Tracheal Agenesis: A Rare But Important Congenital Anomaly

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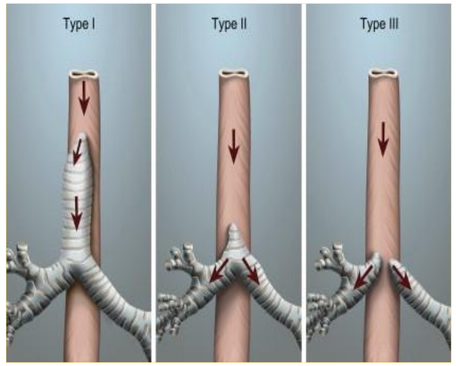
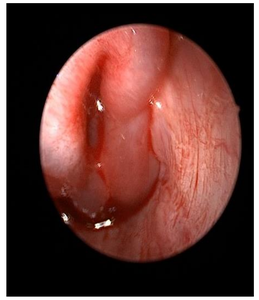
Introduction

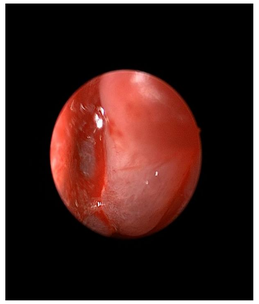
- Tracheal agenesis is a rare and lethal congenital malformation in which the trachea is absent or underdeveloped.
- The prevalence of tracheal agenesis is less than 1:50,000 with a male to female ratio of 2:1.
- There are 3 types of tracheal agenesis:
- Type I, characterized by agenesis of the proximal trachea, normal short distal trachea, normal bronchi and a distal tracheoesophageal fistula.
- Type II, defined by absence of the trachea and normal bifurcating bronchi with or without a broncho-esophageal communication.
- Type III, defined by absence of the trachea with the two mainstem bronchi arising independently from the esophagus.
- We report a case of a term newborn with Type I tracheal agenesis.

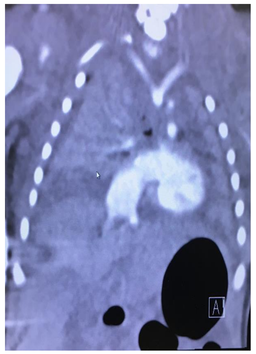
Case Presentation

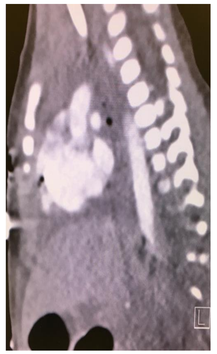
- A full term female was born via spontaneous vaginal delivery to a healthy 31 y/o G3P011 mother. Ultrasound in the first trimester showed an echogenic bowel that resolved by 21 weeks.
- Upon delivery, thick meconium, apnea, poor tone, and pallor were noted. Bulb suction with positive pressure ventilation was initiated by the NICU team. Apgar scores were 1 and 2 at one and five minutes.
- Because of minimal response to mask ventilation, intubation was attempted. The larynx was visualized but attempts at placing the endotracheal tube were unsuccessful. A laryngeal mask airway was placed with end-tidal CO2 detected. CPR was initiated and multiple doses of epinephrine were given along with dopamine and nitric oxide.
- The infant was transferred to our center with the diagnosis of difficult airway.
- On arrival at the emergency room, the infant was assessed by the multidisciplinary difficult airway response team (DART) and found to be dusky, intermittently bradycardic, and hypoxic. A rapid decision was made for OR evaluation.











Case Presentation

- In the OR, with ongoing CPR, direct laryngoscopy by ENT revealed Grade 1 view of vocal cords with no airway below the cords. A rigid bronchoscope could not access the trachea.
- Because of declining infant condition, extra-corporeal membrane oxygenation (ECMO) was initiated.
- Ultrasound and CT after ECMO cannulation could not locate the trachea.
- Following a multidisciplinary discussion in the PICU, support was terminated because the infant would not likely benefit from additional surgical intervention.
- Postmortem exam revealed Type I Tracheal Agenesis, a bi-lobed right lung, narrowing of the left ureter at the bladder and a membrane covering a portion of the ascending colon.

Discussion

- Tracheal agenesis is a rare diagnosis that should be considered in the delivery room.
- Approximately 90% of patients with tracheal agenesis also have cardiovascular, gastrointestinal, and genitourinary abnormalities.
- Severe respiratory distress immediately after birth, accompanied by cyanosis and a weak or absent cry, may help in the diagnosis, especially in the setting of difficult intubation.
- Temporary ventilation with an endotracheal tube placed in the esophagus may be helpful in the presence of a fistula.
- The success of tracheostomy depends on the length of the distal trachea. In most cases, long-term solutions for tracheal agenesis are limited and the outcome is almost always fatal.

References

1. BMC Pediatrics (2017) 17:49
2. Brooklyn Med Journal (1900)14:56