G41 An Unusual Cause of Sudden Death in Infancy: Hypertrophic Cardiomegaly

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Hypertrophic cardiomegaly in infancy may cause sudden death in infancy. This presentation discusses the clinicopathologic features, describes the gross and microscopic features, and addresses the differential diagnostic features.

Introduction: The number of cases of Sudden Infant Death Syndrome (SIDS) is decreased by an increasing number of cases that can be ascribed to specific medical conditions. A case of the sudden death of an infant is described that autopsy revealed was due to the unusual finding of hypertrophic cardiomegaly in a two-month-old.

Clinical History: The infant was a healthy, full-term twomonth-old white male, who was found unresponsive in his crib following a morning nap. The father began CPR and called emergency personnel. The infant was transported to the hospital, where resuscitative efforts were unsuccessful, and the baby was pronounced dead.

The infant was born to a G2P2 30-year-old woman, who had a history of a motor vehicle accident that resulted in brain trauma. She later underwent temporal lobe resection for control of seizures. During pregnancy, seizure activity required that she was started on levetiracetam (Keppra), beginning at approximately 20 weeks gestation at a dose of 1000 mg per day that was later increased to 1500 mg per day. Towards the end of the pregnancy, she was also started on gabapentin (Neurontin) at a dose of 5400 mg per day. Her pregnancy otherwise was uneventful. At 32 weeks gestation, a second level ultrasound showed no fetal abnormalities, and the heart was normal in size.

Autopsy Findings: At autopsy, the body was that of a well developed and well nourished two-month-old white male, weighing 11 pounds and measuring 23 inches in length. There were no signs of trauma. All internal organs were in their normal anatomic relationships. The heart, in the fresh state, weighed 72 grams (average weight for age was 30 grams) and was structurally unremarkable. Upon sectioning, the myocardium was red/brown, without fibrosis, hemorrhage, or distinct lesions. The interventricular septum was intact. The atria were unremarkable, and no interatrial defects were present. The coronary ostia were normally located, and the distribution showed right dominance with a circulation pattern within normal limits. The valves were thin and unremarkable.

Microscopic Examination: Hypertrophic cardiomyocytes found on H&E sections of the heart. No other microscopic abnormalities were found. Electron microscopy and special histochemical stains were performed.

Discussion: Hypertrophic cardiomegaly in many ways resembles hypertrophic cardiomyopathy. There is left and/or right ventricular hypertrophy that is usually asymmetric and involves the interventricular septum. The hallmark features are myocardial hypertrophy and structural derangement. In this case, the heart was approximately double the normal weight and size.

Neonatal cardiomegaly may resemble hypertrophic cardiomyopathy. However, at autopsy a variety of cardiovascular defects, such as aortic coarctation or malformation of the coronary arteries, are usually found. In addition, in some patients without cardiovascular defects, another cause for ventricular hypertrophy may be present, such as chronic renal failure or maternal diabetes. In this case, no other cardiovascular defects, besides left ventricular hypertrophy, were found, and no additional medical conditions were known.

The differential diagnosis in this case includes amyloidosis, glycogen storage disease, and Fabry disease. These conditions can be eliminated from consideration by microscopic examination of the heart. In this case, sections of the heart showed no abnormal intracellular accumulations.

The etiology of this infant's hypertrophic cardiomegaly is unknown. The parents were concerned that the anticonvulsant medications taken by the mother during pregnancy may have adversely affected the fetus. This question cannot be answered with certainty. Both levetiracetam and gabapentin are pregnancy risk category C drugs, meaning there are no adequate studies of the drugs in humans, although animal studies have shown an adverse effect on the fetus. Category C drugs may be useful in pregnant women despite their potential risk.

It is possible that there may be a familial component to this case. The infant's paternal grandfather has a history of hypertrophic cardiomyopathy.

Conclusion: Hypertrophic cardiomegaly is a rare finding in infants and may be a cause of unexpected sudden death. This condition should be considered in the differential diagnosis of SIDS. A careful examination of the cardiovascular system is warranted in each case. Microscopic and special studies may be used to rule out other causes of cardiomegaly in infants.

Infancy, Hypertrophic Cardiomegaly, Sudden Death

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