Congenital high airways obstruction syndrome – first and second trimester diagnosis

Abstract

Congenital High Airways Obstruction Syndrome (CHAOS) is a rare condition associated with high fetal or neonatal mortality. Prenatal diagnosis in the second trimester is made by the typical ultrasound features of large echogenic lungs, a flattened or inverted diaphragm, and fetal ascites or hydrops. We present two cases diagnosed at our institution; one in the second trimester, and a first trimester diagnosis. To the best of our knowledge, first trimester diagnosis of CHAOS has not been previously reported.

Keywords: laryngeal atresia, prenatal diagnosis, tracheal agenesis.

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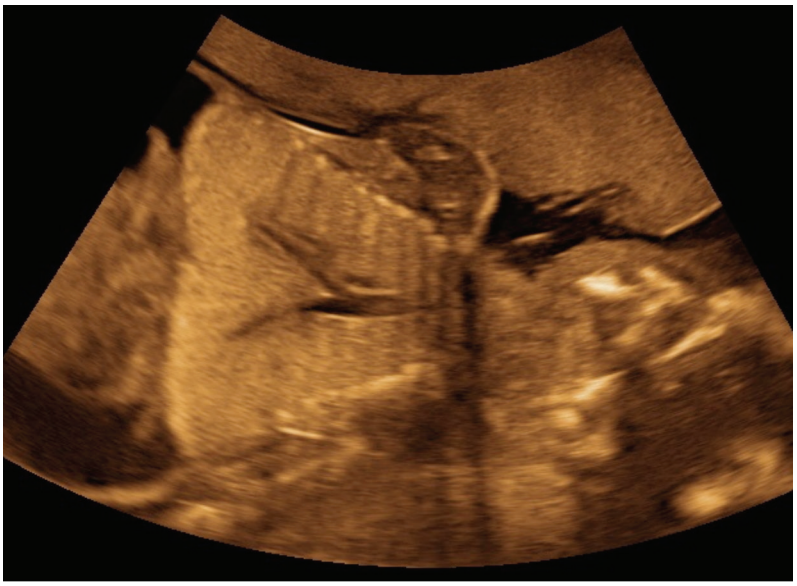

Figure 1: Coronal image through the fetal thorax and upper abdomen. Note the large echogenic lungs with a convex inferior aspect inverting the diaphragm and the dilated bronchial tree.

Case report 1

A 39-year-old G6 P5 was referred to our unit at 21 weeks gestation, following a routine mid-trimester scan demonstrating fetal ascites. Ultrasound findings included large echogenic lungs, dilated bronchi and an inverted diagram (Figure 1). The fetal abdomen was grossly distended due to ascites (Figure 2). The couple elected to terminate the pregnancy. Post mortem confirmed high airways obstruction with subglottic tracheal atresia involving the proximal 25 mm of the trachea (Figure 3). A tracheoesphageal fistula was not demonstrated. The lungs were markedly hyperplastic, with a combined weight of 67 g (normal is about 13 g at 21 w). The diaphragm was inverted and the bronchial tree dilated. Gross ascites and small pleural and pericardial effusions were present.

Case report 2

A 22 year old G2 P1 was referred to our unit at 12 weeks gestation, following a routine 12 week scan which demonstrated an increased nuchal translucency, a posterior fossa cyst, and a hand abnormality. These ultrasound findings were confirmed, along with the additional finding of echogenic lungs (Figure 4). The couple elected to terminate the pregnancy at 13 week gestation in view of the cluster of findings. Post mortem was not able to confirm the posterior fossa abnormality due to maceration; however, the right hand demonstrated a lobster claw deformity. The lung size was in normal limits for gestation and there was disruption of anterior neck structures (Figure 5). In addition, a posterior cleft palate and imperforate nostrils were present.

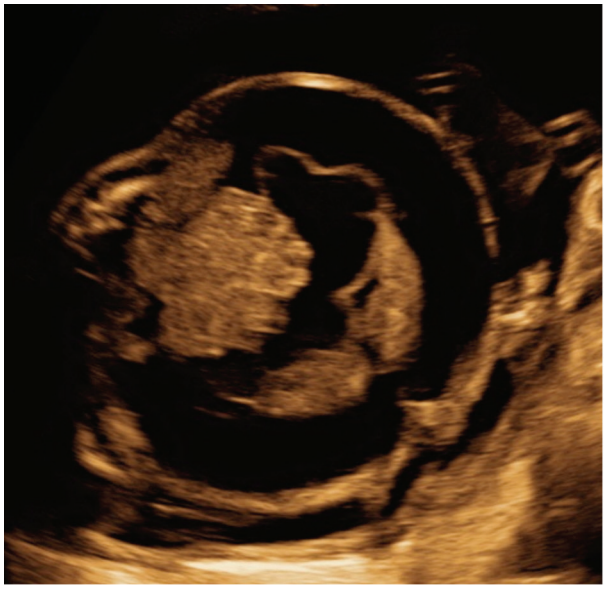

Figure 2: Transverse image through the fetal abdomen demonstrating gross ascites.

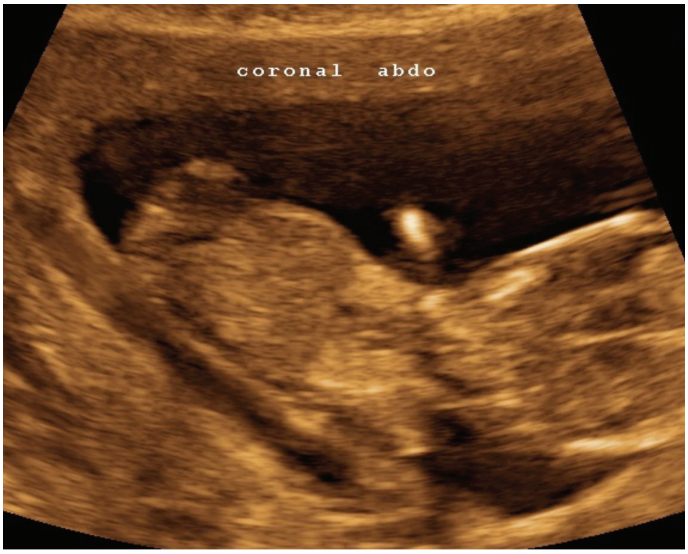

Figure 4: Coronal image through the fetal thorax and abdomen demonstrating echogenic lungs.

Discussion

CHAOS results from complete or near complete obstruction of the fetal airway. This may occur at the larynx (laryngeal atresia), or along the trachea (tracheal agenesis). Tracheal agenesis may be complete or partial, and there may be a tracheoesophageal fistula (TOF).[1]

Floyd, *et al.* (1962) described three classifications of tracheal agenesis.

- Type 1: agenesis of the proximal trachea with presence of the distal trachea and a TOF.
- Type 2: (most common) agenesis of the entire trachea with normal bronchi fused at the level of the carina. A fistula is often present between the oesophagus and carina.
- Type 3: Complete agenesis of the trachea with main bronchi arising from the oesophagus.[2]

Laryngeal atresia is the most common cause for CHAOS, however, isolated tracheal stenosis or subglottic stenosis may also occur. The mechanism for CHAOS is thought to occur around the 10th week of gestation. At this time, an epithelial lamina temporarily occludes the upper airway of the fetus. Failure of this membrane to properly recanalise results in laryngeal atresia and obstructed airways prevent fetal lung fluid escaping. Increasing intratracheal pressure leads to the distension of the tracheobronchial tree and proliferation of lung tissue. As the lungs expand, the diaphragm flattens and may invert. Cardiac compression and mediastinal shift occurs as the lungs enlarge. Increased intrathoracic pressure causes decreased venous

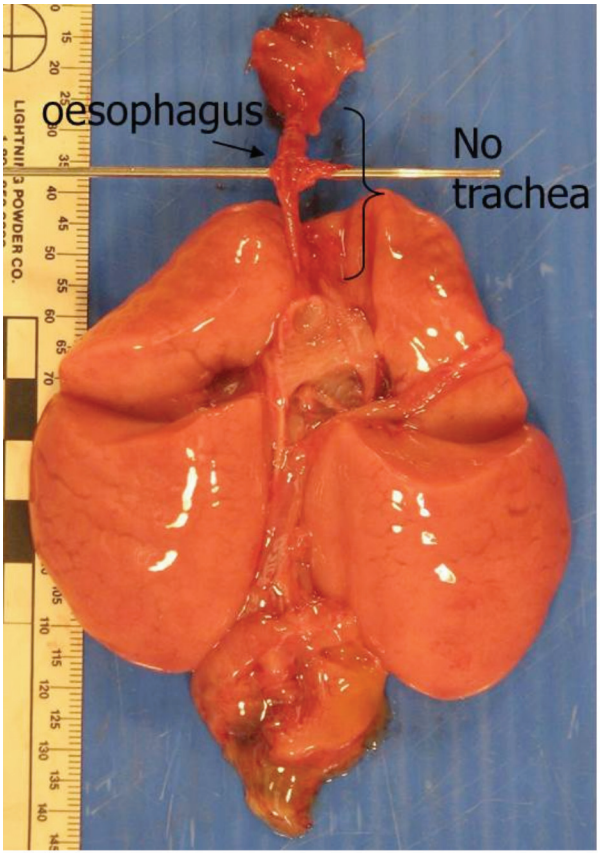

Figure 3: Post mortem dissection demonstrating tracheal atresia and pulmonary hyperplasia. Note the convex inferior aspect of the lungs.

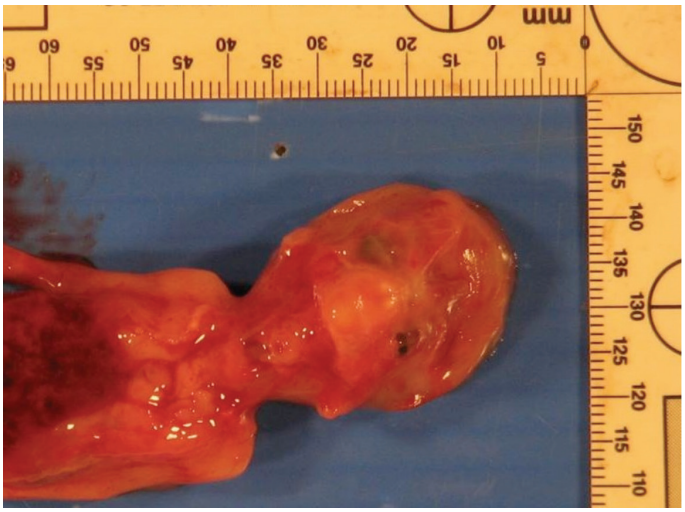

Figure 5: Post mortem dissection demonstrating disruption of anterior neck anatomy.

return, cardiac failure and ascites.[3] This sequence explains the characteristic ultrasound features of large echogenic lungs, a flattened or inverted diaphragm, fluid filled dilated trachea and bronchi, mediastinal shift/compression of the heart, and fetal ascites or hydrops.[1, 3]

When a TOF is present, lung fluid can pass into the stomach or amniotic sac, and lung pressure will remain normal. The ultrasound features of CHAOS will therefore not be present and prenatal diagnosis almost impossible. In a recent literature review, de Groot-van der Mooren (2012) and colleagues reported that in 72% of cases, polyhydramnios was present in fetuses with tracheal atresia (TA) and a TOF, and in more than half of these cases there were no other mechanisms for polyhydramnios. They suggested that the diagnosis of TA should be considered when polyhydramnios develops, particularly in combination with associated anomalies[4].

The presence of other anomalies suggests CHAOS may occur as part of a syndrome or association. Fraser syndrome, an autosomal recessive disorder, is characterised by laryngeal atresia, cryptophlathmos, syndactyly, and urogenital defects.[3] The VACTERL association (vertebral anomalies, anal atresia, cardiac anomalies, tracheoesophageal fistula/oesophageal atresia, renal and limb anomalies)and TACRD association (tracheal atresia/laryngeal atresia, cardiac and renal anomalies, and duodenal atresia) are also considerations when CHAOS is suspected.[3,5]

Therapeutic options, namely ex utero intrapartum treatment (EXIT), may now be possible for presentations of CHAOS where enough proximal or distal trachea is present to allow for a tracheostomy.[5] For these cases, fetal MRI has been shown to be helpful in localising the level of obstruction.[6, 7] Performed during caesarean section, EXIT procedures allow for fetal interventions while maintaining placental circulation, and have been used to re-establish the fetal airway in cases of diaphragmatic hernia. EXIT procedures have been reported in CHAOS with reasonable neonatal survival rates.[3,5,8] These children, however, may remain tracheostomy dependent, require complicated staged airways reconstruction, are delayed in oral feeding and have speech limitations.[3] The ability to provide effective therapy relies on early and accurate prenatal diagnosis.

One interesting development in relation to this defect is the creation and clinical use use of stem cell tissue-engineered airways. The first transplant of this artificially created airway has already taken place successfully.[9] This has significant implications for both the pediatric and adult populations but it is important to remember that early detection of these cases(i.e. pre-viability) puts them most likely in a much poorer prognostic group and termination of pregnancy remains a realistic option in terms of management.

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