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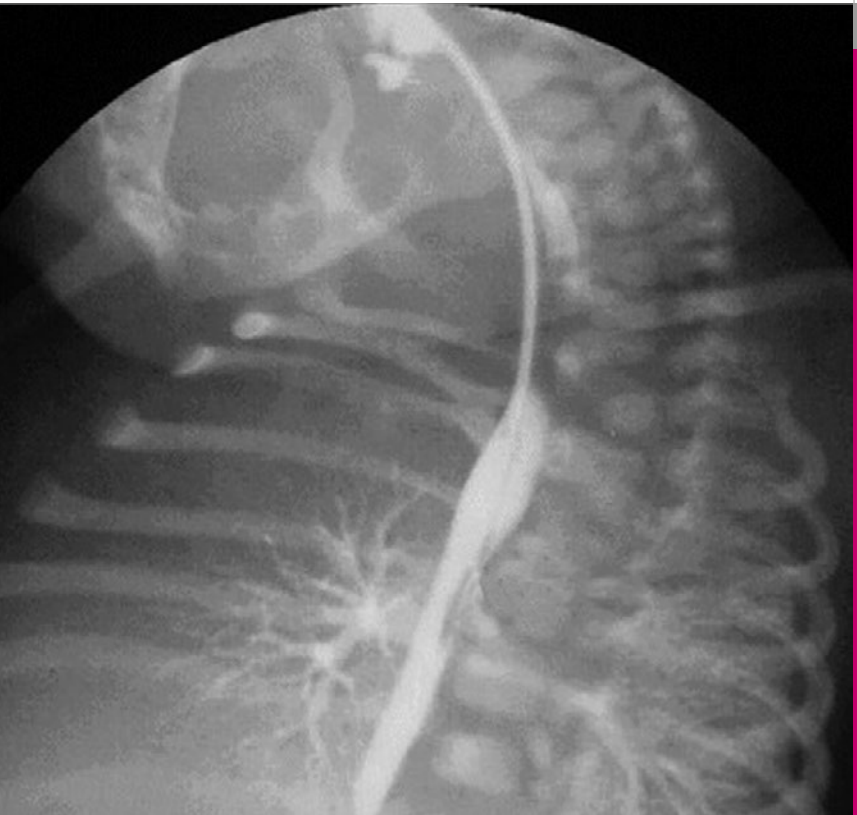
Tracheal agenesis – a rare congenital anomaly



November 2004

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A female infant was born at term by normal vaginal delivery. Her parents were non-consanguineous. Maternal antenatal screening was negative; however, polyhydramnios was noted at an antenatal scan at 30 weeks. Ultrasound scan at 36 weeks revealed hydronephrosis of the fetal left kidney.

At birth, the baby was floppy, gasping, cyanosed and bradycardic. At intubation, the vocal cords were visualized clearly. A Portex (size 3.0) endotracheal tube was passed but resistance was felt beyond the cords. Intermittent positive pressure ventilation was commenced, but there was no air entry to the chest or the stomach on auscultation. The endotracheal tube was removed and bag and mask ventilation started. Further attempts to intubate past the cords were unsuccessful, despite using smaller endotracheal tubes.

Full resuscitation was commenced and intermittently breath sounds could be heard in both lungs. The heart rate picked up to 100 beats per minute but the infant's color failed to improve. Crystalloids and sodium bicarbonate were administered through an umbilical venous catheter and bag and mask ventilation was continued with little success. Attempts to intubate with an introducer by an otolaryngologist were also unsuccessful. With airway patency in question, and a heart rate not sustainable without adrenaline, resuscitation was halted following discussion with the parents. A preliminary diagnosis of tracheal stenosisor agenesis was

made. This baby had a rudimentary right ear, absent right radius, thumb and index finger and a grossly distended, tense abdomen.

A postmortem contrast esophagogram performed underdigital fluoroscopy demonstrated an anomalous fistulous communication between the esophagus and carina (Figure). There was retrograde filling of the carinal stump and both bronchi. A skeletal survey showed a sixth dorsal hemivertebra and an absent right radius.

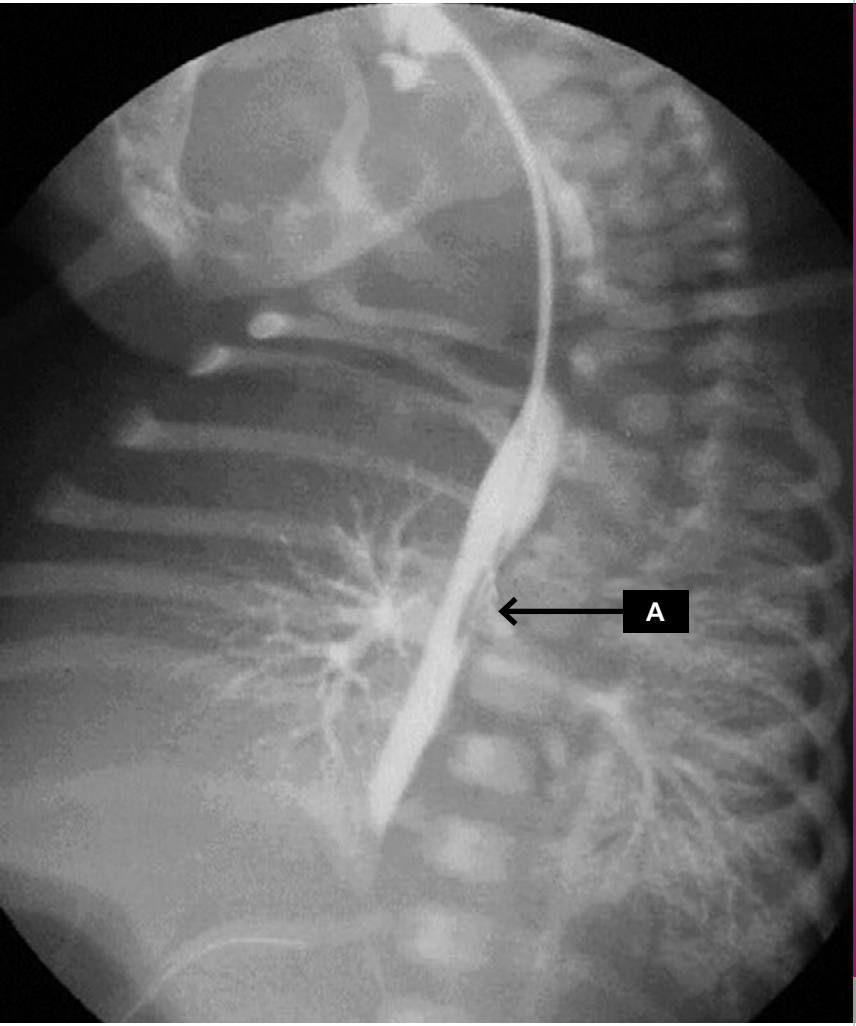

Fig. 1

Contrast esophagogram showing an anomalous fistulous communication (A) between the esophagus and carina.

DISCUSSION

Tracheal agenesis is a rare congenital anomaly occurring in association with anomalies of the heart, genitourinary, gastro-intestinal, pulmonary and central nervous systems. Incidence is reported to be less than 1:50'000 with a male predominance (1).

At birth, these babies present with asphyxia, absent cry or respiratory distress. An inability to intubate alerts one to the presence of tracheal agenesis. An accidental esophageal intubation may improve the respiratory status temporarily if a tracheo-oesphageal fistula is present.

Success with intubation is operator-dependent but failure to intubate a neonate at birth should bring this condition to mind and a trial of esophageal intubation and ventilation could be attempted. Chest movement should be observed to check if this manoeuvre is useful. A supraglottic device like a laryngeal mask airway may be tried (1). Standard neonatal resuscitation manuals do not go into depth about what to do in cases of impossible intubation (2).

Tracheal agenesis was first described by Payne in 1900 (3). Kluth et al. showed that the esophagus and trachea develop as the foregut decreases in size by infolding without formation of a fused septum. They proposed that tracheal atresia with fistula may result from a ventral deformation of the foregut and a concomitant dorsal dislocation of the tracheo-esophageal space (4).

In 1962, Floyd proposed an anatomical classification of this malformation. Type I is atresia of part of the trachea with a normal but short distal trachea, normal bronchi, and a tracheo-esophageal fistula. This type makes up approximately 20% of the malformations. Sixty percent of the reported cases are type II, where there is complete tracheal atresia but with normal bifurcation and bronchi, as in our case. Type III has no trachea and the bronchi arise directly from the esophagus and account for 20% of cases (3).

Antenatal ultrasound may show bilateral uniform hyperechoic lungs and ascites if the trachea or larynx is completely obstructed. The inspissated lung fluid and secretions cause overdistension of the lungs. Inversion or flattening of the diaphragm can occur. Contiguous compression of the fetal heart results in low output heart failure. In the presence of an esophageal fistula, the lungs do not become enlarged as fluid escapes through the fistula into the gastrointestinal tract. Color and spectral Doppler ultrasonographic findings may show absence of blood flow at the laryngeal level (4).

In our case, VACTERL association is a possibility as the anomalies included tracheal agenesis, a carino-esophageal fistula, a 6th dorsal hemivertebra, left-sided hydronephrosis and an absent right radius,thumb and index finger. In VACTERL association, tracheo-esophageal fistula (70% cases) is more frequently seen with esophageal atresia (5).

Survival of infants with tracheal agenesis is rare and correction is very difficult. Short term survival may be possible if there is a fistulous connection between the esophagus and bronchus. The first surgical correction was attempted in 1960 (3). Since then 11 cases with varying degress of tracheal agenesis have been resuscitated and treated with a variety of palliative and tracheal reconstructive procedures. Published reports show long-term survival in 3 of them (1,6).

It has to be emphasized that attempts at utilizing the esophagus as an air conduit is only temporary. However, this may allow time for the diagnosis to be confirmed. It is hoped that the use of tissue engineered cartilage may improve the outcome of those babies fortunate enough not to have other major associated congenital anomalies.

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