Congenital Tracheal Aplasia Without Prenatal Diagnosis Masked by Maternal Obesity and Gestational Diabetes: A Case Report

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This case report describes a neonate with tracheal aplasia first diagnosed after birth due to the presentation of respiratory distress, absence of crying, and unsuccessful tracheal intubation. The most common finding with tracheal aplasia is polyhydramnios. However, diagnosis remains challenging in the prenatal period. In this case, maternal obesity and gestational diabetes made diagnosis more difficult. The only lifesaving treatment available is ventilation through esophageal intubation or tracheostomy. However, in some cases, tracheostomy is not an option. (A&A Practice. 2020;14:e01200.)

GLOSSARY

BMI = body mass index; **CO_2** = carbon dioxide; **CT** = computed tomography; **ETT** = endotracheal tube; **ID** = inner diameter; **MRI** = magnetic resonance imaging; **NICU** = neonatal intensive care unit; **PROM** = premature rupture of membranes; **Spo_2** = peripheral oxygen saturation; **TACRD** = tracheal agenesis/atresia, complex congenital cardiac abnormalities, radial ray defects, and duodenal atresia; **TEF** = tracheoesophageal fistula; **VACTERL** = vertebral defects, anal atresia, cardiovascular defects, tracheoesophageal fistula and/or esophageal atresia, radial dysplasia, renal defects, and limb defects

This case report discusses a 2290-g neonate born by cesarean delivery at 37 weeks of pregnancy due to the premature rupture of membranes (PROM) and previous resection of maternal uterine myomas. The infant was struggling to breathe and did not cry. The vocal cords were closed, pinker, and their edges appeared somewhat more angular than usual, when viewed using a laryngoscope, and tracheal intubation was impossible. Prenatal growth was normal at each stage of pregnancy with no prenatal diagnosis of congenital tracheal aplasia. Written informed consent for publication was obtained from the parents of the infant.

CASE DESCRIPTION

The mother was 38 years of age with a history of laparoscopic surgery to treat uterine myomas. Following this procedure, she attended an infertility clinic, became pregnant, and was subsequently closely monitored. Before pregnancy, she was 158-cm tall and weighed 91-kg, with a body mass index (BMI) 36.5. During pregnancy, the mother's weight increased, and gestational diabetes developed, but blood glucose levels were controlled by diet alone. Polyhydramnios was noted during pregnancy.

The amniotic fluid pocket was deep, and thus, fetal ultrasound examinations were only able to measure fetal growth, which was normal. A cesarean delivery was scheduled at 38 weeks of pregnancy due to previous uterine surgery. One week before the scheduled cesarean delivery, the mother was admitted with PROM, and a category 4 cesarean delivery was performed that day. The body weight of the mother was 96-kg (BMI 38.5) when the infant was delivered.

Spinal-epidural anesthesia was provided. A neonatologist and a nurse from the neonatal intensive care unit (NICU) were present during delivery, as is typical in our hospital. Despite PROM, 5000 mL of amniotic fluid was collected. When the infant was born, she did not cry and had weak muscle tone. The neonatologist administered ventilatory support using a bag-valve-mask. However, effective ventilation was not achieved. Peripheral oxygen saturation (Spo_2) was 70%–90% and pulse rate was 120–140 beats per minute. The neonatologist diagnosed fetal asphyxia and unsuccessfully attempted tracheal intubation.

The maternal anesthesiologist then inserted a 3.5-mm inner diameter (ID) cuffed endotracheal tube (ETT) into the larynx of the infant via the nasal passage and attempted to ventilate by closing the nose and mouth so not to leak ventilation.[1] The assisted ventilation appeared to be effective. However, it was noted that the infant's stomach had extended. Thus, the anesthesiologist attempted to intubate the infant orally using a 3.0-mm ID uncuffed ETT with a stylet. The larynx and the area around the vocal cords were observed using a laryngoscope (Cormack-Lehane classification[2] Grade 1) and initially appeared normal. However, the vocal cords were closed, pinker, and the edges of the

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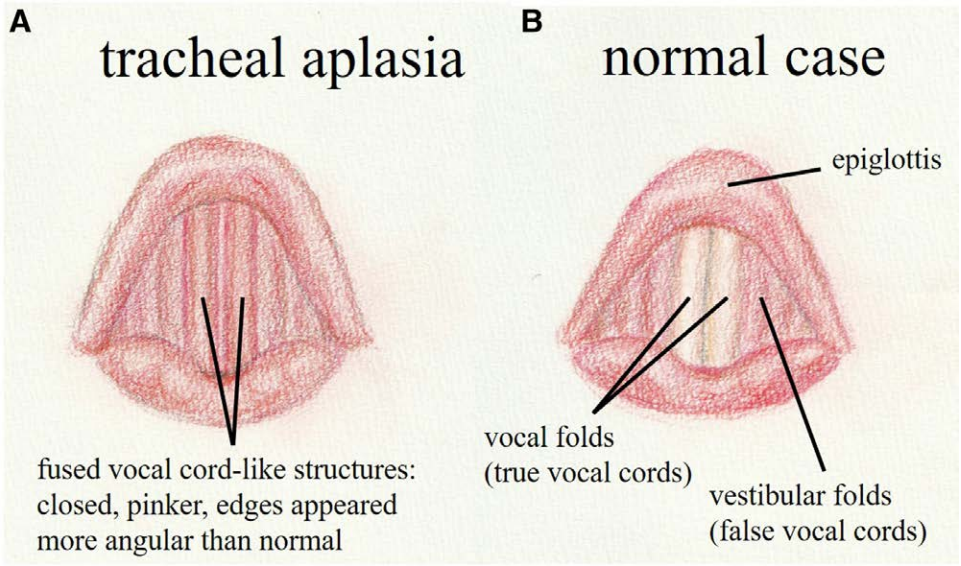

Figure 1. Schema of the larynx of the newborn in the current case and in a normal case. A, A sketch representation showing the closed vocal cords, which also appear pinker with the edges appearing more angular than normal. B, A sketch representation of the vocal cords, which appear whiter than the surrounding tissue in normal cases.

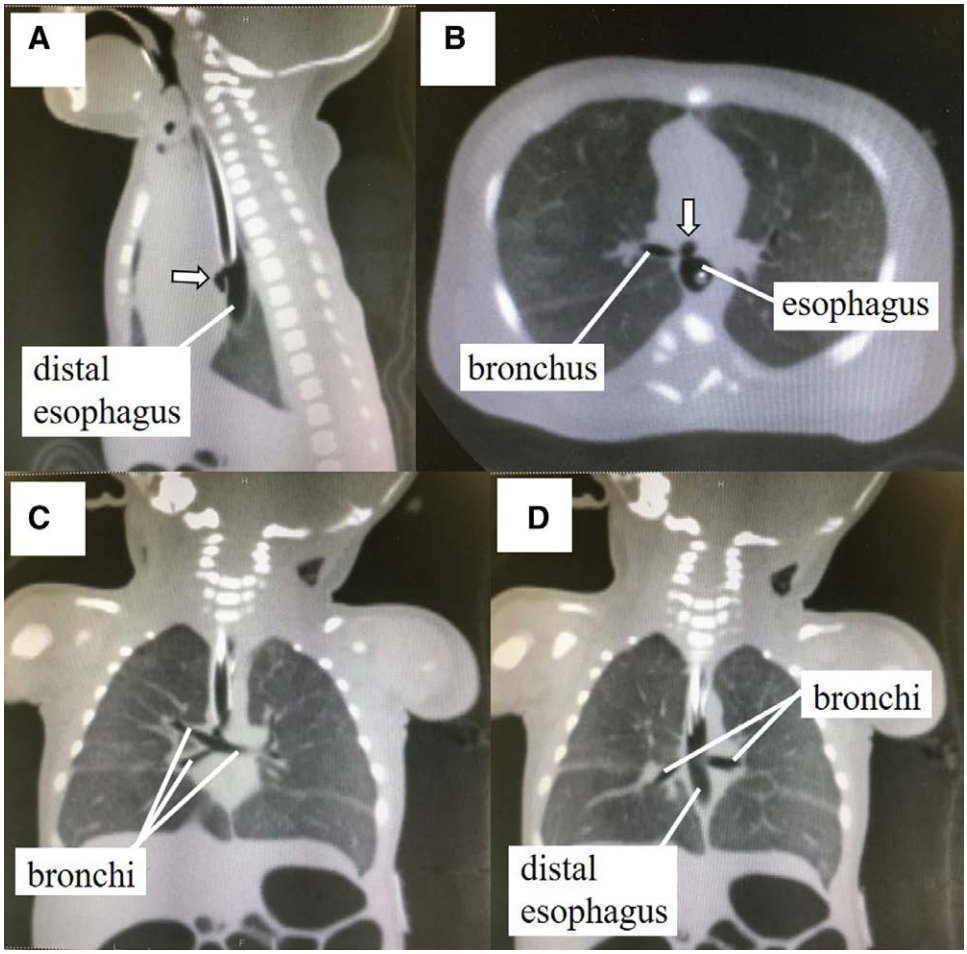

Figure 2. Findings of emergency CT scan. A, The sagittal section showing the ETT, which has been inserted into the esophagus and the TEF (arrow). B, The horizontal section showing TEF (arrow). C and D, The coronal sections showing the left and right bronchi branched from the TEF. The CT scan also shows abnormality of the lung lobes. CT indicates computed tomography; ETT, endotracheal tube; TEF, tracheoesophageal fistula.

vocal cords appeared somewhat more angular than usual (Figure 1). The anesthesiologist attempted to open the vocal cords using a 3.0-mm ID uncuffed ETT without stylet and perform tracheal intubation by advancing gently and rotating the ETT slightly. From his experience, tracheal intubation can be achieved without muscle relaxants in routine neonatal cases. However, this attempt was unsuccessful. At this point, an intravenous line was not yet secured. The anesthesiologist was able to visualize the vocal cords, and therefore, he attempted to advance a 3.0-mm ID uncuffed styletted ETT with a little more force, but the vocal cords remained closed. A further attempt using a 2.5-mm ID

uncuffed styletted ETT was also unsuccessful. A second neonatologist then arrived and attempted tracheal intubation, which appeared to be successful on the first attempt. A carbon dioxide (CO_2) indicator confirmed the return of CO_2 from the ETT. However, bag-mask ventilation did not result in chest expansion, the stomach continued to distend, and the Spo_2 remained around 70%–90%. The pulse rate remained stable at 120–140 beats per minute, and the patient was immediately admitted to the NICU.

Following admission to the NICU, ventilation continued to be difficult. The newborn had repeated episodes of bradycardia, hypoxemia, and acidemia, and required cardiopulmonary resuscitation. Echocardiography revealed a double-outlet right ventricle and pulmonic stenosis. Otolaryngologists were requested to perform an emergency tracheostomy. The otolaryngologists examined the patient's neck and located the hyoid bone, but could not locate the thyroid cartilage, cricoid cartilage, or trachea. Additionally, they could feel the ETT, which had been inserted into the esophagus. The newborn's transfer to obtain an emergency computed tomography (CT) scan was very challenging. However, the CT scan confirmed that the ETT had been inserted into the esophagus and that a tracheoesophageal fistula (TEF) was present, and the left and right bronchi were branched from the TEF (Figure 2). The CT scan findings confirmed the diagnosis of tracheal aplasia of Floyd type II[3]/Faro C[4] without a proximal trachea, which makes a tracheostomy impossible (Figure 3). The possibility of surgical interventions such as distal esophagus occlusion, banding of the abdominal esophagus, transection between the esophagus and the stomach and gastrotomy were considered to provide effective ventilatory management and gastric decompression[5] for initial stabilization, but the patient's condition continued to worsen. Veno-venous extracorporeal membrane oxygenation was also considered.[6] However, the parents refused further treatment. The neonate died 6 hours after birth. Consent for an autopsy was not obtained.

DISCUSSION

Tracheal aplasia is a rare (<1/50,000 births)[7] but severe congenital malformation, which poses a threat to life immediately after delivery.[8,9] The embryologic pathogenesis remains unclear,[10] and the condition is more prevalent in males (65%).[9] Tracheal aplasia is often associated with malformations such as vertebral defects, anal atresia, cardiovascular defects, tracheoesophageal fistula and/or esophageal atresia, radial dysplasia, renal defects, and limb defects (VACTERL) or tracheal agenesis/atresia, complex congenital cardiac abnormalities, radial ray defects, and duodenal atresia (TACRD) syndrome.[8,9] Tracheal aplasia is the complete or partial absence of the trachea with or without TEF.[3,4] Not every type of tracheal aplasia is classified.[8] When TEF is absent, the outflow obstruction of lungs leads to fluid accumulation and enlarged echogenic fetal lungs, dilated trachea, and ascites (congenital high airway obstruction syndrome). Therefore, tracheal atresia is usually suspected during prenatal ultrasound examination.[8] However, the prenatal diagnosis of tracheal aplasia is challenging in cases in which TEF is present, as the lungs appear normal on ultrasound scans. In such cases, polyhydramnios is often

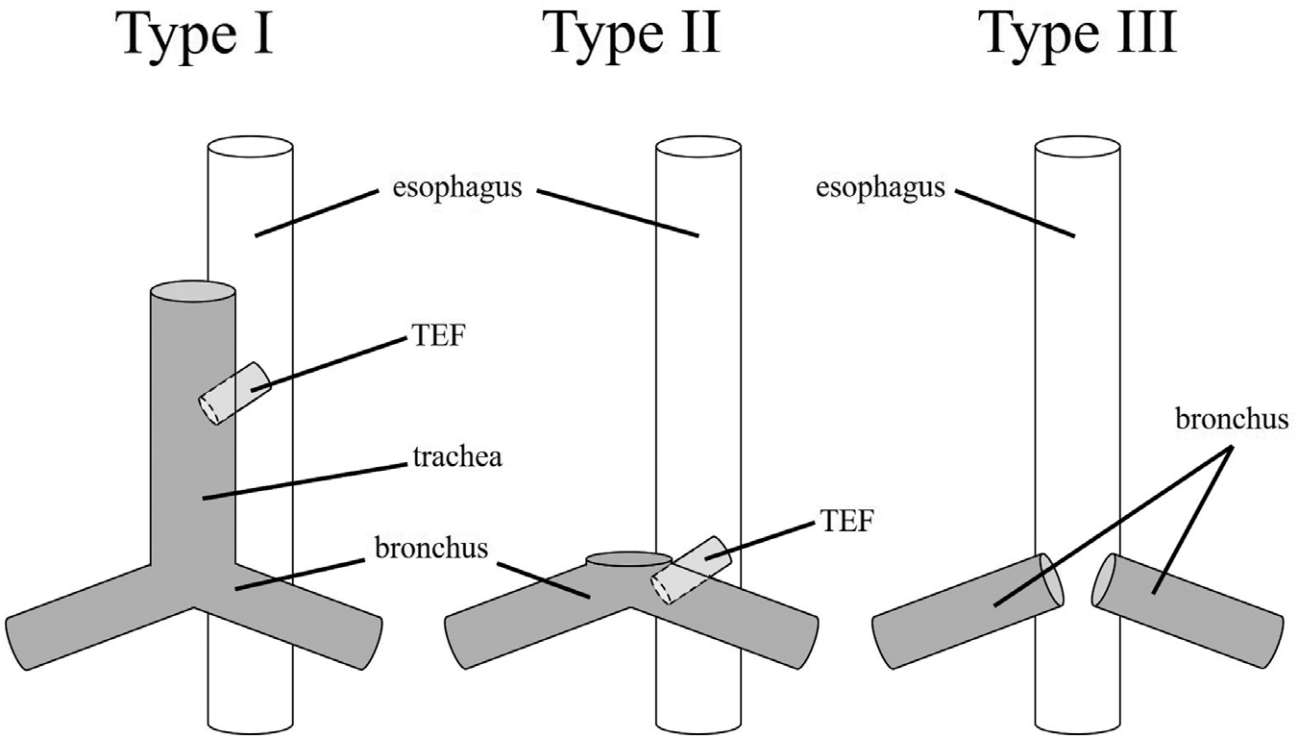

Figure 3. Diagrams of Floyd's classification.[3] Tracheal aplasia comprised of 3 categories based on the types of TEF and the presence or absence of the trachea. Type 1: Defect only in the proximal trachea; distal trachea, trachea bifurcations, and bronchi are present. Type 2: Complete defect of the trachea; trachea bifurcations and bronchi are present, TEF is either present or absent. Type 3: The left and right bronchi branch directly from the esophagus. TEF indicates tracheoesophageal fistula.

the only abnormal ultrasound finding when the infant has no additional congenital malformations.[8] It is reported that prenatal abnormal findings are present in only 56% of tracheal aplasia cases, and the most common finding is polyhydramnios.[9] The infant in the current case had tracheal aplasia with TEF. In addition, the present case was complicated by gestational diabetes, which is also associated with polyhydramnios. Furthermore, the mother was obese, making an accurate fetal diagnosis, except for fetal growth measurement by ultrasound, difficult.

Several reports concluded that magnetic resonance imaging (MRI)[9] and CT[11] may help to diagnose the tracheal aplasia prenatally. However, it is unrealistic to perform these examinations in all cases of polyhydramnios in daily clinical practice. Thus, the majority of tracheal aplasia cases are likely to be diagnosed for the first time after birth. Tracheal aplasia should be suspected in newborns demonstrating "respiratory distress with breathing movement," "absence of crying," and "impossible tracheal intubation," especially when polyhydramnios has been diagnosed during pregnancy.[12] If a fetal diagnosis had been made, surgical interventions could be considered in advance for initial stabilization. However, the prognosis of tracheal aplasia is very poor and varies based on the type of tracheal aplasia.[5,13–15]

In the present case, if the anesthesiologist had forcibly broken the fused vocal cord-like structures without a patent trachea, fatal iatrogenic complications would have occurred. To avoid iatrogenic complications, it is vital that clinicians distinguish types of tracheal aplasia in which a tracheostomy is not an option due to a proximal tracheal defect, as was seen in our case.[3,4] The patient's neck should be examined on suspicion of tracheal aplasia for the presence or absence of structures such as the thyroid cartilage.

The important message is to consider congenital tracheal aplasia when newborns present with the 3 signs mentioned above, and realize that the only lifesaving emergency treatment is ventilation via esophageal intubation. This is effective only in cases in which there is a connection between the esophagus and the trachea or bronchi (TEF). ■

DISCLOSURES

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