CASE REPORT

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Cardiac Rhabdomyoma Presenting as Sudden Infant Death Syndrome

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ABSTRACT: A case of cardiac rhabdomyoma presenting as sudden infant death in a four-and-one-half month-old infant is reported. The child was the product of an essentially uncomplicated pregnancy and enjoyed good health before his unexpected, sudden death. Autopsy examination revealed the presence of multiple cardiac lesions which histologically were diagnosed as rhabdomyomas. Death was attributed to fatal cardiac arrhythmia caused by the tumor. To the authors' knowledge this represents the first reported case in the forensic science literature of death as a result of cardiac rhabdomyoma presenting as sudden infant death syndrome (SIDS).

KEYWORDS: pathology and biology, cardiovascular system, sudden infant death syndrome, cardiac rhabdomyoma

Primary cardiac tumors are extremely rare with a reported incidence of 1 in 10 000 in the general autopsy population [*1*]. The incidence is even less in infancy and childhood, where the most frequently observed primary cardiac tumor is the rhabdomyoma [*2*]. These lesions are generally believed to be hamartomas rather than true neoplasms because they are most often multiple rather than solitary [*3*] and strongly associated with tuberous sclerosis [*3-6*]. Despite the fact that these lesions are biologically benign, patients with these lesions have a rather poor prognosis, with death frequently occurring in the first years of life [*3*]. To our knowledge, there have been no previous reports in the forensic science literature of cardiac rhabdomyoma presenting as sudden infant death syndrome (SIDS).

Case Report

A four-and-one-half month-old male attended by a babysitter was fed and put in his crib for a nap at approximately noon on the date of death. The babysitter checked on the child about 2 1/2 h later to find him breathless and unresponsive. The sitter called for an ambu-

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lance, and paramedics at the scene noted the infant to be cool, cyanotic, and in asystole with no spontaneous respirations. The pupils were dilated and nonreactive. Emergency resuscitation including cardiopulmonary resuscitation (CPR), endotracheal intubation, and 100% oxygen were instituted with the child arriving at a teaching hospital emergency room 23 min after the paramedics received the initial call. At the hospital, 23 min of advanced cardiac life support were unsuccessful, and the infant was pronounced dead. A rectal temperature of 94°F (34.4°C) was obtained in the emergency room.

The infant was the product of a full-term, uncomplicated pregnancy. The child was delivered vaginally at 41 weeks gestation to a serology negative, healthy, married, 26-year-old female. Labor lasted approximately 14 h, and at delivery the infant was noted to have a nuchal cord ×2, however, no resuscitation was necessary. The 1- and 5-min Apgar scores were 5 and 8, respectively. The infant was discharged with his mother on the third hospital day with the only abnormalities noted being bilateral ptosis and a sebaceous nevus on the left cheek. At age 3 months a neurology consult for persistent bilateral ptosis revealed no evidence of myasthenia gravis, and the child was diagnosed as having congenital ptosis. The child was found to be in good health with an otherwise normal physical examination. The child had 2 siblings, aged 2 and 4 years, who enjoy good health.

At autopsy, the infant was normally developed and well-nourished. Indices including heel-crown length and weight, as well as head, chest, and abdominal circumference were within normal limits. Laboratory studies revealed normal vitreous electrolytes. Abnormal findings were confined to the heart upon internal examination.

The heart had a normal weight of 27.5 g and was normally developed with no abnormalities being noted when opened along the flow of blood. Transverse sections through the myocardium revealed a firm, well-circumscribed, tan-white nodular mass measuring 4 by 3 by 3 mm occupying the subepicardial portion of the superior, anterior left ventricular wall adjacent to the ventricular septum. The mid and inferior portions of the ventricular septum contained multiple identical tumor nodules measuring less than 1 mm in greatest diameter each in the subendocardial myocardium of the left and right ventricles. The left and right ventricles showed normal trabeculation with smooth endocardial surfaces. The nodules produced no protrusions into the ventricular chambers. The atria were unremarkable. Light microscopy of hematoxylin and eosin (H&E) sections of the heart showed predominately well-preserved myocardium. Scattered throughout sections of the anterior ventricular wall and ventricular septum were multiple, well-circumscribed, round to oval, nonencapsulated tumor masses (Fig. 1). In some areas these nodules were grouped and located just beneath

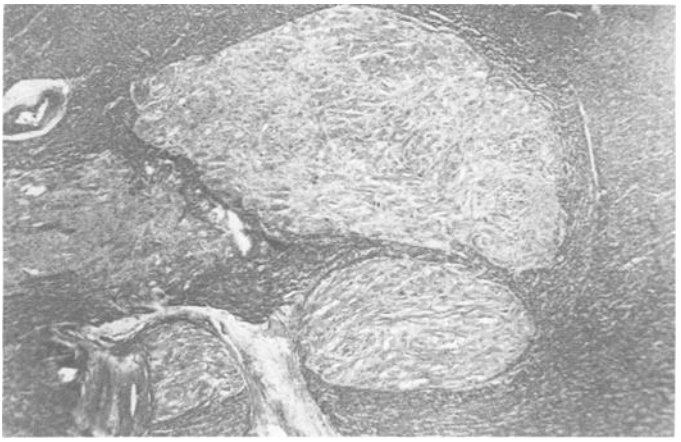

FIG. 1—*Multiple well-circumscribed tumor masses within the ventricular wall (hematoxylin and eosin, ×100).*

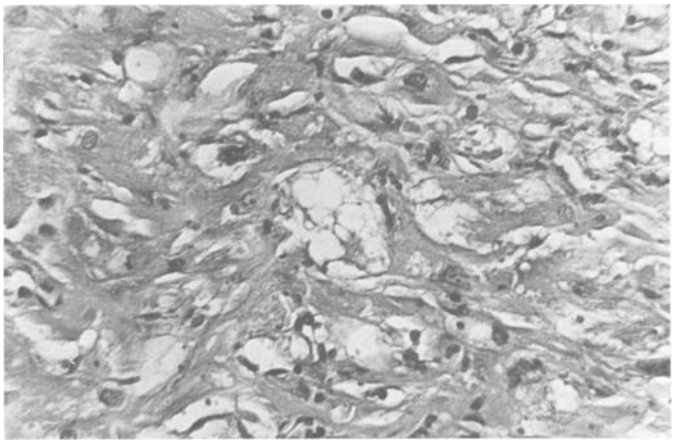

FIG. 2—*Polygonal cells with clear vacuolated cytoplasm within the tumor nodule (hematoxylin and eosin, ×400).*

the endocardium of the left ventricle. In these areas the nodules were separated by thin bands of unremarkable myocardium. The nodules were composed of large polygonal cells with clear vacuolated cytoplasm and uniform nuclei (Fig. 2). Interspersed among these cells were haphazardly arranged, slightly enlarged myocytes with uniform nuclei and fibillary to slightly granular cytoplasm. Rare "spider cells" with centrally located nuclei surrounded by fibillary cytoplasmic fibrils extending to the cell periphery were observed. Cellular features of malignancy were absent, as were mitotic figures. There was no inflammatory infiltrate. Periodic acid-Schiff (PAS) stain revealed focal areas of cytoplasmic positivity within vacuolated tumor cells consistent with the presence of glycogen. A histologic diagnosis of cardiac rhabdomyoma was made.

Discussion

Cardiac rhabdomyoma was first described by Von Rechlinghausen in 1862 [*7*]. Classically, the lesion is composed of multiple distinct masses, one of which is dominant in size [*8*]. It is generally accepted that these lesions are hamartomas rather than true neoplasms, and they are frequently attributed to be but one part of the tuberous sclerosis complex with other developmental abnormalities being found in the skin, brain, and kidneys [*3,5,9*]. Thirty to fifty percent of infants with cardiac rhabdomyoma have tuberous sclerosis [*10*].

Information on the clinical presentation of cardiac rhabdomyomas is scanty, and, unfortunately, a variety of cardiac tumors, as well as some storage diseases of the heart, have been misdiagnosed as rhabdomyomas in the past [*3*]. Most often these tumors are small, intramural, and produce no symptoms with discovery made at autopsy [*4,11*]. However, patients with these lesions have a high mortality rate. A series of 36 patients with cardiac rhabdomyomas revealed a mortality rate of 70% by 1 year of age and 92% by 5 years of age [*3*].

When clinically evident, these tumors have been reported to produce fatal obstruction of the cardiac outflow tracts [*5,11,12*], cyanosis [*11,13*], cardiogenic emboli [*14*], or cardiac failure resulting from myocardial involvement [*11-13*], or some combination of these. These lesions have also been reported to produce a variety of potentially fatal cardiac arrhythmias including right bundle branch block [*15*], ventricular preexcitation syndrome [*16*], ventricular tachycardia [*17,18*], nodal extrasystoles [*18,19*], and heart block with atrial flutter [*18,20*]. Interestingly, the site of the lesion does not always correlate with the arrhythmia [*3,18*].

In 1980, Bohm and Krebs [4] reported a case of an eleven-month-old boy with a clinically silent rhabdomyoma that resulted in a sudden, unexpected death. While actively crawling on the floor, the subject suddenly collapsed and died. In 1981, Violette et al. [21] reported a case of a sudden unexpected death in a six-year-old boy as a result of an asymptomatic cardiac rhabdomyoma. In these cases, both subjects were active before suffering a sudden, unexpected death.

In our case, the subject's death presented initially as a sudden infant death in a four-and-one-half-month-old infant. The diagnosis of sudden death attributable to a rare primary cardiac tumor was made possible only because a thorough and complete postmortem examination was performed. This case serves to point out the absolute necessity of performing such an examination on all infants who die suddenly and unexpectedly. Yet, in 1983, 12% of reported sudden infant deaths had no autopsy performed [22]. In addition, because rhabdomyomas are frequently associated with tuberous sclerosis, a heterofamilial, autosomal dominant inherited disease [23], failure to perform an autopsy would have denied the family the opportunity of seeking appropriate genetic counseling. For these reasons a thorough postmortem examination in cases of suspected sudden infant death cannot be overemphasized.

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