Case Report

Cardiac Rhabdomyoma—A Case Report

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Summary

Cardiac tumors are rare in neonates, most are benign hamartomas (rhabdomyomas) of the muscle cells. Due of large size, they may cause homodynamic instability and even death in neonatal period. They are rarely diagnosed prenatally and are found in multiple forms. In ~50% of the cases, rhabdomyoma is associated with tuberous sclerosis.

Key words: tuberous sclerosis, cardiac rhabdomyoma, orthotropic cardiac transplantation.

Cardiac tumors are rare in neonates, most are rhabdomyomas. Because of large size, they may cause homodynamic instability and even death in neonatal period. In ~50% cases, rhabdomyoma is associated with tuberous sclerosis [1]. About 100 cases have been reported in literature [1, 3]. We are reporting a case of term neonate, with antenatally diagnosed cardiac masses, who expired at 36 h of life and ultimately diagnosed as rhabdomyoma. Other family members were found to be affected with tuberous sclerosis.

Case Report

A term male newborn, a product of nonconsanguineous marriage, born to primigravida at 41 weeks of gestation, whose antenatal ultrasonography at 33 weeks of gestation revealed three avascular echogenic masses in fetal heart, one each in left atrium, left ventricle and right ventricle, measuring 17, 19, 20 mm in diameter, respectively.

The baby cried immediately after birth, physical examination was normal and baby was asymptomatic at birth. However, in view of antenatally diagnosed cardiac masses, baby was admitted in nursery for observation and further management. At 20 h of life, baby developed respiratory distress [Respiratory rate (RR) 68 min⁻¹, nasal flaring and chest retractions], tachycardia [Heart rate (HR) 176 bpm] and prolonged capillary refill time. All the peripheral pulses were palpable, but feeble. On auscultation, there was systolic murmur of grade II. Liver was palpable 4 cm below costal margin in mid-clavicular line.

Chest X-ray revealed cardiomegaly with normal pulmonary vasculature and no other congenital malformation and blood gas was suggestive of metabolic acidosis. Baby was managed for congestive cardiac failure. Baby’s condition continued to worsen despite all supportive measures and baby expired at 36 h of life. Partial autopsy was performed and gross examination of heart showed multiple, well-circumscribed, non-encapsulated and homogeneously solid nodules in left atrium, left ventricle and right ventricle with largest being 3 × 2 cm (Fig. 1).

Histopathological examination revealed well-circumscribed tumors, with normal myofibrils in periphery, centrally placed oval nuclei and eosinophilic granular cytoplasm having thin radiating processes, comprising typical features of ‘SPIDER CELLS’, pathognomonic of rhabdomyoma (Fig. 2).

Family members were examined for stigmata of tuberous sclerosis, because of known association with rhabdomyoma. Father had adenoma sebaceum, axillary freckling and multiple café-au-lait spots suggestive of tuberous sclerosis. Seven-year-old paternal aunt had a history of recurrent seizures since infancy, not responding to anti-epileptic drugs. CT head of both revealed multiple calcified subependymal nodules suggestive of tuberous sclerosis (Fig. 3).

Discussion

Cardiac tumors are very rare in neonates, rhabdomyoma being the commonest, accounting for up to 60% of cases. About 100 cases were described in literature till 1976, with an addition of 10 more cases recently [1]. As many as 50% of children with rhabdomyoma have tuberous sclerosis; 50% of children with tuberous sclerosis have rhabdomyomas [1]. Crawford suggested that prenatal diagnosis of rhabdomyoma indicates that baby may have other postnatal manifestations of tuberous sclerosis [2]. Recently, their spontaneous regression has been
documented, consistent with hamartomatous nature. Farooki suggested that their circumference decreases by \( \frac{2}{24} \) mm month \(^{-1} \) \([3]\).

Rhabdomyomas are benign hamartomas of muscle cells, occur most commonly involving ventricular and septal walls. Rarely, they block circulation within heart causing heart failure \([1, 4]\). Problem may resolve as baby’s heart grows. Extremely rarely, much of the heart muscle is replaced by rhabdomyomas that the heart cannot support circulation independently. Affected babies may be either stillborn or die within few hours. Clinical profile varies from still-birth to intrauterine myocardial infarct (due to coronary artery compression). Most fetuses with rhabdomyomas do well in antenatal period. Occasionally, they cause fetal hydrops \([5]\) or arrhythmias on fetal heart rate monitoring \([6]\) requiring serial ultrasound monitoring. Echocardiography is useful for diagnosis of even small lesions. Delivery at a center, with pediatric cardiology and surgery facilities, is recommended.

After delivery, if no hemodynamic compromise, expectant observation for tumor regression is considered. Serial ultrasound evaluation, MRI and in some cases, angiography may be indicated. Prostaglandin E1, by maintaining patency of the ductus arteriosus, may help to stabilize critically ill newborns with right or left ventricular obstruction \([1]\). Surgery may not be possible because of tumor size or position. Definitive indications for surgery include cardiac outflow obstruction, persistent arrhythmias, cardiac failure and cardiogenic emboli \([7]\). Orthotropic cardiac transplantation is indicated if neonate presents with severe myocardial ischemia \([8]\).

Pathologically, rhabdomyomas are single or multiple, non-capsulated soft lesions. Diffuse rhabdomyomas are rare. Large tumors may show intracavitary extension with almost obliteration of cavity. Microscopically, they consist of discrete masses of large, rounded vacuolated cells predominantly with peripherally placed nuclei (‘spider cells’). Glycogen is present in cytoplasm, concentrated towards periphery \([1, 9]\). Cross-striations are observed in few cells at periphery. Electron microscopy shows myofibrils with \( z \)-bands, and clusters of
leptofibrils with striations at a periodicity of \(\sim 1600\) nm.

Allele loss in hamartomas from patients with tuberous sclerosis, for markers spanning chromosome 16q13.3 in region of TSC2 gene and chromosome 9q34 in region of TSC1 gene, has been found supporting their role as growth suppressor [10]. Mouse model of cardiac rhabdomyoma has been generated by loss of TSC1 gene, which developed dilated cardiomyopathy with enlarged ventricular myocytes similar to spider cells [11].

Well-conducted studies estimate that about two-thirds of cases of tuberous sclerosis are mutations, remaining are familial in autosomal-dominant pattern [1]. For proper diagnosis and genetic counseling, mutation studies should be carried out on affected child. Prenatal diagnosis is possible with DNA technology.

References