Case Report

Indian Journal of Genetics and Molecular Research
Volume 4 Number 1, January - June 2015
DOI: https://dx.doi.org/10.21088/ijgmr.2319.4782.4115.7

Tracheal Agenesis: Role of Fetal Autopsy

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Abstract

Tracheal agenesis is a very rare congenital airway anomaly of respiratory system with incidence of 1 in 50,000 newborns. The condition may be diagnosed antenatal if associated with Tracheo-esophageal fistula (TEF) presenting as Congenital High Airway Obstruction Syndrome (CHAOS). It may pose a great challenge to the attending neonatologist both in diagnosis and in establishing the airway soon after birth and associated with high mortality even with best possible facilities. Tracheal agenesis is difficult to detect prenatally. We report a case of tracheal atresia Faro type E associated with other congenital anomalies.

Keywords: CHAOS; Tracheal Atresia; Tracheal Abnormality; Tracheal Agenesis.

Introduction

Tracheal agenesis is an extremely rare anomaly and is mostly incompatible with life. It usually comes as surprise to attending neonatologist with failure to intubate trachea despite repeated attempts. TA was first described in 1900 by Payne and was classified based upon the presence and length of the tracheal remnant (1, 2). Dilated airways, enlarged lungs with flattened diaphragm, fetal ascites and severe non-immune hydrops can be observed in Congenital High Airways Obstruction Syndrome (CHAOS). Early diagnosis helps in deciding options including Extra-utero intra-partum treatment (EXIT) procedure. The condition is being reported for its rarity and its association with other anomalies.

Case Report

A 25 years old primigravida unbooked woman arrived in labor room of the hospital with labor pain. Emergency evaluation suggested term, IUGR fetus with severe oligohydramnios and fetal distress. Antenatal third trimester ultrasound at arrival to hospital revealed severe oligohydramnios. There was no history of any major medical or surgical illness, drug intake or radiation exposure in the antenatal period. Female baby delivered with gasping respiration and bradycardia. After initial steps of resuscitation, Bag & Mask ventilation started. As it failed to give chest rise, intubation was attempted but unsuccessful despite repeated attempts. The endotracheal tube couldn't be negotiated through normal appearing larynx, Needle cricothyroidotomy was tried for ventilation but failed. Despite all efforts to resuscitate, baby could not be revived.

On physical external examination, baby had single umbilical artery and flat chest with no other obvious external malformation. After getting written consent from parents, photographs were taken; a whole babygram and autopsy were performed. There was normal appearing laryngeal opening which was blind below vocal cords, tracheal atresia of 2 cm in upper part which was patent in lower segment with normal division as two patent bronchi. Lungs were occupying fifty percent of the thoracic cavity and were dense, firm and without crepitus. The lungs showed atelectasis with intense congestions.

Histopathology revealed solid area of trachea with no opening showing cartilaginous tissue surrounded by fibromuscular tissue and minor salivary glands. Area with patent trachea showed normal tracheal

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tissue. The esophagus was normal and there was no communication with trachea. There was no vascular ring or abnormality in the adjoining areas and thymus was normal. The condition is also considered as congenital high airway obstruction syndrome (CHAOS) – Tracheal atresia Faro type E. Other anomalies were also found like single umbilical artery and a small muscular ventricular septal defect. Radiograph was normal.

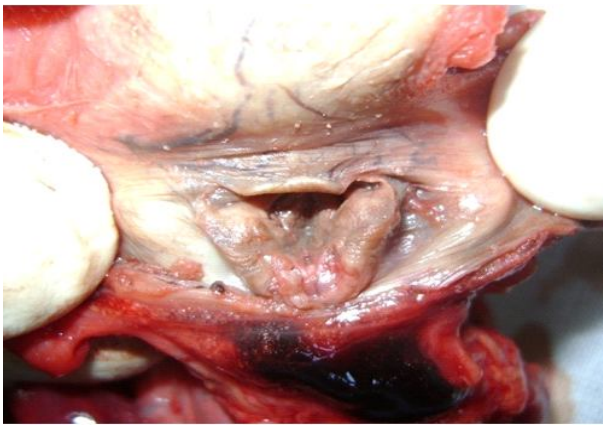

Fig. 1a: Laryngeal inlet-Laryngeal inlet-normal epiglottis, arytenoid, aryepiglottic folds & vocal cords.

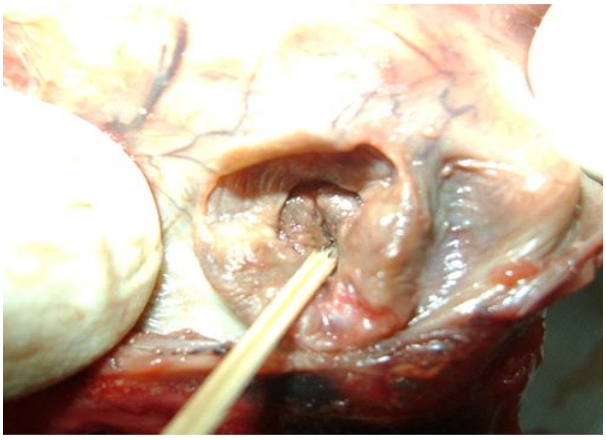

Fig 1 b: Absent opening below vocal cords

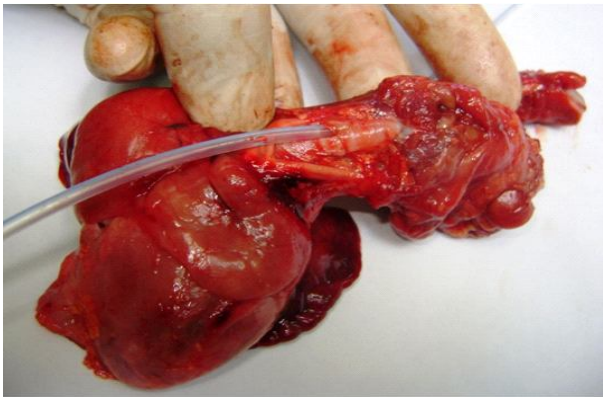

Fig. 2: Tracheal atresia

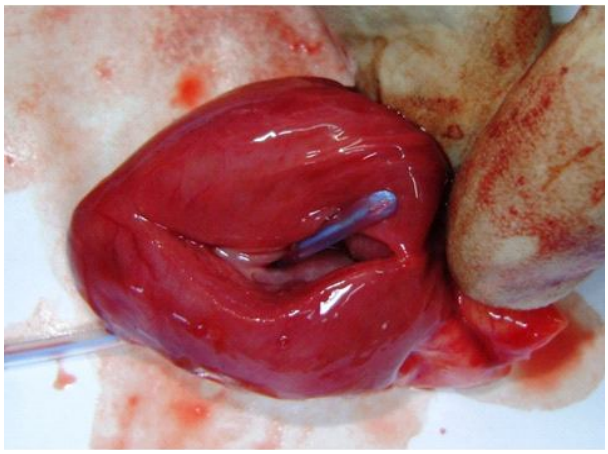

Fig. 3: Muscular Ventricular septal defect

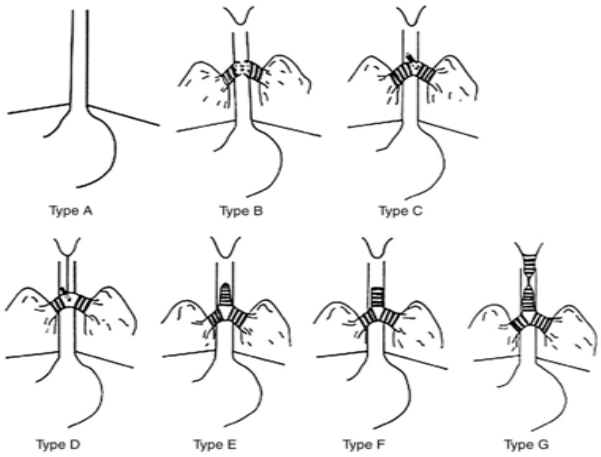

Fig. 4: Faro's classification of Tracheal agenesis (source: Ref 2)

Discisson

Tracheal agenesis (TA) is a rare congenital defect that confronts obstetricians, neonatologists, and anesthesiologists with an unexpected emergency presentation. This usually lethal defect has an incidence of 1 per 50,000 newborns and consists of complete or partial absence of the trachea below the larynx, with or without a concomitant tracheoesophageal fistula (TOF). It has shown a male predominance[1]. It was first described by Payne in 1900. The exact cause for this condition is unknown. Severe respiratory distress, cyanosis, absence of audible crying. Difficult or impossible endotracheal intubation and successful ventilation through accidental esophageal intubation are the hallmark of the diagnosis. DeLuca et al have discussed various resusucitative issues related to tracheal agenesis [3].

The embryo pathology resulting in this abnormality is not completely understood but tends to occurs during the first eight weeks of gestation.

The trachea- pulmonary complex develops from the respiratory diverticulum at the ventral aspect of the primitive foregut. A compromised vascular supply to the developing trachea during this stage may cause tracheal agenesis or tracheal stenosis with complete tracheal ring. According to Merei *et al*., the point of bifurcation between the developing trachea at ventral and developing esophagus at dorsal foregut remains fixed in relation to the cervical vertebra. Caudally, the respiratory diverticulum will develop into the carina and bronchi-pulmonary tree. The cephalic aspect of the respiratory diverticulum will be elongated to form the trachea and the infra- glottic structure. Tracheal agenesis results when this normal elongation process fails to take place [4].

Faro *et al* divided tracheal atresia into categories "A" to "G", representing various types of airway anomalies decreasing in severity, with "A" representing total pulmonary atresia and "G" representing tracheal stenosis [2]. Other malformations are associated in more than 80% of cases. These include VATER association, complex congenital heart disease, genitourinary, gastrointestinal, central nervous system, aneuploidies and musculoskeletal system defects.

Antenatal diagnosis is difficult, challenging and can be confirmed prenatally by magnetic resonance imaging (MRI) if suspected [5]. Our case didn't had polyhydramnios and therefore difficult to suspect prenatally. At present there is no specific surgical management technique that is associated with survival of tracheal agenesis. Antenatal diagnosis allows the chance for intervention strategies such as the ex-utero intrapartum treatment (EXIT), using placental circulation during hysterectomy or transfer to centre with this facility [6,7]. Improving awareness about the condition, antenatal diagnosis and EXIT procedure has led to improved survival rate in this rare anomaly [8,9]. Promptly performed surgical tracheotomy is the only mean to ventilate such a baby. Besides environmental factors, animal models of different genetic defects, such as (conditional) inactivation of *Gli2, Gli3, Shh, Foxf1*, and *b-catenin*, show Tracheal Atresia or incorrect septation of the foregut. A major role for the *BMP type I* receptor genes in mouse models is suggested [10]. However, no causal gene has been identified in human TA patients yet.

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