H37 A Case of Congenital Laryngeal Stenosis Diagnosed at Autopsy

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After attending this presentation, attendees will understand the usual pathophysiology and presentation of congenital laryngeal stenosis and its diagnosis at autopsy.

This presentation will impact the forensic science community by demonstrating the methods necessary for recognizing and diagnosing cases of congenital laryngeal stenosis at autopsy.

Congenital laryngeal stenosis is a rare condition in which the diameter of the upper airway at any point from the epiglottis to the thyroid cartilages is markedly narrowed.[1-7] This condition is usually diagnosed early in pregnancy as a part of Congenital High Airway Obstruction Syndrome (CHAOS) with ultrasound-detected pulmonary abnormalities, abnormalities of the heart and diaphragm, and increases in amniotic fluid levels.[1-7] While most cases are sporadic occurrences, a few cases reported in the literature suggest an autosomal dominant inheritance.[1,4] CHAOS has reportedly occurred in association with other syndromes such as Fraser syndrome, short rib polydactyl syndrome, Vertebral Defects, Anal Atresia, Cardiac Defects, Tracheo-Esophageal Fistula, Renal Anomalies, and Limb Abnormalities (VACTERL) syndrome, or chromosomal abnormalities such as deletion of chromosome 5p and partial trisomy 5.[1-5] Laryngeal stenosis with prenatal care is usually manageable and survival rates are good when emergent treatment is provided; however, if the stenosis goes unrecognized prior to delivery, the mortality rate can range between 80% to 100%.[2,6,7]

This presentation describes a case of congenital laryngeal stenosis diagnosed at forensic autopsy of a 39-week term gestation infant delivered to a 36-year-old *gravida* 5 *para* 3 mother. The maternal gestational history was largely unknown except for reports of a prior pregnancy ending with preterm delivery of a still-birth fetus who had numerous physical deformities. The prenatal history of the current pregnancy was unremarkable except for gestational diabetes; the mother received regular ultrasounds that exhibited appropriate fetal growth and movement. The mother declined an amniocentesis for genetic testing.

After presenting for induction of labor, the infant was delivered in the hospital and was noted to have APGARs of 1, 1, 1, and the following physical abnormalities: "webbed fingers, small ears, abnormal genitalia, and left eye deformity." The infant survived for approximately one hour, during which time she experienced marked respiratory distress and eventually died. The case was referred to the forensic pathologist by the coroner's office for autopsy.

At autopsy, the body measured 49cm long and weighed 3,200 grams. Significant external findings included minor facial abnormalities, including a small left eye with a partially fused lid, a small left nasal opening, syndactyly, and ambiguous genitalia. Internally, there was stenosis of the larynx at the level of the glottis/subglottis and lumen diameter of ~0.2cm; airways distal to the stenosis were dilated with mucus congestion. Additionally, the right and left lungs weighed 38.3 grams and 27.2 grams, respectively, and were congested but otherwise normal in appearance. Ascites was noted with approximately 20ml of serous fluid in the peritoneal cavity. Microscopically, there was vascular congestion of the alveolar capillaries and numerous intra alveolar foamy macrophages. The remaining organs examined were all histologically appropriate for gestational age.

In this case, the clinical presentation and finding of markedly narrowed larynx are consistent with a death due to the obstruction/constriction of the airway; thus, the primary cause of death was determined to be due to congenital laryngeal stenosis. The reported case is an unusual presentation of a very uncommon anomaly.[8] The presentation of congenital laryngeal stenosis at a full-term delivery is rare, especially when the presentation is unexpected.[8]

Reference(s):

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