CASE REPORT

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Sudden Unexpected Death Due to Asymptomatic Cardiac Rhabdomyoma

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ABSTRACT: A case of cardiac rhabdomyoma producing sudden unexpected death in an asymptomatic six-year-old boy is reported. Cardiac rhabdomyoma is a rare tumor, and its ability to cause sudden unexpected death in children beyond infancy is not generally known. The great majority of afflicted patients die in early infancy; few survive beyond five years of age. The signs and symptoms present in most cases are said to be due to interference of intracardiac blood flow or to interruption of the cardiac conduction system. Numerous cases have been clinically misdiagnosed because of this tumor's ability to mimic other cardiac diseases.

KEYWORDS: pathology and biology, death, cardiovascular system, rhabdomyoma

Cardiac rhabdomyoma is a distinctly rare tumor, especially when one considers that cardiac tumors as a group have an incidence between 0.0017 and 0.25% in reported autopsy series [*1,2*]. Forty to fifty percent of cardiac rhabdomyomas are associated with tuberous sclerosis [*3*]. Since many of these tumors are congenital, death usually occurs during the first few days of life; when death occurs later than early childhood, it usually results from associated congenital abnormalities. Less than 15% of diagnosed cases survive beyond five years of age. To our knowledge, there have been no previous reports in the forensic science literature of sudden unexpected death of a child resulting from asymptomatic cardiac rhabdomyoma.

Case Report

A six-year-old, 128.75-cm (51.5-in.) tall, 40.9-kg (90-lb), well-developed, well-nourished white boy in apparent good health returned home from school and ate a tuna fish sandwich. Immediately after eating the sandwich, he complained of a stomach ache and then collapsed next to a couch as he went to lie down. He was cyanotic and was immediately taken to a health center where resuscitation was initiated and continued during transport to a hospital. Continuous electrocardiographic monitoring demonstrated a coarse ventricular fibrillation

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that degenerated to an idioventricular bradycardia. The patient did not respond to epinephrine, lidocaine, or procainamide. Resuscitative measures failed and the boy was pronounced dead 2 h after initial collapse.

The deceased was an only child. Family history was positive for febrile convulsions and was negative for mental illness, serious medical illness, and congenital abnormalities. The deceased's medical history was unremarkable except for a single episode of febrile convulsions at eight months of age, when he had a temperature of 40°C (104°F), generalized tonic-clonic movements, flushed skin, and no arrhythmia.

Postmortem examination revealed a lean white boy with equally dilated pupils, slight bilateral scleral congestion, and pale nail beds.

The heart weighed 200 g (mean for body length plus or minus two standard deviations is 119 ± 22 g) [*4*]. The coronary arteries originated normally and were widely patent on serial sectioning. The atrial and left ventricular chambers were normal. Exploration of the right ventricle demonstrated a bulky interventricular septum protruding into the apical half of the right ventricular cavity (Fig. 1). The basal interventricular septum was 0.9 cm thick while the apical septum was 2.1 cm thick. Incision of the septum revealed an intramural, well-circumscribed, nonencapsulated, 3.0- by 2.5- by 2.0-cm multinodular mass of glistening, bulging, soft, pale tan tissue containing focal areas of recent hemorrhage (Fig. 2). The basilar border of the tumor was 2.0 cm inferior to the membranous septum. The myocardium of the basal interventricular septum was congested; the remainder of the myocardium was unremarkable. The left ventricular free wall was 1.0 cm thick. The anterior mitral valve leaflet and proximal aorta demonstrated a few fatty streaks. The remaining valves were within normal limits.

FIG. 1—*View of right side of interventricular septum, showing subendocardial tumor mass protruding into lower half of right ventricle.*

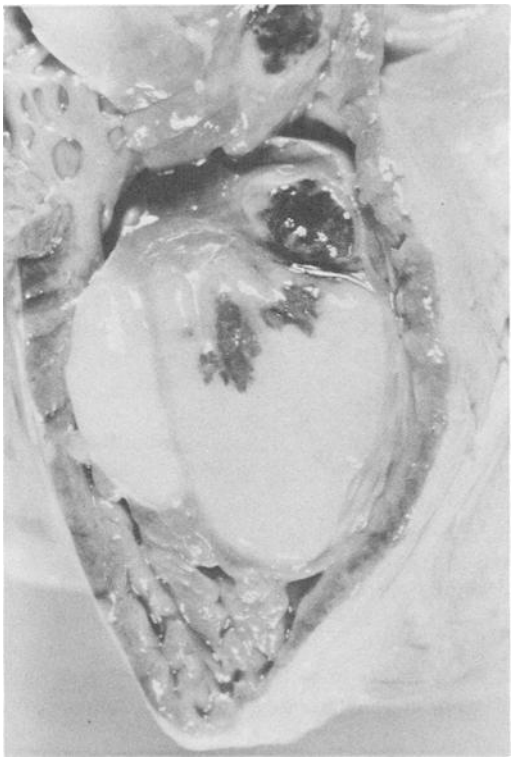

FIG. 2—*Multinodular tumor mass with focal hemorrhage, occupying apical portion of interventricular septum.*

The lungs were congested and edematous, weighing 250 g left and 275 g right (normal for body length is 152 g left and 174 g right [*4*]). Brown gastric contents were found to the level of the tertiary bronchi. The thymus was unremarkable. The liver was congested, weighing 1150 g (normal for body length is 736 g [*4*]). The spleen, remainder of the gastrointestinal tract, urinary tract, adrenals, and cervical organs were unremarkable.

The brain weighed 1650 g (normal for body length is 1273 g [*4*]) and was without evidence of hemorrhage. Uncal grooving was present, greater on the left than on the right. No tonsillar herniation was seen. The vessels of the circle of Willis were unremarkable. Multiple coronal sections of cerebrum, cerebellum, and brain stem demonstrated vascular congestion without other neuroanatomic abnormality.

Microscopic Findings

Microscopically, the tumor in the interventricular septum was composed of nodules of empty-appearing large cells with predominantly eccentric, moderately pleomorphic nuclei (Fig. 3). The tumor mass was generally well demarcated but not encapsulated; several borders were indefinite and showed insertion of tumor cells between myofibers. A few classically described "spider cells" were randomly distributed throughout the tumor mass. There was a prominent vascular stroma without significant fibrosis. Centrally, small foci of recent hemorrhagic necrosis were present within the tumor. Hypereosinophilia and waviness of myofibers, sometimes associated with contraction bands, were seen in multiple sections of myocardium predominantly in subendocardial regions. No margination of neutrophils was

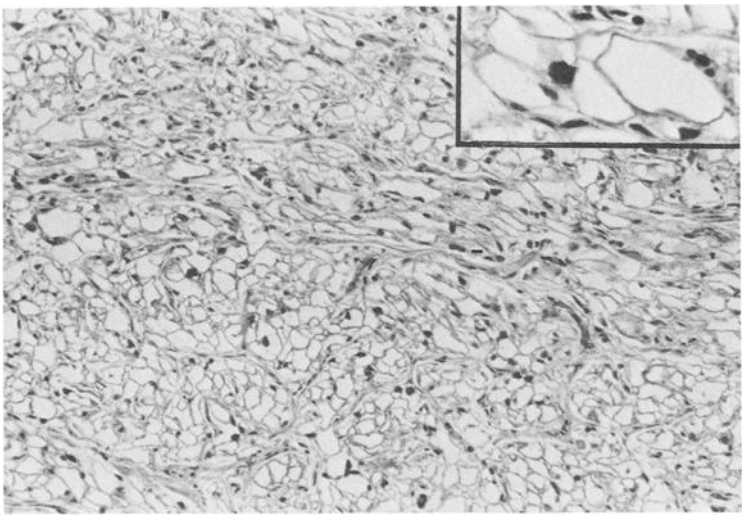

FIG. 3—*Microscopic view of tumor with characteristic "empty-appearing" cells with eccentric, often pleomorphic nuclei and (inset) occasional "spider" cells (hematoxylin and eosin stain).*

seen. These changes did not appear to be limited to the region of the tumor. Sections from the region of the high interventricular conduction system did not reveal tumor or other abnormality.

Sections of the lungs exhibited congestion, intra-alveolar edema, and food particles (meat fragments) in tertiary bronchi with postmortem overgrowth. The liver and kidneys were congested. The brain was congested and showed hypereosinophilia of scattered neurons in the hippocampus, consistent with anoxic change.

Discussion

Cardiac rhabdomyoma was first described by von Rechlinghausen in 1862 [5]. Although rare, it is the most common of the childhood cardiac tumors [6]. There is a male predominance with a ratio of approximately 2:1. The mortality rate is high: 40% of affected individuals die within the first six months of life; by the end of four years, 92% of patients are dead [7].

Signs and symptoms can usually be attributed to interference with the cardiac conduction system or obstruction of the intercardiac blood flow. Syncope, dyspnea, cyanosis, congestive heart failure, and systolic murmurs of varying intensity have been reported [2,4,6,8]. Clinically, cardiac rhabdomyomas may mimic subaortic stenosis [9], pulmonary stenosis [10], tricuspid atresia [11], and tetralogy of Fallot [12]. Patients with a cardiac rhabdomyoma have a predilection to arrhythmias, especially of the atrial type: premature atrial contractions, atrial tachycardia, atrial flutter, or atrial fibrillation. Ventricular preexcitation syndrome has been reported [12], as has fatal ventricular tachycardia [13]. Although these patients may die suddenly, they are usually symptomatic. In our review of the medical literature (*Index Medicus* from 1952 to present) we could find no cases of sudden unexpected death in asymptomatic patients.

The ultimate mechanism of death in this case was ventricular arrhythmia as demonstrated by monitoring by electrocardiography during attempted resuscitation. Several possible causes for a ventricular arrhythmia may be postulated. First, the tumor mass, by protruding

into the right ventricular cavity, could have caused obstruction of the right ventricular outflow tract, leading to hypoxia and, thereby, to an arrhythmia. However, the tumor protruded only into the apical half of the right ventricle and could not be demonstrated to compromise the right ventricular outflow tract, even with manipulation of the heart at autopsy. Second, the tumor could have directly interfered with cardiac conduction, but the tumor came no closer than 2 cm to the membranous septum and did not impinge directly on the atrioventricular node or the bundle of His: sections from this area showed no tumor or abnormality of the high ventricular conduction system. Right bundle branch block might have been expected antemortem but was never documented clinically in this asymptomatic child. Third, a more likely possibility, we think, is that because of local pressure by the tumor the myocardium immediately adjacent to the tumor became ischemic and therefore caused the arrhythmia. As expected because of the short interval between onset of symptoms and death, we found no unequivocal anatomic changes to either support or refute this possibility.

The amounts of hemorrhage in the tumor (Fig. 2) were too small to have expanded the tumor significantly and are probably secondary to resuscitation attempts.

The acute myofibril necrosis and contraction bands are consistent with either (1) ischemia resulting from hypotension following cardiac arrest, (2) catecholamine-induced myofibrillar degeneration [*14*], or both. In either case, these changes are recent and entirely consistent with development during the resuscitative period.

Cardiac rhabdomyomas were previously believed to be a manifestation of a localized glycogen storage disease [*15*] or derivatives of Purkinje's fibers [*16*] or mycocardial cells. Recent ultrastructural studies by Fenoglio et al [*7*] at the Armed Forces Institute of Pathology now suggest that these tumors evolve from cardiac myoblasts prior to the development of T-tubules and should be regarded as hamartomatous rather than as neoplastic in nature.

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