Evolution of Congenital High Airway Obstruction Syndrome (CHAOS): A case report

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Introduction

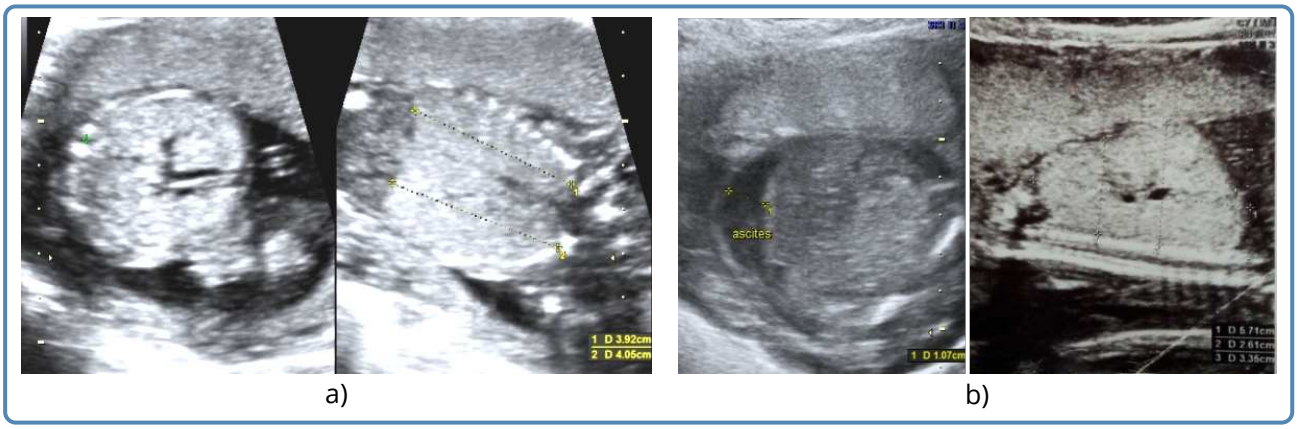
Congenital High Airway Obstruction Syndrome (CHAOS) is a rare, mostly lethal abnormality which is characterized by congenital obstruction of the upper airway usually at the level of the larynx or trachea. This condition is usually diagnosed in utero by ultrasonography which typically shows large echogenic lungs, flattened or inverted diaphragm with or without dilated airways distal to the obstruction, and fetal hydrops [Hoffer et al.,1994]. Reported here is a case of CHAOS with tracheal stenosis which was detected in antenatal USG at 16 weeks and which evolved into fetal hydrops at 18 weeks.

Case Report

A primigravida with nonconsanguineous marriage was referred at 17 weeks 6 days (by dates) of gestation for a detailed fetal anomaly scan in view of findings of oligohydramnios with hyper inflated, enlarged and highly echogenic lungs with inversion of diaphragm in an ultrasound scan done elsewhere. She had been diagnosed to have hypothyroidism 3 years back and was on thyroxin replacement therapy for the same. There was no history of fever with rash, radiation exposure or teratogenic drug intake in the antenatal period. Antenatal USG done at our hospital, showed presence of a single live fetus with a mean gestational age (GA) of 16 weeks 4 days with bilateral homogenously hyperechoic and enlarged lungs, inverted diaphragm, compressed and narrow four chamber view of the heart and non visualization of the stomach suggestive of the possibility of congenital high airway obstruction syndrome (CHAOS) (Figure 1a). The lungs were enlarged and pressing the diaphragm leading to convex lower border of the lungs. The family was counseled regarding the poor prognosis of the condition. In the follow up USG after two weeks, the fetal growth parameters corresponded to a gestational age of 18 weeks 4 days and the fetus had similar findings as earlier along with ascites (Figure 1b). The lung length had increased from 3.92 cm to 5.71 cm. Amniocentesis was done and karyotype of cultured amniocytes was found to be normal. The couple opted for termination of the pregnancy and the fetus was brought for autopsy evaluation.

On external examination of the fetus the chest was small and the abdomen was distended (Figure 2a). On internal examination ascites was present. Both lungs were enlarged with a smooth surface. The diaphragm was thinned out and the liver & other abdominal viscera were pushed down by enlarged lungs (Figure 2b). An elongated part of the trachea below the larynx was narrowed suggestive of lower tracheal stenosis proximal to the bifurcation. There was no tracheoesophageal communication (Figure 2c, 2d). There were no other malformations.

Histopathological examination showed stenosed area of trachea measuring 0.5 cm in length. The stenosed region was 2.5 cm from the upper end of the trachea and 0.5 cm from the lower end. Sections from this area showed tracheal canal lined by pseudo stratified ciliated columnar epithelium with an underlying zone of hemorrhage. Surrounding areas showed cartilage, thyroidal tissue and congested vessels (Figure 2e). After autopsy and histopathological examination, a final diagnosis of CHAOS with tracheal stenosis



a) b)

Figure 1 Showing images of antenatal ultrasound done at 16 weeks (fig 1a) and 18 weeks (fig 1b) gestation. Note the hyperechoic lungs, convex lower border and compressed heart. Ascites is seen in fig 1b.

with fetal ascites was made.

Discussion

We describe a case of CHAOS diagnosed in the antenatal period at 16 weeks of gestational age. After diagnosis at 16 weeks, the fetus was followed up at 18 weeks by which time fetal ascites also developed. The evolution into fetal hydrops helped the family in taking the decision of termination of pregnancy. CHAOS is a rare lethal malformation and to the best of our knowledge, no similar case has been reported earlier where features of CHAOS were evident as early as 16 weeks and its evolutionary trend followed. Findings were confirmed by fetal autopsy which showed the presence of tracheal stenosis. In CHAOS, obstruction of larynx or trachea results in retention of bronchial secretions and pulmonary distension. Overdistension of the lung with flattening or inversion of the diaphragm cause compression of the heart and inferior vena cava, impairing venous return thereby resulting in fetal hydrops and ascites, as was seen in our case at 18 weeks of gestational age (Figure 1a,1b) [Manjula et al., 2014]. The most frequent cause of CHAOS is laryngeal atresia but other etiologies include laryngeal or tracheal webs, laryngeal cysts, tracheal atresia, subglottic stenosis or atresia, and laryngeal or tracheal agenesis. Neck masses can also cause compression and narrowing of lumen.

The overall incidence of congenital tracheal stenosis is not clear. In 1994, Hoffer et al. described fewer than 70 reported cases of varying grades of congenital tracheal stenosis. However the number of cases which presented with CHAOS was not mentioned in the study [Hoffer et al., 1994; Lorri et al., 2006]. Hedrick et al. reported 4 cases of CHAOS and on autopsy 3 fetuses showed laryngeal atresia while one had tracheal stenosis [Hedrick et al., 1994].

Although most cases of CHAOS are sporadic, some cases have been linked to genetic syndromes, the commonest being Fraser syndrome. This comprises of laryngeal or tracheal atresia, cryptophthalmos, microphthalmia, renal agenesis, orofacial clefting, mental retardation, musculoskeletal anomalies and syndactyly or polydactyly [Berg et al., 2001].

Syndromic etiologies like Fryn syndrome need to be considered, as being inherited in an autosomal recessive manner; they could have a risk of recurrence of 25%. Other syndromes which have been reported in association with CHAOS are Cri-du-chat syndrome, short-rib polydactyly syndrome, and velo-cardio-facial syndrome [Joshi P et al., 2012]. Few familial cases have been reported in published literature. A case of CHAOS with autosomal dominant inheritance of the father and his two affected children was reported by Vanhaesebrouck et al. [2006]. In our case the family was counseled that the risk of recurrence is not significantly increased. However, detailed and serial ultrasonographic evaluation of the next pregnancy, starting from 14 to 16 weeks of gestation is indicated.

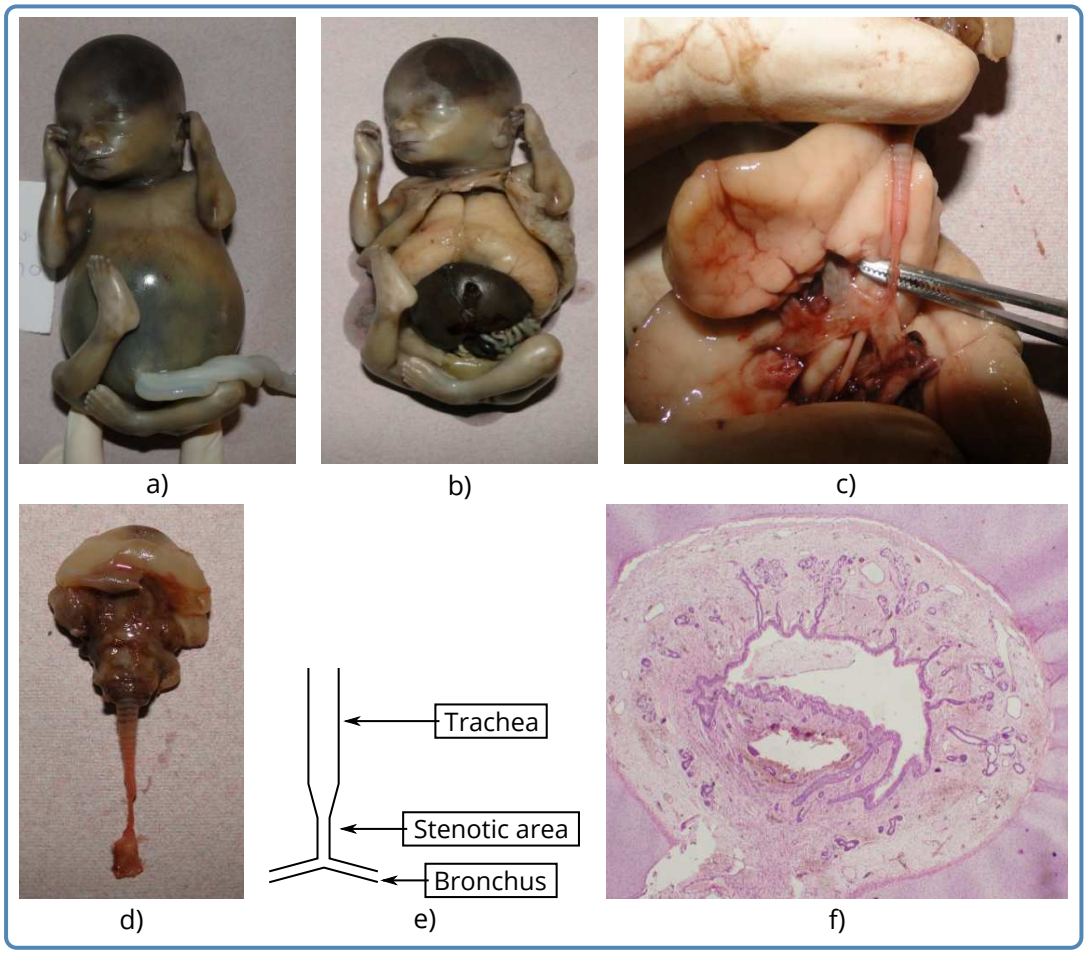


Figure 2 Showing autopsy images (2a, 2b, 2c, 2d) and diagrammatic representation of tracheal stenosis (2e). Figure 2f showing stained sections (X 40) from the tracheal stenotic area which shows the tracheal canal lined by pseudostratified ciliated columnar epithelium of mucosa with an underlying zone of submucosa, a fibrocartilage layer and an adventitia.

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