SECTION EDITOR: WALTER W. TUNNESSEN, JR, MD

Picture of the Month

N. F. Nik Abdul Rashid, MD; Jayendra Sharma, MD

AN ECHOCARDIOGRAPHIC examination performed on the fetus of a 23-year-old primigravida woman at 36 weeks' gestation revealed multiple densities in the right and left ventricles (**Figure 1**). An earlier fetal echocardiographic examination, performed at 20⅝ weeks' gestation, had normal findings. The mother had received prenatal care throughout her pregnancy, which was without complications or known risk factors. Antenatal screening studies were all normal. The mother's medical history was unremarkable. She took multivitamins and ferrous sulfate regularly and denied ingestion or abuse of alcohol or illicit drugs. Family history was positive for a seizure disorder in a sibling of the mother. There was no family history of fetal death, cardiac or renal diseases, or mental retardation. On physical examination, multiple erythematous papules were noted over the mother's nose and cheeks (**Figure 2**).

The infant was born at term by spontaneous vaginal delivery. An echocardiographic examination confirmed a structurally normal heart with multiple cardiac tumors within both ventricles (**Figure 3**). Findings from Holter monitoring were unremarkable without the appearance of arrhythmias. Head and renal ultrasonographic examination results were normal, without evidence of tumors. A complete eye examination by an ophthalmologist and an examination of the infant's skin revealed no abnormalities.

From the Department of Pediatrics, Lincoln Medical and Mental Health Center, Bronx, NY.

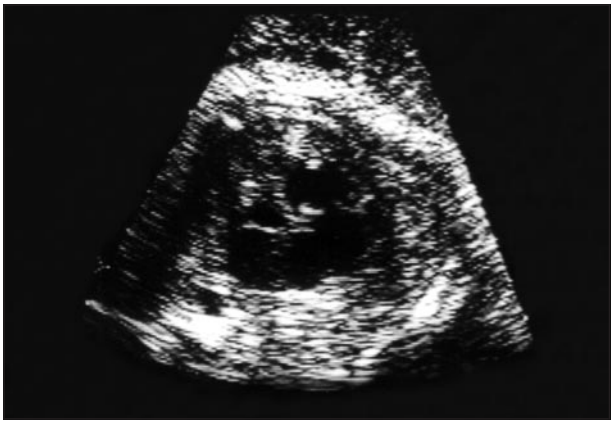

Figure 1.

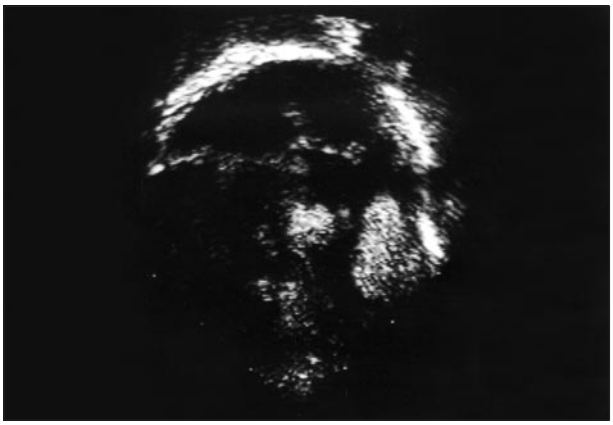

Figure 3.

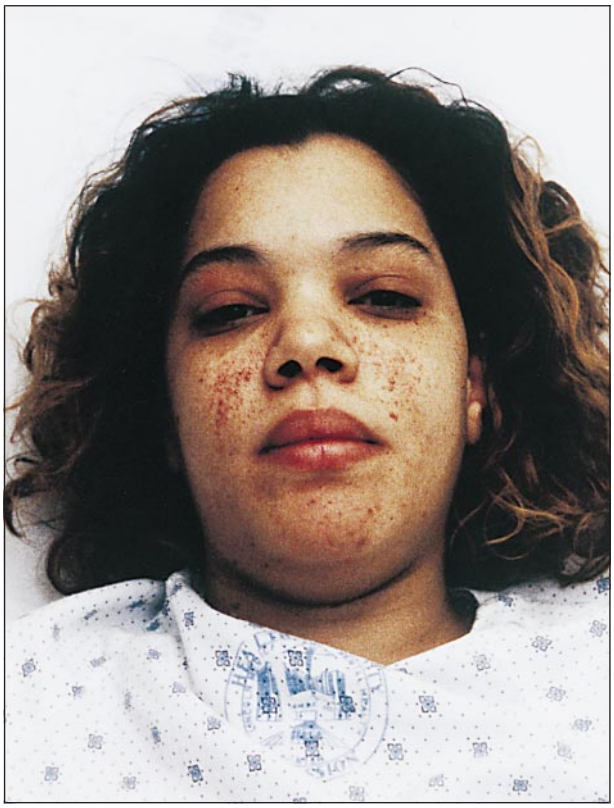

Figure 2.

©2001 American Medical Association. All rights reserved.
Downloaded From: https://jamanetwork.com/ on 08/16/2022

Denouement and Discussion

Cardiac Rhabdomyoma in Tuberous Sclerosis

Figure 1. On echocardiographic examination, a 4-chamber view of the fetal heart shows multiple tumors attached to both sides of the ventricular septum.

Figure 2. Angiofibroma (adenoma sebaceum) are prominent on the mother's face.

Figure 3. The 4-chamber view of the postnatal transthoracic echocardiographic examination shows multiple echodense masses attached to the interventricular septum and left ventricular free wall.

Cardiac rhabdomyoma, while rare at all ages, is the most common primary cardiac tumor of infancy. Although the true incidence of cardiac tumors in the pediatric age group is difficult to ascertain, in 2 hospital series, 0.0017% to 0.003% of pediatric admissions had cardiac tumors.[1] Of these tumors, 75% were rhabdomyomas and teratomas in infants younger than 1 year. Cardiac rhabdomyoma is classified as a hamartoma.[2] The tumors vary in size from a few millimeters to massive tumors that may cause obstruction. The tumors, which may be multiple or single, are most commonly located on the ventricular septum.

CLINICAL MANIFESTATIONS

Cardiac rhabdomyomas may be totally asymptomatic and discovered incidentally on echocardiographic examination or present with severe congestive heart failure, including hydrops fetalis. A large tumor at a vulnerable location, such as the inflow or outflow tract of a ventricle, may cause early symptoms of obstruction to blood flow. Tumors may also present as abnormalities in the conduction system such as ventricular tachycardia, supraventricular tachycardia, and Wolf-Parkinson-White syndrome.[3] Symptomatic rhabdomyomas are associated with fatality rates of 53% in the first week of life and 78% by 1 year.[4]

ASSOCIATION WITH TUBEROUS SCLEROSIS

Cardiac rhabdomyomas are strongly associated with tuberous sclerosis, especially when multiple tumors are present. In one study, 80% of individuals with cardiac rhabdomyomas had tuberous sclerosis, while 60% of children with tuberous sclerosis were documented to have these tumors.[5] If a cardiac rhabdomyoma is found in a fetus or infant with a family history of tuberous sclerosis, it can almost certainly be concluded that the infant may have tuberous sclerosis. If there is no family history of tuberous sclerosis, the presence of a cardiac rhabdomyoma should raise a high suspicion for this disorder.

More than 80% of children with tuberous sclerosis who had cardiac rhabdomyomas documented at birth had no clinical manifestations.[6] Tumor regression or disappearance occurred in 70% of children by age 4 years, whereas only 17% had regression of the tumors after that age.[6] Tuberous sclerosis is inherited as an autosomal dominant disorder with a high rate of sporadic mutation, which accounts for approximately 50% of cases. Two separate genetic mutations may be responsible for the disorder, one on chromosome 9 and the other on chromosome 16. The clinical manifestations of tuberous sclerosis vary widely, and the disorder may not be recognized in mildly affected individuals. Hamartomas occur in a variety of organs in an unpredictable fashion, including the brain, eyes, skin, kidneys, heart, lung, and skeleton, which results in a wide spectrum of signs, symptoms, and complications.

DIAGNOSIS AND TREATMENT

Echocardiography is the most definitive method of diagnosing cardiac tumors in children. This noninvasive test has greatly advanced our knowledge of the incidence, morbidity, and course of cardiac rhabdomyomas, particularly those in association with tuberous sclerosis. Since most cardiac rhabdomyomas in children with tuberous sclerosis spontaneously regress, treatment for these tumors is indicated only if they result in critical blood flow obstruction or arrhythmias. Surgical resection of the tumor may be indicated in symptomatic infants and children in an attempt to reduce morbidity and mortality.[7]

Accepted for publication June 13, 2000.

Reprints: N. F. Nik Abdul Rashid, MD, Department of Pediatrics, Lincoln Medical and Mental Health Center, 234 E 149th St, Bronx, NY 10451.

REFERENCES

1. Ludomirsky A. Cardiac tumors. In: Garson A Jr, Bricker JT, Fisher DJ, Neish SR, eds. *The Science and Practice of Pediatric Cardiology*. 2nd ed. Baltimore, Md: Williams & Wilkins; 1997:1885-1893.
2. Marx GR. Cardiac tumors. In: Emmanouilides GC, Riemenschneider TA, Allen HD, Gutgesell HP, eds. *Heart Disease in Infants, Children, and Adolescents*. 5th ed. Baltimore, Md: Williams & Wilkins; 1995:1773-1785.
3. Jayakar PB, Stanwick RS, Seshia SS. Tuberous sclerosis and Wolf-Parkinson-White syndrome. *J Pediatr*. 1986;108:259-260.
4. Fenoglio JJ Jr, McAllister HA Jr, Ferrans V. Cardiac rhabdomyoma: a clinicopathologic and electron microscopic study. *Am J Cardiol*. 1976;38:241-251.
5. Webb DW, Thomas RD, Osborne JP. Cardiac rhabdomyoma and their association with tuberous sclerosis. *Arch Dis Child*. 1993;68:367-370.
6. Nir A, Tajik AJ, Freeman WK, et al. Tuberous sclerosis and cardiac rhabdomyoma. *Am J Cardiol*. 1995;76:419-421.
7. Murphy MC, Sweeney MS, Putnam JB, et al. Surgical treatment of cardiac tumors: a 25 year experience. *Ann Thoracic Surg*. 1990;49:612-618.

New Feature: Clinical Problem Solving

The Editor is seeking submissions for a new feature, *Clinical Problem Solving*, which will combine *Picture of the Month*, *Radiological Case of the Month*, and *Pathological Case of the Month*. Our aim is to demonstrate the thinking process of a master clinician involved in approaching a patient with an unknown disease. The discussion of such cases should place the clinician's expertise into the context of the prevailing medical literature on the topic. Manuscripts should be between 3000 and 4000 words and may include photographs and radiographs.

©2001 American Medical Association. All rights reserved.
Downloaded From: https://jamanetwork.com/ on 08/16/2022