05. PENTALOGY OF CANTRELL: REPORT OF A UNIQUE PHENOTYPE

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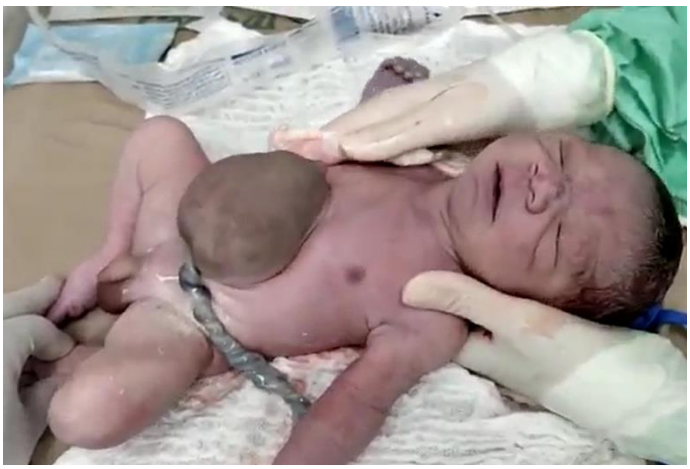
https://www.youtube.com/live/fSpXH-3Xy5w?t=19394s

BACKGROUND: Pentalogy of Cantrell (PC) is a rare congenital anomaly, characterized by the association of ectopia cordis, defects in the thoracoabdominal wall, diaphragm, sternum, pericardium and intrinsic cardiac anomalies. Embryologically, the alteration that causes PC takes place in the fourth week of human development, due to a failure in the processes involved in the formation of the thoracoabdominal wall. The etiology has not been elucidated, but it has been proposed that heterogeneous mechanisms are involved, with an important genetic component linked to the X chromosome. **THE CASE:** We describe the case of a masculine patient product of a non-consanguineous marriage, born at 39 weeks of gestation. Her mother, a 15-year-old, gravida 1 patient, presented to the outpatient OB/GYN service of the Hospital Universitario del Valle in Cali, Colombia, at 37 weeks of gestation, she had no previous prenatal care due to her condition as a recent immigrant from Venezuela with low socioeconomic status, a through history and physical was performed reporting no abnormalities, she was immediately scheduled for a detailed anatomy ultrasound, it showed a fetus with thoracoabdominal ectopia cordis, suggesting PC as a diagnosis. A physician meeting between OBGYNs, pediatric surgeons, neonatologists and pediatric cardiologists was carried out, they opted for a C-section at 39 weeks with the possibility of surgical intervention after birth. After the C-section, the initial evaluation revealed dysmorphic features, a midline defect of the abdominal wall extending from the inferior portion of the sternum to the hypogastrium, with heart and bowel protrusion and absence of the muscle wall, remarkably, the protruded organs were completely covered by skin (Figure). APGAR scores were 7 at one minute and 6 at 5 minutes, due to irregular breathing and cyanosis, the patient was intubated and transferred to the NICU. The echocardiogram showed tetralogy of Fallot and partial diaphragm agenesis. He then was scheduled for a contrast enhanced CT in order to better detail the anatomy with the goal of planning a corrective surgery, unfortunately, before the imaging was performed, and, after 8 days in critical condition in the NICU, the patient died. Multiple genetic, embryologic and environmental factors have been described to explain the etiology of this type of congenital anomalies, however it is yet to be established clearly. **CONCLUSION:** The aim of this study was to provide to the scientific literature the first case of Cantrell's Pentalogy with the described unique phenotype (ectopia cordis, large thoracoabdominal wall defect with evisceration and complete skin coverage, and Tetralogy of Fallot), highlighting the importance of an early prenatal diagnosis, the role of social determinants of health in maternal care, and the involvement of a multidisciplinary team, in order to build rapport with patients, regarding follow-up, genetic and reproductive counseling in challenging scenarios.

Figure: Newborn with Pentalogy of Cantrell: Dysmorphic Features, Thoracoabdominal Wall Defect, and Skin-Covered Evisceration.



Key Words: Pentalogy of Cantrell, Ectopia Cordis, Congenital Abnormalities.