

A case of tuberous sclerosis presenting with dysrhythmia in the first day of life

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Cardiac rhabdomyoma (CR) is the most common primary cardiac tumor in childhood. Although CRs are asymptomatic in many cases, they may cause arrhythmia, heart failure and fetal hydrops. Babies with arrhythmia in the neonatal period must be investigated for structural heart disease including CR. Cardiac rhabdomyoma may either present as an isolated tumor or may be related with tuberous sclerosis. Arrhythmia due to CR may be the initial sign of tuberous sclerosis. We report a case of tuberous sclerosis presenting with ventricular premature beats and second-degree atrioventricular block in the first day of life who was found to have multiple CR during echocardiographic examination.

Key words: arrhythmia in neonates, cardiac tumor, rhabdomyoma, tuberous sclerosis.

Cardiac rhythm abnormalities are not rare in neonates and may be detected in healthy neonates; however, the infants with dysrhythmia must be evaluated for structural heart disease, fetal distress or infection¹. Cardiac tumors are another cause of arrhythmia, and cardiac detected by echocardiography, and about 50% of CR are related with tuberous sclerosis^{2,3}. The cranial tubers found in cranial imaging studies may be helpful in diagnosis of tuberous sclerosis in such cases⁴. We report a neonate presenting with dysrhythmia in the first day of life and found to have CR due to tuberous sclerosis.

Case Report

ED, a one-day-old male infant, was born following an uneventful pregnancy. He was the first child of a healthy 25-year-old mother. Family history was negative for cardiac or genetic disorders. His birth was 3,200 g. Cardiology consultation was needed because of irregular rhythm found during routine examination in the first day of life. The physical examination was completely normal except for the irregular heart sounds. No murmurs were present on cardiac auscultation. The size and configuration of the

heart was normal on the chest X-ray. Electrocardiogram showed frequent atrial and ventricular premature beats (Fig. 1) and low-grade second-degree atrioventricular (AV) block. The echocardiographic examination showed multiple hyperechogenic masses, 1-2 mm in diameter, within the left ventricular myocardium, and involving the lateral wall, interventricular septum and left ventricular outflow tract; they were presumed to be rhabdomyoma (Fig. 2). Propranolol 2 mg/kg/day was started for



Fig. 1. The electrocardiogram showing the non-conducted atrial premature beats and ventricular premature beats.



Fig. 2. The multiple rhabdomyomas within the interventricular septum demonstrated by two-dimensional echocardiography.

treatment of arrhythmia. The 24-hour Holter monitoring performed both before and under propranolol treatment revealed frequent atrial and ventricular premature beats and low-grade second-degree AV block. The cranial computerized tomography (CT) and magnetic resonance imaging (MRI) studies revealed multiple calcified tubers located on the subependymal region of both lateral ventricles and bilateral parietal subcortical white matter (Figs. 3a, 3b). Electroencephalography showed multiple epileptic foci, and vigabatrin was started. Tuberous sclerosis was diagnosed with these findings. Two hypopigmented patchy lesions on the left ankle located dorsally and on the back of the patient were found during the follow-up

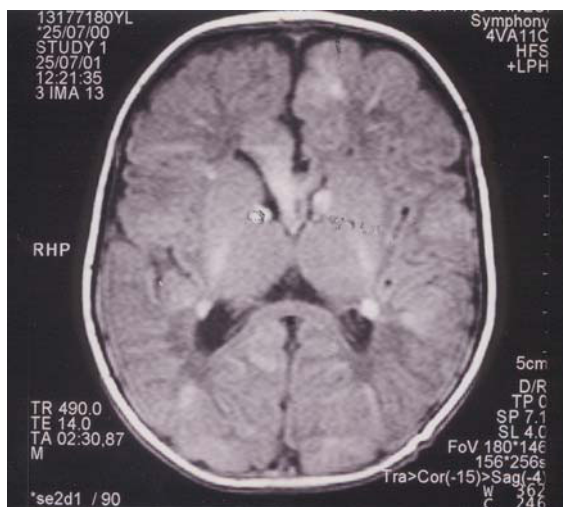


Fig. 3a. Subependymal and subcortical multiple tubers demonstrated by T1 weighted axial cranial magnetic resonance imaging.



Fig. 3b. Subependymal calcified tubers on the left lateral ventricle demonstrated by cranial computerized tomography.

examination of the baby in the first month; they were not present during initial examination. At the age of nine months, he had attained normal weight and height but neurologic development was retarded. No seizures were observed during the follow-up period; however, the ventricular extra-systole existed on the electrocardiogram.

Discussion

Tuberous sclerosis (TS) is an autosomal dominant multisystem disease and its incidence is 1: 6,000-30,000 in the normal population. Genetic heterogeneity is present and gene locus is found on the 9th and 16th chromosomes, which are both tumor suppressing genes^{2,4}. TS is characterized by hamartomas involving primarily the brain, retina, skin, kidneys, heart and lungs. The specific findings are cortical dysplasia (tubers), subependymal nodules, giant cell astrocytoma, retinal astrocytic hamartoma, and facial angiofibroma, less specific findings are cardiac rhabdomyoma, renal angioepithelioma and angiomyolipoma of lungs, liver, gonads or adrenal glands⁵. Mental retardation and epilepsy are common. Our patient did not show clinical evidence of seizures but epileptic foci were found on EEG. Neuromotor development of the patient was found to be retarded during his follow-up examination, and he still had head lag at six months.

Primary cardiac tumors are rare in childhood. CR are the most common primary cardiac tumors in children³. The number of diagnosed cases with CR is increasing due to the development of non-invasive diagnostic methods. CR are the earliest recognizable hamartomas in TS; they can be detected even during fetal life. CR are found in 50% of patients with TS³; on the other hand, 40-80% of the patients with CR have TS^{6,7}. Early diagnosis of TS was possible by echocardiographic diagnosis of CR in our patient.

Cardiac rhabdomyoma may present with a murmur, arrhythmia, heart failure or exercise intolerance⁷. Heart failure due to left ventricular outflow tract obstruction and severe arrhythmia may cause fetal hydrops. The reported types of dysrhythmia are Wolff-Parkinson-White syndrome, supraventricular tachycardia, atrioventricular block and ventricular tachycardia⁶. CR are composed of embryonal Purkinje cells, are postulated to act as microscopic or macroscopic accessory conduction pathways and may predispose ventricular preexcitation². Serious arrhythmia is rare in neonates but may be found during intra-uterine life. The other causes of dysrhythmia may be congenital cardiac abnormalities, electrolyte disturbances, neonatal asphyxia, and neonatal lupus¹. Echocardiographic evaluation is needed for diagnosis of structural abnormalities in neonates presenting with dysrhythmia.

Conservative approach is recommended in patients with CR since spontaneous regression of the tumor occurs in about 60-100% of cases⁷. Medical or surgical intervention is considered according to the severity of clinical findings. Surgical resection is needed in patients with significant left ventricular outflow obstruction. Radiofrequency ablation is another option in patients with intractable arrhythmia who are responding to medical treatment^{8,9}. Our patient

did not develop life-threatening arrhythmia during follow-up, and we preferred medical treatment with propranolol in order to prevent ventricular tachycardia.

In conclusion, in rhythm problems in neonates, cardiac rhabdomyoma and tuberous sclerosis must be suggested and echocardiographic examination must be performed. Cranial imaging is usually helpful in definitive diagnosis of tuberous sclerosis in such patients.

REFERENCES

1. Fish FA, Benson DW Jr. Disorders of cardiac rhythm and conduction In: Allen HD, Gutgesell HP, Clark EB, Driscoll DJ (eds). *Heart Disease in Infants, Children and Adolescents* (6th ed). Vol 1. Philadelphia: Lippincott Williams & Wilkins; 2001: 482-533.
2. Di Mario FJ Jr, Diana D. Evolution of cardiac rhabdomyoma in tuberous sclerosis complex. *Clin Pediatr (Phil)* 1996; 35: 615-619.
3. Webb DW, Thomas RD, Osborn JP. Cardiac rhabdomyomas and their association with tuberous sclerosis. *Arch Dis Child* 1993; 68: 367-370.
4. O'Callaghan FJ, Osborne JP. Advances in the understanding of tuberous sclerosis. *Arch Dis Child* 2000; 83: 140-142.
5. Stowens D. Congenital malformations of the central nervous system: neurocutaneous syndromes. In: David R (ed). *Child and Adolescent Neurology*. St Louis: Mosby; 1998: 419-440.
6. Krasuski RA, Hesselson AB, Landolfo KP, Ellington KJ, Bashore TM. Cardiac rhabdomyoma in an adult patient presenting with ventricular arrhythmia. *Chest* 2000; 118: 1217-1221.
7. Beghetti M, Gow RM, Haney I, Mawson J, Williams WG, Freedom RM. Valvular and congenital heart disease. Pediatric benign cardiac tumors: a 15-year review. *Am Heart J* 1997; 134: 1107-1114.
8. Enbergs A, Boorggreffe M, Kurlemann G, et al. Ventricular tachycardia caused by cardiac rhabdomyoma in a young adult with tuberous sclerosis. *Am Heart J* 1996; 132: 1263-1265.
9. Kohli V, Mangru N, Pearse LA, Cantwell P, Young ML. Radiofrequency ablation of ventricular tachycardia in an infant with cardiac tumors. *Am Heart J* 1996; 132: 198-200.