H141 When the Autopsy Is the Only Possibility to Obtain a Diagnosis: The Investigation of a Rare Case of Aortopulmonary Window (APW)

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Learning Overview: The goal of this presentation is to report the autopsy investigation of a neonatal death due to a rare congenital defect, APW.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by outlining the role of forensic pathology in the management of a difficult task, such as the diagnosis of congenital heart defects.

APW is one of the rarest congenital heart defects, accounting for 0.5% of all congenital heart defects. The malformation is caused by incomplete development of the aortopulmonary septum during fetal life. APW can be isolated or seen as a part of a wide variety of other cardiac anomalies, such as atrial or ventricular septal defects, patent ductus arteriosus, Berry syndrome, pulmonary atresia, aberrant subclavian artery, and tetralogy of Fallot. Patients with APW have increased pulmonary flow, due to the presence of a left-to-right shunt, which leads to pulmonary hypertension in early life and is associated with a poor prognosis in the absence of surgical correction.

This is the report of a case of a male newborn delivered via caesarean section at full-term (39 weeks) who rapidly developed cyanosis, poor reactivity, and hypotonia, necessitating partial resuscitation and oxygen therapy. The quick clinical deterioration resulted in rapid transportation to an advanced hospital where a diagnosis of respiratory distress was made. Despite the intensive care, the symptoms worsened, and the newborn died about 15 hours after birth. An autopsy investigation was performed in order to establish the cause of the symptoms and death.

At autopsy, the cervical, thoracic, and abdominal organs were removed en masse, according to the Letulle technique. After external examination, the heart, lungs, and aorta were separated as a block, and subsequently fixed in formalin. Gross examination after formalin fixation showed an increased heart volume and a pseudo-aneurysmal dilatation of the pulmonary artery. Sectioning of the great vessels highlighted a wide communication between the aorta and pulmonary artery due to lack of an aortopulmonary septum; the defect was located about 3mm before the origin of the left subclavian artery. Main branches of the aortic arch were normal, except for a stenosis of the left subclavian artery approximately 6.6mm from its origin. The findings identified during the postmortem examination made it possible to identify the cause of death as acute cardiopulmonary insufficiency due to a congenital heart defect with a left-to-right shunt (i.e., APW).

Despite advancements in medical technology and practice, congenital heart diseases are still encountered at autopsy of many stillbirths, newborns, and infants. The rarity and the rapid clinical evolution of these conditions—as well as the low likelihood of antemortem diagnosis in many cases—constitutes a challenge for the forensic pathologist. The current case report emphasizes the importance of choosing the appropriate autopsy technique and the need of training in the detection of congenital heart defects, especially in situations of antemortem misdiagnosis or where ancillary methods, such as forensic radiology, are unavailable.

Congenital Heart Defects, Aortopulmonary Window, Autopsy Investigation

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