G76 Cause of Sudden Death Due to Cardiac Rhabdomyoma in an 11-Month-Old Baby

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After attending this presentation, attendees will become familiar with the possibility that a completely asymptomatic benign cardiac tumor may induce sudden death in a previously healthy infant.

This presentation will impact the forensic science community by making attendees aware of the insidious development of benign cardiac tumors also in infants and children, focusing the possible responsible mechanisms of sudden death in such cases and providing a reference for additional study on these subjects.

Neoplasms of the heart can be characterized as primary and secondary. Primary cardiac neoplasms occur infrequently in both adult and pediatric age groups. In the general population, their incidence ranges between 0.17% and 0.19% in unselected autopsy series. In infants and children, cardiac tumors were reported at a frequency of 0.027%. Approximately 75% of primary cardiac tumors are benign, and 25% are malignant, in the general population. Benign lesions usually predominate, making up more than 90% of all pediatric tumors. Approximately 50% of the benign tumors are myxomas, and about 75% of the malignant tumors are sarcomas.

Rhabdomyoma is the most frequently occurring cardiac tumor in children. It usually presents during the first few days after birth. It is associated strongly with tuberous sclerosis, a hereditary disorder characterized by hamartomas in various organs, epilepsy, mental deficiency, and sebaceous adenomas. Fifty percent of patients with tuberous sclerosis have rhabdomyoma, but more than 50% of patients with rhabdomyoma have or will develop tuberous sclerosis. The exceptional patient is one with a solitary, single rhabdomyoma who does not have or develop tuberous sclerosis.

Over 90% of rhabdomyomas are multiple and occur with approximately equal frequency in both ventricles. The atrium is involved in fewer than 30% of patients. Pathologically, these tumors are firm, gray, and nodular and tend to project into the ventricular cavity. Micrographs show myocytes of twice normal size filled with glycogen and containing hyperchromatic nuclei and eosinophilic-staining cytoplasmic granules. Scattered bundles of myofibrils can be seen within cells by electron microscopy.

The most common presentation is heart failure caused by tumor obstruction of cardiac chambers or valvular orifice flow. Clinical findings may mimic valvular or subvalvular stenosis. Arrhythmias, particularly ventricular tachycardia and sudden death, may be a presenting symptom. Atrial tumors may produce atrial arrhythmias. The diagnosis is suggested by clinical features of tuberous sclerosis and is made by echocardiography.

Benign cardiac tumors in childhood have an excellent prognosis when completely excised and appear to have a good short-term prognosis even when excision is incomplete. Symptomatic tumors often are both multiple and extensive, particularly in patients with tuberous sclerosis, who unfortunately, have a dismal long-term outlook. In such circumstances, surgery offers little benefit.

Case Report: A mother was bathing her 11-month-old baby. Suddenly the infant showed a worsening dyspnoea. Parents accompanied the baby to the emergency room immediately, but despite the reanimation manoeuvres, the doctor could only pronounce the death. The infant had a negative obstetric, remote and recent pathological

anamnesis, except for a documented fall two days before. Also the familiar history was negative for sudden death.

A complete postmortem examination was performed within 48 hours after death. The body was that of a regularly developed 11-month- old infant. External examination was insignificant, except for the presence of a little and superficial wound on the sternal region.

The internal examination revealed a peduncolated mass at the cardiac apex, a second superficial subepicardial neoformation at the posterior wall of the left ventricle and a third transmural nodule of the posterior wall of the left ventricle. A polyvisceral congestion, cerebral and pulmonary oedema, with a massive increase in lung weight were also evident.

The histological examination of cardiac specimens, stained with haematoxylin–eosin, showed a demarcation and separation of the three masses from the surrounding regular parenchyma. The striated

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muscle cells appeared diffusely vacuolized, enlarged, with round to oval slightly irregular nuclei and variable cytoplasmatic clearing. There were occasional spider cells; muscular tissue residues were also visible. The immunohistochemical studies documented a positive expression of myoglobin, Actin, Vimentin, Desmin, CD34. The result with antibodies Anti-Ki67, -S100 was negative. This microscopic examination was consistent with rhabdomyoma.

Cultural tests and toxicological screening resulted negative. There were no signs of sclerosi tuberosa.

It was concluded that the infant had three cardiac lesions consistent with a primary cardiac tumor, the rhabdomyoma, which caused the sudden death. In particular one tumoral mass occupied almost the whole posterior wall of the left ventricle, rising from the apex to the valvular level, so compromising the regular contraction of the left ventricle. The neoplasms probably had caused two days before a near syncopal episode that the parents erroneously referred as a fall.

Sudden Infant Death, Cardiac Rhabdomyoma, Benign Cardiac Tumors

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