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Learning from errors

Congenital laryngomucocoele: a rare cause for CHAOS

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Abstract

Congenital high airway obstruction syndrome (CHAOS) is a rare but life-threatening condition that results from the obstruction of the upper airways.

We describe a female newborn, from a Grávida II, Para 0, 36-year-old woman, with a routine ultrasound at 30 weeks' gestation that showed polyhydramnios. She delivered a live-born female baby at 36 weeks without any dismorphic features but with respiratory distress. Attempts at endotracheal intubation were unsuccessful due to the presence of a mass obstructing the larynx. The reanimation process was stopped after 20 minutes. Post-mortem examination demonstrated the presence of a total occlusion of the larynx by a laryngomucocoele. Laryngocele, a congenital cyst of the larynx, occurs rarely and hardly ever as a cause of CHAOS. What is more, laryngomucocoele has not been previously reported as a cause of CHAOS. These conditions represent a neonatal emergency with reserved prognosis unless diagnosed antenatally allowing for a programmed ex utero intrapartum treatment (EXIT) by performing tracheostomy while maintaining the placental circulation.

BACKGROUND

Congenital high airways obstruction syndrome (CHAOS) is characterised by a set of echographic findings related to the complete or almost complete obstruction of the upper airways resulting in large echogenic lungs, flattened or inverted hemidiaphragms, widening of the airways distal to the obstruction, fetal ascites and hydrops.[1] The majority of the cases are sporadic; the incidence unknown.[2] Laryngeal atresia is the most frequent cause, followed by tracheal atresia or stenosis and laryngeal membranes or septs.[1]

Prenatal diagnosis is mandatory to allow programmed delivery with assistance of a multidisciplinary team in order to re-establish a patent airway before placentary circulation is interrupted.[3]

CASE PRESENTATION

Mother, 33 years old, white, hypertensive, clerical worker; father, 39 years old, smoker and heavy drinker, furniture worker; both Ukrainian emigrants. Uneventful pregnancy; serology negative (venereal diseases research laboratory, hepatitis B surface antigen (HBsAg), toxoplasmosis, HIV 1 and 2); blood group O Rh negative. Single routine obstetric echography at 30 weeks' gestation; head presentation, biparietal diameter, abdominal area and femur within the 50th percentile. Increased amniotic fluid.

Live-born female weighing 2230 g, delivery by venteuse at 36 weeks' gestation without dismorphic signs or external malformations. Hypotonic newborn, pink-coloured, fetal pulse 100 pm, not crying and showing inefficient respiratory efforts.

Several unsuccessful intubation attempts due to the presence of a whitish mass the size of a "table-tennis ball"; the glottis and epiglottis were not visualised and the only visible orifice was posterior and corresponded to the oesophagus. Progressive deterioration with Apgar score 6/1/1 at 1st, 5th and 10th minutes. The resuscitation procedure was abandoned after 20 minutes due to lack of vital signs.

INVESTIGATIONS

Post-mortem examination showed a female newborn with general development according to gestational age. Gross examination revealed a laryngeal cyst 3 cm in diameter arising from the posterior surface of the epiglottis concerning the larynx with a near total occlusion of the laryngeal orifice (fig 1A). This cyst extended below the posterior surface of the epiglottis and the anterior border of the larynx and had a mucinous content. The subglotic trachea was almost completely obstructed (fig 1B). Histological examination confirmed the presence of a laryngomucocoele.

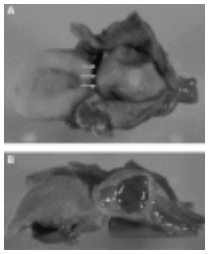

Figure 1

A. Cystic lesion of the larynx, 3 cm in diameter, arising from the posterior epiglottis and obstructing the laryngeal aperture (white arrows). B. Laryngomucocoele, 3 cm maximum diameter, concerning the posterior aspect of the epiglottis and proximal larynx (more ...)

On histology, the lungs showed overall architecture in keeping with the gestational age but there was marked dilatation of the distal airways, including terminal bronchioalveolar structures (fig 2).

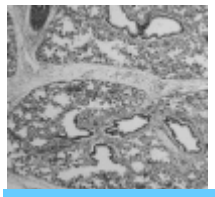

Figure 2
Lung histology showing a marked dilatation of the distal airways, including terminal bronchioalveolar structures (H&E, ×40).

DISCUSSION

Congenital laryngocoele is a rarely reported condition most frequently located in the supraglottic region.[4] In addition, there are but occasional reports of congenital laryngeal cysts related to CHAOS-like presentations in newborns.[4,5] To the best of our knowledge there is no known report of congenital laryngomucocoele as a cause of CHAOS. Lim *et al*[6] included a case of laryngomucocoele in a fetal termination at 22 weeks' gestation when discussing the natural history and management of CHAOS.

Our case showed a polyhydramnios on antenatal ultrasound, which is part of the sonographic triad for diagnosis of CHAOS: large echogenic lungs, flattened or inverted diaphragm, and fetal ascites and/or hydrops. Post-mortem examination allowed for the diagnosis of a laryngomucocoele as the cause of this upper airway obstruction and the fatal outcome is a frequent event in undiagnosed CHAOS. The absence of the other echographical parameters of the triad may be explained if it is assumed that the obstruction at 30 weeks was only partial.[2,7] Oesophageal compression by dilated airways and increased lung volume are probably the cause of polyhydramnios due to decreased ingestion of amniotic fluid.[1,6]

Our report further underlines the need for a specific prenatal echographic assessment of the larynx in order to detect congenital alterations of the upper airways when confronted with the presence of isolated ultrasound findings as polyhydramnios or any of the other elements of CHAOS. The absence of prenatal diagnosis is fatal in the majority of the cases.[7,8]

It is possible to perform echographic assessment of the laryngeal cartilaginous structures after 22 weeks.[9] In some cases, MRI may characterise the lesions better[6] and colour/spectral Doppler may be helpful in the differential diagnosis of foetuses with CHAOS.[10] In the presence of hyperechogenic lungs, it is necessary to consider as differential diagnosis the bilateral cystic adenomatoid malformation.[2]

The majority of typical cases of CHAOS with complete obstruction of the airways are sporadic and of unknown incidence. The study of abnormalities in the context of a familial history may demonstrate autosomic dominant transmission with varying expression or an association with Fraser syndrome, polydactyl short ribs syndrome and

chromosomal alterations such as deletion of chromosome 5 (“Cri du Chat” syndrome) and microdeletion of the long arm of chromosome 22 (velocardiofacial syndrome).[2]

The most common causes of CHAOS are laryngeal atresia, subglotic stenosis and laryngeal membrane. Laryngomucocoele is a rare cause of airways obstruction in the neonatal period.[4–6] A literature search showed only one reference to a single case of a termination at 22 weeks’ gestation with a laryngocele[6] since it is more frequent in the adult.[11–13]

Prenatal diagnosis allows intrauterine treatment for foetuses with CHAOS when at high risk of fetal death[1,8,14] and assists with planning of ex utero intrapartum treatment (EXIT) with tracheostomy while the placental circulation is maintained.[1] This management requires a multidisciplinary team: obstetrician, neonatologist, paediatric otolaryngologist, anaesthetist specialising in neonatology, obstetric/gynaecologist anaesthetist, and so forth. The EXIT procedure starts with assessment of the larynx—that is, with laryngoscopy plus or minus rigid bronchoscopy—in an effort to secure an airway; failing this, a tracheotomy is performed. The placental circulation is maintained, allowing for the clearing of the fetal airway and, thus, affording its patency at birth before the newborn breathing and circulation are established. The main differences between the EXIT procedure and a caesarean section is the uterine hypotonia in EXIT and the use of inhalation anaesthetic gases, contrary to the standard caesarean section where uterine hypertonia is required to prevent post-partum haemorrhage.[3,6,15]

It has been successfully applied in cases of cervical masses, CHAOS syndrome, thoracic masses, unilateral pulmonary agenesis and congenital diaphragmatic hernia.[3,6]

LEARNING POINTS

- Laryngomucocoele has not been previously reported as a cause of congenital high airways obstruction syndrome (CHAOS).
- These conditions represent a neonatal emergency with reserved prognosis unless diagnosed antenatally allowing for a programmed ex-utero intrapartum treatment (EXIT).
- In the centres with experience in this procedure, complications for the mother and the newborn are rare.

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Footnotes

Competing interests: none.

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