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Prenatal detection of congenital high airway obstruction syndrome with encephalocele

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Abstract

Congenital high airway obstruction syndrome (CHAOS) causes secondary morphological changes which can be detected on ultrasound. Here we report a case of congenital high airway obstruction with an occipital encephalocele detected at 23 weeks of gestation.

Key words: Congenital high airway obstruction syndrome; dilated airway; encephalocele

Introduction

Congenital high airway obstruction syndrome (CHAOS) is defined as complete or partial obstruction of the fetal upper airways. The cause may be laryngeal atresia, tracheal atresia, or laryngeal cyst, but the clinical presentation is the same. Due to advances in antenatal imaging, more cases are being detected in the prenatal period. CHAOS can occur in isolation or can be syndromic. The association of CHAOS with encephalocele and limb anomalies has been reported in two cases.[1] Both these cases had an anterior encephalocele. Here we report a case of CHAOS with occipital encephalocele.

Case Report

A 23-year-old primigravida was referred to us for evaluation of fetal anomaly, occipital encephalocele, at 23 weeks of gestation. It was a spontaneous conception. She was married for 6 months, it was a non-consanguineous marriage. USG done showed a single fetus with parameters corresponding to 22 weeks of gestation with an occipital encephalocele [Figure 1]. The lungs were enlarged and echogenic with everted diaphragm [Figure 2]. High airways were dilated and the division of the trachea was seen [Figure 3]. The heart was centrally placed and compressed [Figure 4]. There was cutaneous edema and hydramnios. The stomach shadow was not visualized. The fetal karyotype was normal. The couple was counseled regarding the poor prognosis.

She had an intrauterine fetal demise at 24 weeks. Labor was induced and she delivered a dead male fetus weighing 770g. There was generalized soft tissue edema. There was no external malformation other than the encephalocele. The couple refused autopsy.

Discussion

Congenital high airway obstruction is a rare anomaly. The most common cause is laryngeal atresia.[2] Antenatal diagnosis is possible because of the typical morphological changes that occur secondary to the airway obstruction. Lungs normally secrete fluid which flow through the fetal airway and become a component of the amniotic fluid. In cases of congenital airway

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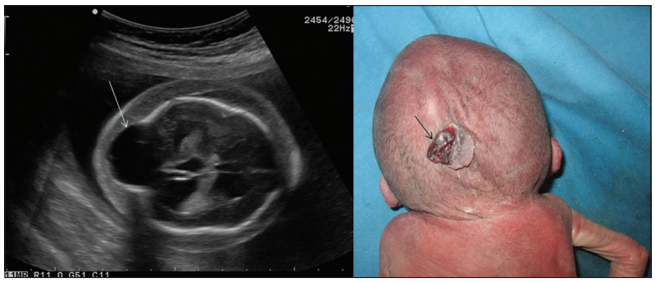

Figure 1: Occipital encephalocele (arrow)

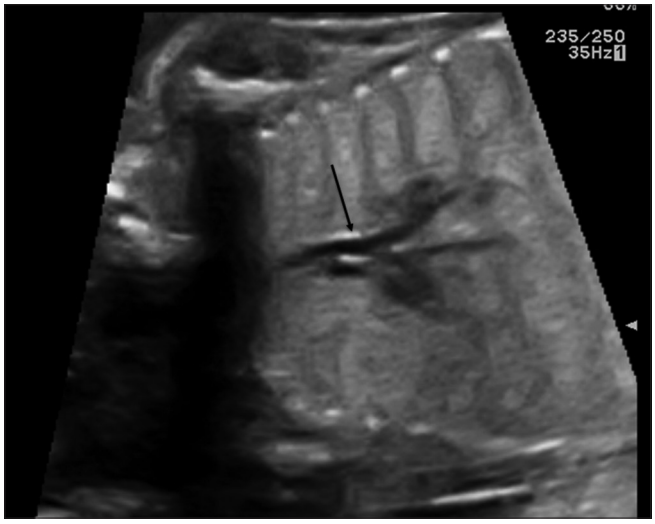

Figure 3: Dilated airway (arrow)

obstruction, the fluid accumulates within the lungs and the airways. This fluid retention results in the clinical entity termed CHAOS. Sonographic features are characteristic and these include bilateral enlarged echogenic lungs, flattened or everted diaphragm, dilated airway distal to the obstruction, small compressed heart, fetal ascites, and/or non immune hydrops. The enlarged lungs cause cardiac and caval compression and this can lead on to hydrops. Esophageal compression can impair fetal swallowing and this, in turn, can cause polyhydramnios.[2-5]

When CHAOS is detected on prenatal ultrasound, a detailed evaluation of the fetus is warranted to exclude other associated anomalies. CHAOS may be seen in Fraser syndrome. The other reported associated anomalies include short long bones, facial cleft, flexion deformities, radial and tibial aplasia, renal agenesis, esophageal atresia, microphthalmia, and single umbilical artery.[4,6,7]

In the past, CHAOS was considered to be a lethal anomaly, but of late, survival in fetuses with CHAOS has been reported with EXIT procedure and postnatal correction.[3] Kohl *et al*. have reported in utero decompression in a fetus with CHAOS.[7]

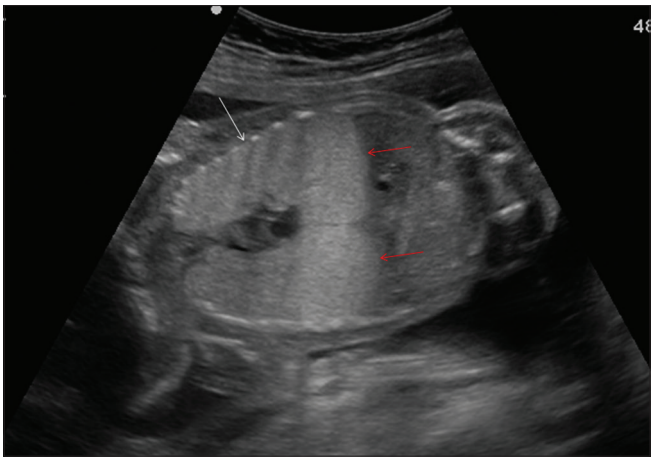

Figure 2: Bilateral echogenic lungs (white arrow) with everted diaphragm (red arrow)

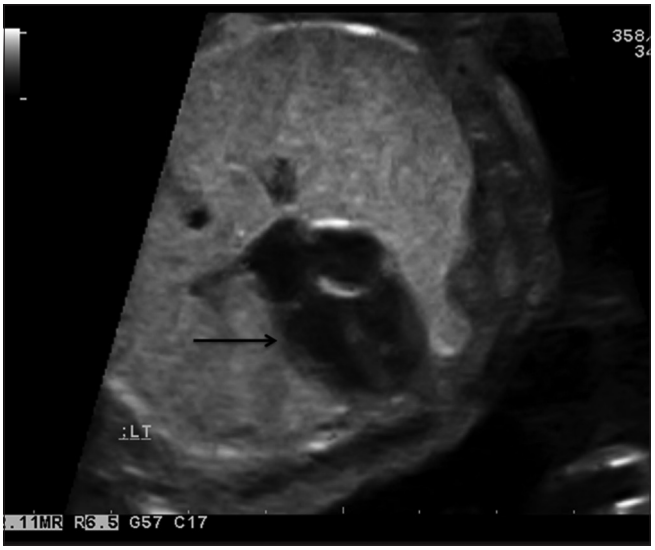

Figure 4: Compressed centrally placed heart (arrow)

The differential diagnosis of echogenic lung is congenital cystic adenomatoid malformation (CCAM). In CHAOS, both lungs are echogenic, whereas CCAM is usually unilateral, but rarely can be bilateral. Dilated airway is another differentiating feature and is seen in CHAOS.[8]

Kalache *et al.* have described the association of laryngeal atresia, encephalocele, and limb anomaly as a possible syndrome -the LEL syndrome.[9] Our case had CHAOS with encephalocele, the limbs were normal. The association of CHAOS with encephalocele may not be coincidental, but whether these features are genetically inherited will remain unclear until further cases are described.

Conclusion

To conclude we have described a case of CHAOS with an encephalocele detected in utero.

CHAOS has characteristic sonographic features, can be detected in the prenatal period and is associated with a poor prognosis

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