Tracheal agenesis: a lethal malformation

Tracheal agenesis is a rare and lethal congenital defect consisting of complete or partial absence of the trachea below the larynx. This report describes a newborn infant with type II tracheal agenesis and a broncho-oesophageal fistula. Presence of the fistula made prenatal diagnosis difficult and allowed the infant to be ventilated via oesophageal intubation with detection of exhaled carbon dioxide.

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Key points

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1. Tracheal agenesis is a rare and lethal congenital anomaly with an incidence of 1 in 50,000 and a male to female ratio of 2:1.
2. It may present postnatally as respiratory distress, no audible cry and failed endotracheal intubation. Most infants will be diagnosed after birth unless congenital high airway obstruction syndrome (CHAOS) is present, which can help in antenatal diagnosis.
3. Presence of polyhydramnios, although not specific for tracheal agenesis, may alert the clinician to potential underlying tracheal problems.

The case report

The neonatal team was alerted at a fetomaternal meeting about a 32-year-old primigravida woman who presented with severe polyhydramnios and ultrasound findings of dilated bowel loops in the fetus, raising the possibility of bowel atresia. Labour began at 34 weeks' gestation and, following a course of steroids, a female infant with a birth weight of 2.1kg was delivered by vaginal route.

The newborn infant had to be intubated at 12-15 minutes of age by the attending medical team because of respiratory distress and her inability to maintain oxygen saturation. Intubation was reported to be uneventful, including a colour change with a colorimetric carbon dioxide detection device (NeoStat, colorimetric capnography). Following admission to the neonatal intensive care unit, the baby received surfactant (Curosurf) and commenced synchronised intermittent mandatory ventilation (SIMV). Subsequently it was noted that the infant had ongoing chest wall recessions, increasing oxygen requirement and persistent flashing alarms on the ventilator denoting air leak and poor blood gases.

The team reviewed for 'DOPE' – **d**isplaced endotracheal tube (ETT), **o**bstruction, **p**neumothorax and **e**quipment failure – none of which were found to be conclusive. Tracheal placement of the ETT was checked again by connecting the colorimetric carbon dioxide detection device. The colour changed to yellow indicating detection of carbon dioxide, which led the medical team to believe that the ETT was correctly placed in the trachea. A chest and abdominal X-ray (**FIGURE 1**) confirmed the ETT to be high: consequently the ETT was adjusted.

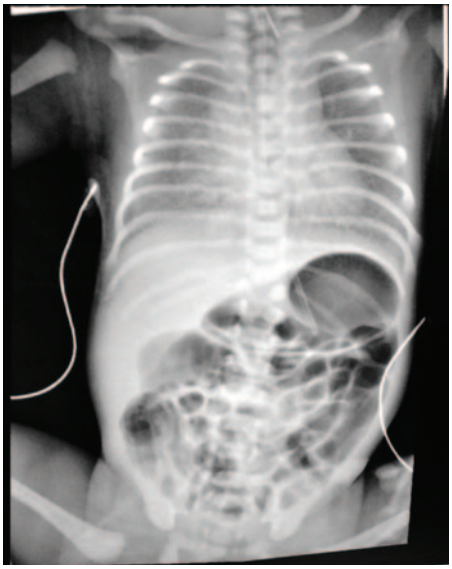

FIGURE 1 A chest and abdominal X-ray showing a high endotracheal tube and dilated bowel loops.

The infant continued to have significant chest wall recessions and difficulty in ventilation despite adequate sedation and optimisation of ventilatory parameters. A decision was taken by the senior doctor to re-intubate the infant. Attempts at intubation were unsuccessful as no laryngeal inlet could be identified, except for a 'blind pit'. The ETT was placed back into the oesophagus and again, exhaled carbon dioxide was detected by colorimetric capnography. It was suspected that, since its first placement, the ETT had been residing in the oesophagus and ventilation was probably occurring via a fistula between the trachea and the oesophagus.

The infant was subsequently transferred to the tertiary centre for investigations and specialist opinion to confirm an anatomical abnormality and to ascertain whether the anomaly might be repairable.

A bronchogram (**FIGURE 2**) and computerised tomography (CT) scans (**FIGURE 3**) confirmed the diagnosis of complete tracheal agenesis with a broncho-oesophageal fistula. An echocardiogram showed a double outlet right ventricle and a ventricular septal defect. The family was made aware that these airway anomalies were non-correctable and incompatible with life. Following discussion with the extended family, care was redirected to palliation and the infant died on the third day.

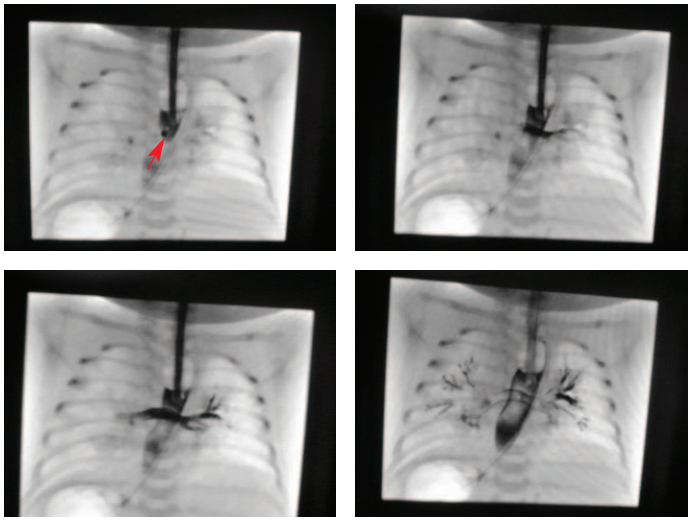

FIGURE 2 Bronchogram sequences showing the fistula connection (arrow) between the carina and the oesophagus (the endotracheal tube is in the oesophagus). Note the unusual horizontal alignment of the right and left main bronchi.

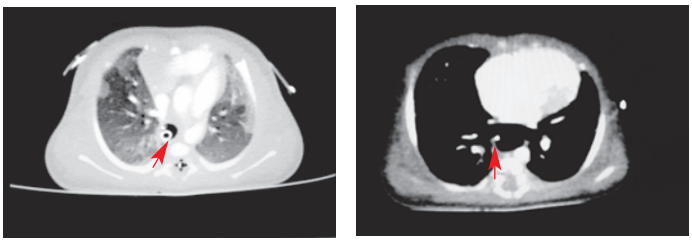

FIGURE 3 CT scans of the chest. Left: the endotracheal tube and the nasogastric tube can be seen in the oesophagus (arrow). Right: the nasogastric tube is seen located in the oesophagus (arrow). No trachea is seen and note the origin of the right and left main bronchi in continuation with the oesophagus.

Review and discussion

Tracheal agenesis is a rare congenital anomaly; a recently published literature review quoted 49 reported cases up until 2010.[1] Floyd et al classified tracheal agenesis into three subtypes (**TABLE 1**).[2]

In the majority of cases, the diagnosis of tracheal agenesis is established after birth; prenatal diagnosis is difficult and prenatal findings leading to the diagnosis of tracheal agenesis are only found when no tracheo-oesophageal fistula (TOF) is present. Absence of a TOF gives a characteristic appearance on ultrasound, consistent with the definition of congenital high airway obstruction syndrome (CHAOS), showing enlarged hyperechogenic lungs and fluid-filled dilated trachea and bronchi, demonstrating an absent flow in the trachea during fetal breathing. Presence of a TOF allows the lung fluid to pass via the fistula to the stomach or amniotic sac thereby preventing the characteristic appearance of CHAOS on ultrasound. In uncertain cases fetal magnetic resonance imaging can provide additional information.

Postnatal diagnosis can be established by the presence of respiratory distress with severe recession, poor air entry, no audible cry and failed endotracheal intubation. Presence of TOF with tracheal agenesis may allow bag and mask ventilation and accidental oesophageal intubation to temporarily improve the respiratory status.

Using Floyd's classification, the case presented in this report may be classified as type II tracheal agenesis. Presence of a broncho-oesophageal fistula in this case made prenatal diagnosis difficult and allowed the baby to be ventilated via an oesophageal intubation with detection of carbon dioxide on colorimetric capnography.

Type I	Type II	Type III
The trachea is absent except for a short caudal segment with a normal carina A tracheo-oesophageal fistula connects the distal trachea with the oesophagus	There is complete tracheal agenesis and both the main bronchi join in the midline at the carina Sometimes a fistulous connection between the oesophagus and the fused bronchi can exist The most common type of tracheal agenesis	The two main bronchi originate independently from the oesophagus

TABLE 1 Floyd's classification of tracheal agenesis.

Conclusion

Tracheal agenesis is a severe congenital defect that can present as an unexpected difficult airway in a newborn infant. Prenatal diagnosis of tracheal agenesis is rare and only possible in cases of CHAOS. The presence of polyhydramnios should alert clinicians not only to problems such as oesophageal atresia, TOF and bowel atresia, but also to tracheal problems. A TOF in tracheal agenesis can allow ventilation via an oesophageal route with detection of exhaled carbon dioxide. Colorimetric capnography should be used as an adjunct in addition to clinical examination to confirm the tracheal placement of the ETT.

Patient consent

The authors received written consent to publish this report from the patient's parents.

References

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