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"Unveiling the Silent Threat: Single Umbilical Artery with Congenital Heart Disease and Pulmonary Hypertension in a Neonate"

Case Report: Infant with Single Umbilical Artery and Congenital Heart Disease with Pulmonary Hypertension

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KEYWORDS

Umbilical, Arteries, Occasionally, Abnormality, Ultrasound, Chromosomal, Incidence.

ABSTRACT:

The umbilical cord typically contains two arteries and one vein. Occasionally, one artery may be absent, with the left artery being more commonly affected than the right(1). Single Umbilical Artery (SUA) is the most common abnormality of the umbilical cord. SUA can be diagnosed prenatally by ultrasound as early as 12 weeks of gestation. Ultrasound findings useful for this diagnosis include the presence of two vessels on a cross-section of the free loop of the cord(2) and observation of the arteries coursing around the bladder in the fetal pelvis. Numerous studies have reported that the presence of SUA is associated with a variety of congenital anomalies, chromosomal defects, aneuploidy, and low birth weight.(3)The incidence estimates of SUA from different countries range from 0.2 to 0.87%.

Introduction:

The umbilical cord typically contains two arteries and one vein. Occasionally, one artery may be absent, with the left artery being more commonly affected than the right(1). Single Umbilical Artery (SUA) is the most common abnormality of the umbilical cord. SUA can be diagnosed prenatally by ultrasound as early as 12 weeks of gestation. Ultrasound findings useful for this diagnosis include the presence of two vessels on a cross-section of the free loop of the cord(2) and observation of the arteries coursing around the bladder in the fetal pelvis. Numerous studies have reported that the presence of SUA is associated with a variety of congenital anomalies, chromosomal defects, aneuploidy, and low birth weight.(3)The incidence estimates of SUA from different countries range from 0.2 to 0.87%.

Case Report

Background

A 1-month and 7-day-old male infant, born at 37 weeks of gestational age via lower segment cesarean section (LSCS) due to safe confinement concerns, presented with a history of weak cry and hurried breathing for 1 day. The infant was the first child of a non-consanguineous marriage. Antenatal scans, including the early pregnancy scan, NT scan, and triple test, were normal. However, the third-level scan at 21 weeks of gestation revealed polyhydramnios, and the anomaly scan at 25 weeks showed a single umbilical artery (SUA). Subsequent growth scans at 34 and 36 weeks confirmed the SUA along with bilateral mild pelviectasia. The infant's birth weight was 2.2 kg.

Clinical Presentation

The infant presented with weak cry, hurried breathing, noisy breathing, and chest retractions. On retrospective

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questioning, a suck-rest-suck cycle was noted during feeding. Initially formula-fed due to feeding difficulties, the infant was exclusively breastfed at the time of presentation. The child had received birth vaccinations. There was a family history of polydactyly on the father's side, but no other significant family history noted

On examination, the infant was awake, active, and had an open anterior fontanelle measuring 2.5x2.5 cm. The lips were dark, and vital signs included a heart rate (HR) of 160 bpm, respiratory rate (RR) of 58/min, afebrile, non-invasive blood pressure (NIBP) of 100/60 mmHg, and SpO2 of 100% on room air. Systemic examination revealed the following:

- **Cardiovascular System (CVS):** S1S2 heard, pansystolic murmur present.
- **Respiratory System (RS):** Bilateral coarse crepitations and chest retractions present.
- **Per Abdomen (PA):** Liver palpable 3 cm below the right costal margin (RCM).

Investigations

- **Blood Tests:** Routine investigations were within normal limits.
- **Radiology:**
  - **Chest X-ray (AP view):** Cardiomegaly with a cardiothoracic ratio of 0.65.
  - **2D Echocardiography:** Showed raised pulmonary pressures of 85 mmHg, an ostium secundum atrial septal defect (ASD) measuring 2 mm, a small muscular ventricular septal defect (VSD) of 2 mm, a small perimembranous VSD of 2 mm, and a tiny patent ductus arteriosus (PDA) of 2 mm, all with left-to-right shunts.

Management

The infant was started on sildenafil (0.5 mg/kg intravenously for 3 days), which was later transitioned to oral sildenafil. Clinically, the infant improved, and repeat echocardiography showed a reduction in pulmonary pressure to 45 mmHg.

Discussion

This case highlights the association between SUA and congenital heart disease (CHD), specifically the presence of multiple cardiac anomalies including ASD, VSD, and PDA, along with significant pulmonary hypertension. SUA is a well-documented marker for congenital anomalies, particularly cardiovascular defects, and is associated with a higher risk of other structural abnormalities. The management of this infant focused on reducing pulmonary pressures, which was successfully achieved with sildenafil.

In conclusion, this report adds to the growing body of evidence that SUA is not merely an isolated umbilical cord anomaly but a significant indicator of potential congenital defects, particularly those involving the cardiovascular system. Early detection and intervention are key to managing the complex clinical scenarios that may arise in such cases.

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