], and availability of surgical personnel with requisite skills. The index case was diagnosed at 21 wk of gestation and rapidly progressed to hydrops in 1 wk. In the setting where antenatal fetal surgery is unavailable, the possibility or risk for progression to intrauterine fetal death or death shortly after delivery is high. Ex utero intrapartum treatment has been successful for cases with a laryngeal obstruction that survive till the third trimester. The procedure is often carried out at 35 wk during the caesarean section when the fetal head and upper chest are delivered, and the baby is on placental support. Laryngoscopy is performed to confirm the diagnosis, followed by tracheostomy, surfactant administration and mechanical ventilation [11-13,15,16]. Although CHAOS is rare, it is not equivalent to fetal death. Early antenatal sonographic diagnosis coupled with multidisciplinary team involvement and appropriate fetal or perinatal interventions can lead to a good overall outcome.

Conclusion

In conclusion, congenital high airway obstruction syndrome is a rare cause of upper airway obstruction which is not compatible with life in the absence of perinatal intervention. Antenatal sonographic imaging has characteristic findings that can help in establishing an accurate early diagnosis. Magnetic resonance imaging is used as an adjunct diagnostic tool, particularly if fetal intervention is planned. In the absence of requisite surgical skills and in a resource-limited environment, counselling and medical termination will save the couple the emotional trauma of carrying the pregnancy to term, ending up in stillbirth.

DECLARATIONS

Ethical considerations

Informed consent was obtained from the patient for this case report. They also consented to the postmortem. The case

does not contain any information that could lead to the traceability of the patient. The study images were used with consent from RAAJ Specialist Scan Ghana Limited, Cape Coast, Ghana.

Consent to publish

All authors consented to the publication of the manuscript.

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None

Competing Interests

No conflict of interest was reported by the authors.

Author contributions

BJJ conceived the topic, designed the protocol, performed the ultrasound scan, drafted, and edited the manuscript. TAM and EGT counselled, managed the patient, drafted, reviewed, and edited the manuscript. KUA performed the autopsy, reviewed, and edited the manuscript. BDS and DA drafted, reviewed, and edited the manuscript. All authors read and approved the final version of the manuscript.

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Availability of data

All relevant data are provided in the manuscript. The published data is available from the corresponding author on a reasonable request.

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