H38 Congenital Hypertrophic Cardiomyopathy in a Neonate: A Rare Etiology for Unexpected Death

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The goal of this presentation is to highlight the necessity for a thorough cardiac examination, prenatal and pediatric medical record review, and discussions with family members when faced with the unusual finding of an enlarged heart in an infant.

This presentation will impact the forensic science community by highlighting the need to consider a broad differential when approaching a hypertrophic heart in the infant population and recognize the presence of posterior rib fractures due to resuscitative efforts.

Full postmortem examinations are critical in sudden unexpected infant deaths. This study presents a rare case of congenital hypertrophic cardiomyopathy with asymmetric hypertrophy, regional myofibrosis with calcifications, and myofiber disarray causing sudden death in a neonate.

A 2-week-old male neonate with no significant past medical history made an unusual crying sound prior to becoming unresponsive while being fed a formula bottle by his biological father. Paramedics responded and found him apneic and asystolic. Resuscitative efforts were continued in route to a local children's hospital with no change in clinical status. His mother had an unremarkable prenatal course with no evidence of diabetes and a normal 19-week anatomy ultrasound. The infant was born at 38 weeks and 0 days gestation via emergent Cesarean section due to fetal decelerations. Birth weight was 3.35kg (25 to 50 percentile) with a head circumference of 35cm (25 to 50 percentile). Initial APGAR scores were 8 and 8 at one and five minutes, respectively. He received supplemental oxygen at birth due to a dusky appearance and responded appropriately. A soft, grade II systolic murmur was heard but resolved prior to hospital discharge. He appeared healthy with no medical issues. Notably, the newborn screen sent at approximately 36 hours of life was within normal limits and he passed the congenital heart disease screen.

Autopsy findings included an enlarged heart with right ventricular enlargement and asymmetric hypertrophy of the interventricular septum (thickness=1.5cm) as compared to the left ventricle (thickness=0.6cm) and an accompanying dilated ductus arteriosus. Multiple regions of white, firm fibrosis were present throughout the myocardium, with the largest measuring up to 1.0cm in the interventricular septum. Microscopically, multifocal hypocellular myofibrosis was present with adjacent calcification and myocyte disarray, seen most prominently in the interventricular septum with extension into adjacent papillary muscles. No additional cardiac anomalies were identified. Other findings included a tetra-lobated right lung. No dysmorphic features were present. Rib fractures were present anterolaterally (right #3-4, left #3-4) and posteriorly (right #3-6, left #2-7) with minimal soft tissue hemorrhage due to cardiopulmonary resuscitation.

This neonate was diagnosed with congenital (infantile) hypertrophic cardiomyopathy. The diagnosis of hypertrophic cardiomyopathy requires left ventricular hypertrophy with a notable absence of other abnormalities to explain the degree of hypertrophy. Infantile hypertrophic cardiomyopathy is extremely rare, with an annual incidence of 3.6 per one million children. Clinical signs are variable and range from presenting with a heart murmur, evidence of heart failure, or sudden death, such as in this case with discovery at the time of autopsy. The etiology for hypertrophic cardiomyopathy is equally heterogenous in the pediatric population and includes glycogen storage diseases, mitochondrial disorders, neuromuscular disorders, certain genetic syndromes (most commonly Noonan syndrome or Beckwith-Wiedemann syndrome), infants born to obese or diabetic mothers, and inherited or *de novo* mutations in the sarcomeric protein genes. No evidence of a storage disorder, syndromic findings with dysmorphic features, or maternal diabetes was ascertained in the clinical history or postmortem examination. Both parents of the infant subsequently have had normal cardiac anatomy on echocardiograms and normal electrocardiograms. Genetic testing has not been performed at this time. This case highlights the need to consider a broad differential when approaching a hypertrophic heart in the infant population and recognize the presence of posterior rib fractures due to resuscitative efforts.

Hypertrophic Cardiomyopathy, Sudden Unexpected Infant Death, Posterior Rib Fractures

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