E12 A Multiple Congenital Malformation in a Forensic Infant Autopsy

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Learning Overview: After attending this presentation, attendees will understand the pathophysiology and presentation of Congenital High Airway Obstruction Syndrome (CHAOS).

Impact on the Forensic Science Community: This presentation will impact the forensic science community by discussing the methods necessary for diagnosing CHAOS at autopsy for forensic purposes.

CHAOS is a rare anomaly that is characterized by a partial or complete obstruction of the fetal upper airways. Its exact incidence is not known. The most common cause of CHAOS is the laryngeal atresia.[1,2] Other causes are laryngeal webs and cysts and tracheal agenesis or atresia. At prenatal Ultrasonography (USG), the fetus may be characterized by enlarged echogenic lungs, inverted or flattened diaphragm, and fetal ascites.[3] In fact, "the non-clearance of fluid from the lungs, due to obstruction, results in a dilated trachea and lung parenchymal hyperplasia. This further leads to compression of the great veins and right atrium leading to ascites. The compression of the esophagus due to a dilated trachea may lead to polyhydramnios."[2] In addition, the heart may be centrally placed because of the compression of the enlarged lungs.[4]

This presentation reports on a singular case of CHAOS in a female infant in which congenital agenesis of the trachea and atresia of the esophagus led to her death shortly after delivery, despite intubation attempts.

A 26-year-old pregnant woman was transported by ambulance to the hospital because of intense uterine contractions. The information about the woman's pregnancy was limited because she lived in another country during the previous months of her pregnancy. The only information for clinicians included: 33 weeks + 2 days of gestational age, no known maternal pathologies, and amniocentesis had not been conducted. The maternal weight, stature, and Body Mass Index (BMI) were, respectively, 63kg, 170cm, and 19kg/m^2. The woman gained approximately 8kg of weight during the pregnancy and did not receive regular USG or clinical medical evaluations during pregnancy. The only fetal USG available (conducted 13 days before delivery) showed polyhydramnios, suspected esophageal atresia, and arthrogryposis.

Immediately after admission of the woman to the hospital, the physicians decided to perform an emergency cesarean section because the fetal cardiotocography was highly pathologic. At delivery, the baby weighed 1.7kg, was non-reactive to stimuli, atonic and cyanotic, and the APGAR score was 1, 0, 0. The heart rate was 30bpm. The neonatologist attempted to aspirate the secretions from the upper respiratory airways, but to no avail. Therefore, he immediately tried to intubate the baby, inserting the tube as far as the vocal cords, but was unable to advance the tube beyond that point. Finally, despite multiple intubation and resuscitation attempts, the baby died. The case was referred to the forensic pathologist.

On the gross examination, the heart was centrally placed, and both lungs were enlarged and characterized by two lobes. The larynx was normally developed, but the probe did not manage to reach the trachea due to a cartilaginous septum that was located between the larynx and the trachea. The distal part of the trachea and the bronchi were present, and the trachea (in correspondence with the carina) showed a trachea-esophageal fistula. On the other end, the esophagus was atresic. Indeed, the probe did not manage to go beyond the upper part of the esophagus. In addition, the distal part of the esophagus was present and connected to the trachea through the fistula mentioned above. There was also peritoneal effusion, a small stomach, unicornuate uterus, and a recto-cloacal fistula. The microscopic examination only revealed an accumulation of mucus in the alveoli. The death was due to the congenital tracheal malformation that did permit intubation. The manner of death was natural. Reviewing the literature, the pathologists understood that the baby was affected by a peculiar form of CHAOS in which a type 2 of congenital tracheal agenesis (Floyd classification) was associated with a type C esophageal atresia (Gross classification).[5,6] The combination of these congenital anomalies has not yet been described in the literature. This may generate difficulties in the interpretation of anatomical findings at forensic autopsy. For these reasons, this presentation should serve as a stimulus to improve knowledge in the forensic community regarding these types of congenital malformations.

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Congenital High Airway Obstruction, Forensic Infant Autopsy, Multiple Congenital Abnormalities