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Case Series

Prenatal diagnosis of congenital high airway obstruction syndrome: our experience from a tertiary care center

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ABSTRACT

Congenital high airway obstruction syndrome (CHAOS) is an extremely rare and life-threatening condition. It occurs due to obstruction in fetal respiratory tract and is characterized by typical ultrasonographic findings. Risk of recurrence is low, so antenatal diagnosis can help in counselling regarding risk of recurrence. A retrospective record review of all cases referred to our institution for antenatal ultrasound over a period of 5 years from January, 2014 to December, 2018 was done. Cases diagnosed as CHAOS were reviewed in detail regarding the radiological findings, information regarding delivery, fetal karyotype and postnatal/ fetal examination. Between the period of 2014 to 2018 three fetuses with CHAOS were identified. All of them had characteristic radiological features. Two of them were associated with hydrops and one fetus had oligohydramnios. All the pregnancies were terminated after antenatal diagnosis. Amniocentesis was done in 2 out of 3 cases and fetal karyotype was found to be normal. Fetal autopsy was done in one case and site of upper airway obstruction was identified. Confirmation of diagnosis by antenatal ultrasound and if possible, by post-mortem examination is essential for providing estimation of risk of recurrence to the family and genetic counselling.

Keywords: Congenital high airway obstruction syndrome, Echogenic lungs, Hydrops, Prenatal diagnosis

INTRODUCTION

Congenital high airway obstruction syndrome (CHAOS) is an extremely rare and severe life-threatening condition. The term was introduced by Hedrick and colleagues in 1994. It arises due to obstruction in the upper respiratory tract. The obstruction can occur either at the level of larynx or at trachea.¹ The main ultrasonographic findings are enlarged and hyper-echoic lungs, inverted diaphragm, elongated and compressed heart, dilated tracheobronchial tree and ascites which may progress to hydrops. Without antenatal or postnatal treatment, the condition is universally fatal. The risk of recurrence in subsequent pregnancies is low, so antenatal diagnosis can help in counselling the family. Authors describe here, their experience of three cases of CHAOS diagnosed over a period of 5 years.

CASE SERIES

A retrospective record review of all cases referred to our institution for antenatal ultrasound over a period of 5 years from January, 2014 to December, 2018 was done and three fetuses with CHAOS were identified. All of them had characteristic radiological features suggestive of CHAOS.

Case 1

A 28 years old, fifth gravida was referred at 19 weeks with multiple congenital anomalies and hydrops. On ultrasound (Figure 1), hydrops was present (increased nuchal fold thickness, subcutaneous edema and ascites). Heart was narrowed and compressed between enlarged and hyperechoic lungs. Trachea and bronchi were found

to be distended. The diagnosis of CHAOS was made and in view of presence of hydrops, the chance of poor fetal outcome was explained to the family and they opted for termination of pregnancy. Fetal karyotype was found to be normal. On autopsy evaluation (Figure 1), agenesis of the larynx and superior part of trachea was noted along with hyperinflated lungs.

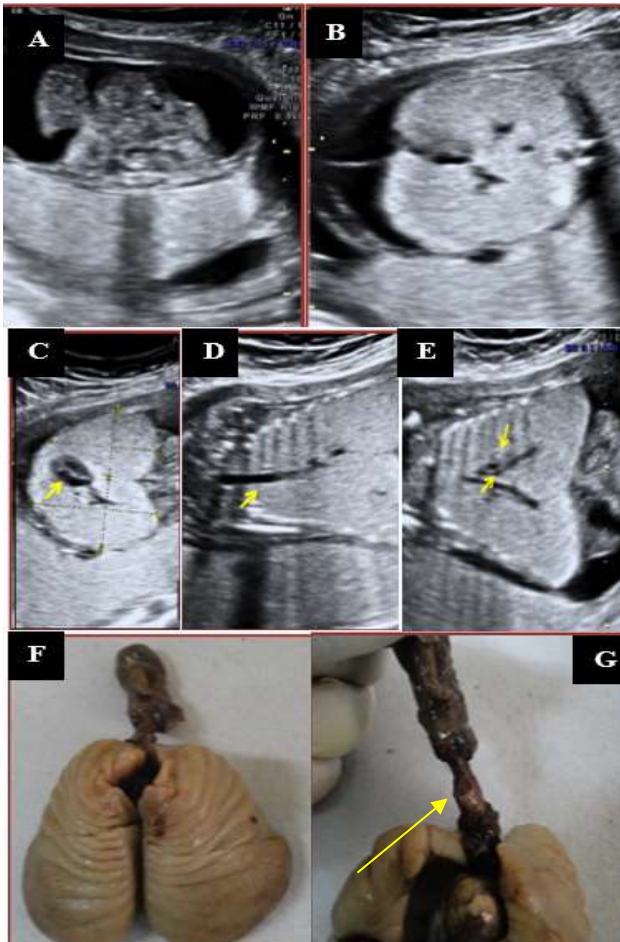


Figure 1: Ultrasound features of fetus 1. Fetus 1 (A and B) ascites and pleural effusion, (C) Transverse scan of elongated and compressed heart (arrow), (D and E) Coronal section of dilated trachea and bronchi (arrow). Fetal autopsy findings in fetus 1 (F) Surface marks of ribs on hyperinflated, enlarged lungs and (G) Agenesis of the larynx and superior part of trachea, inferior part of trachea (arrow) visualized starting from a blind sac superiorly.

Case 2

A 22 years old primigravida was referred at 19 weeks with hydrops and mild cardiac hypoplasia. On ultrasound (Figure 2), heart was found to be compressed between enlarged and hyperechogenic lungs. There was pleural effusion, ascites, scalp and subcutaneous edema. Amniotic fluid karyotype was found to be normal. The

family opted for termination of pregnancy, but the fetus was not brought for postnatal evaluation.

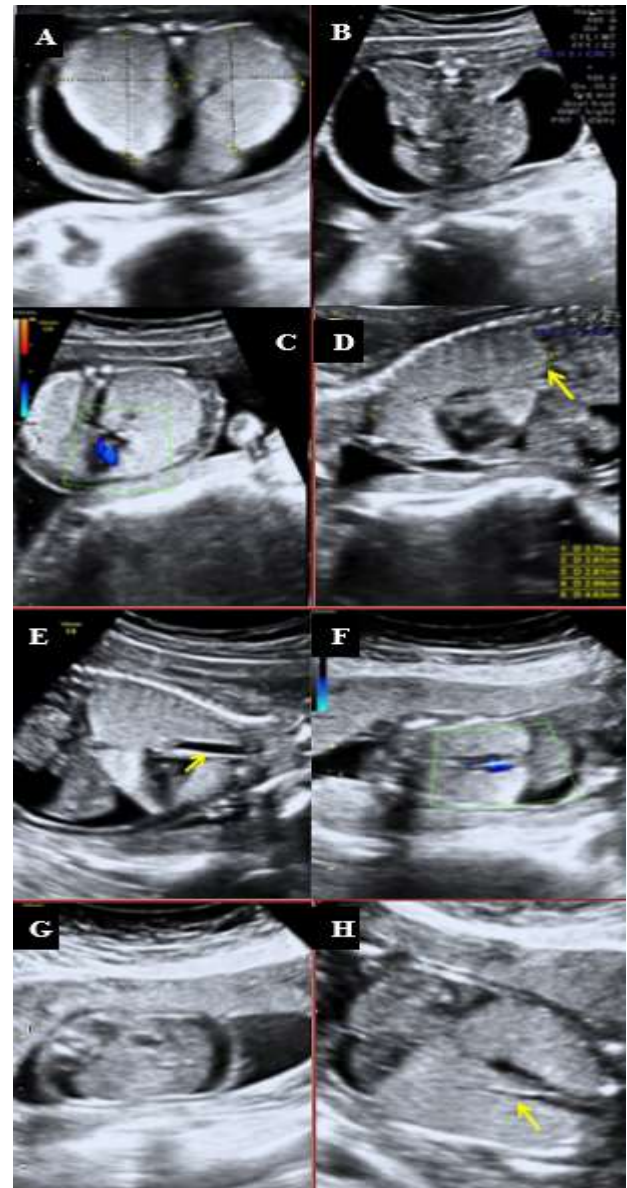


Figure 2: Ultrasound features of fetus 2. Transverse scan of fetus 2 (A) presence of pleural effusion with enlarged, hyperechogenic lungs (B) ascites and (C) compressed heart between enlarged, echogenic lungs (arrow), sagittal scan (D) inverted diaphragm and (E) dilated trachea. Transverse scan of fetus 3 (F) elongated and compressed heart, (G) presence of ascites and (H) enlarged, hyperechogenic lungs with dilated bronchi (arrow).

Case 3

A 30 years old eighth gravida, with history of five pregnancy losses was referred at 18 weeks 4 days for fetal ascites. On ultrasound (Figure 2), amniotic fluid was found to be decreased. Fetal heart was compressed between enlarged and hyperechogenic lungs. Trachea and

bronchus were found to be distended. Fetal ascites was found to be present. The prognosis of the condition was explained to the family. However, the couple didn't opt for fetal karyotyping.

DISCUSSION

CHAOS arises due to obstruction in the upper respiratory tract, most commonly laryngeal atresia which prevents normal egress of fluid secreted by the lung tissue through the larynx.² The fluid accumulates in the tracheobronchial tree resulting in its dilatation and giving bilateral lungs a hyperechogenic appearance on ultrasound. It also causes increased proliferation of the lung tissue and thereby compression of heart in - between the enlarged lungs and inversion of the diaphragm. The elevated intra-thoracic pressure decreases the venous return causing fetal cardiac failure which results in ascites and further hydrops.³ The diagnosis can be made on antenatal ultrasound. All three cases described here were diagnosed on the basis of typical ultrasonographic findings. The characteristic findings are enlarged and hyperechogenic lungs, dilated tracheobronchial tree, inverted or flattened diaphragm, compressed and elongated heart in the midline, fetal ascites and in severe cases features of hydrops. The presence of associated anomalies or hydrops suggests poor prognosis for survival. In two out of three cases, hydrops was present at the time of diagnosis. On providing information about poor outcome, they had opted for termination of pregnancy. The findings were confirmed by fetal autopsy examination in one case and the site of airway obstruction was identified. Antenatal magnetic resonance imaging (MRI) can be done for confirmation of diagnosis and to locate exact site of obstruction, however a study by Mong et al showed 90% concordance rate between detailed ultrasound and MRI evaluation.⁴

The condition is usually sporadic and the risk of recurrence is low. The exact etiology is not known. However, this condition has been found to be associated with various other genetic disorders including some chromosomal abnormalities such as velocardiofacial syndrome, cri-du chat syndrome, trisomy 18, partial trisomy 9, partial trisomy 16q and 47, XXX. It is also seen in some monogenic conditions such as Fraser syndrome, short rib polydactyly, Shprintzen- Goldberg syndrome etc.⁵⁻⁷ In a case series by Jessica L et al. 4 out of 12 fetuses were found to have associated anomalies.²

Also, some conditions need to be considered in differential diagnosis during antenatal ultrasound. One of them is congenital cystic adenomatoid malformation type III (CCAM) of lungs, which is usually unilateral and is associated with presence of normal lung tissue. Whereas, in cases of CHAOS, dilatation of airways distal to the site of obstruction can be seen clearly.⁸ It also needs to be differentiated from some cases of hyperechoic lungs which may be due to transient obstruction to airway due to mucosal plug and later reverts back to normal and

outcome is normal. Severity of findings usually will differentiate.

The prognosis is generally poor without any fetal or postnatal intervention. Fetoscopic management of upper airway obstruction or ex-utero intrapartum treatment (EXIT) has been used in some cases with variable outcome. The outcome is better in cases with no associated anomalies, absence of hydrops and also depend on extent of obstruction.⁹ This emphasizes the need of detailed ultrasound and postnatal evaluation to look for associated anomalies, and also fetal karyotyping for chromosomal abnormalities.¹⁰ It may also help to provide information regarding planning of delivery, fetal or perinatal intervention, mode and place of delivery, and also risk of recurrence in future pregnancies.

CONCLUSION

Detailed evaluation of suspected cases is required to look for associated anomalies considering possibility of associated genetic syndromes. Confirmation of diagnosis by antenatal ultrasound and if possible, by post-mortem examination is essential for providing estimation of risk of recurrence to the family and genetic counselling.

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