ANTENATAL ULTRASOUND DIAGNOSIS OF CARDIAC RHABDOMYOMA IN ONE OF THE TWINS

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Introduction

Primary cardiac tumours in foetus are extremely rare. Rhabdomyoma is the most common tumour and has characteristic echocardiographic findings. About 50% are associated with tuberous sclerosis. A case of cardiac rhabdomyoma in one of the twins is presented. No such case has been reported in the English medical literature to the best of our knowledge.

Case Report

A 23-year old primigravida underwent a routine antenatal ultrasound (US) examination for the first time in the third trimester. It revealed a dizygotic twin pregnancy of 31 weeks gestation (a male foetus in vertex and a female foetus in breech presentation). In the male foetus a 3.5 x 3.3cm sized echogenic mass was found intimately related to the cardiac musculature, in the region of the interventricular septum (Fig 1).

There was no evidence of any other obvious congenital anomaly. The female foetus did not show any cardiac mass or congenital anomaly in major organ systems. The patient was advised weekly follow up examination for the foetal cardiac mass and

reviewed for four weeks. There was no change in the size of the foetal cardiac mass over this period. However the patient discontinued review US examination and proceeded uneventfully to normal delivery at term. She delivered a normal female baby and a stillborn male baby. The parents refused autopsy. Postnatal echocardiography of the female baby did not reveal any cardiac abnormality. No evidence of tuberous sclerosis was noticed in either of the parents.

Discussion

Cardiac rhabdomyoma is the most common primary cardiac tumour in infancy and childhood [1]. The neoplastic nature of cardiac rhabdomyoma is controversial with many workers considering this to be a malformation [2]. Strong association between cardiac rhabdomyoma and tuberous sclerosis is consistent with this concept [3]. Cardiac rhabdomyoma was first described by Von Recklinghausen in 1862 and occurs in about 50 percent of patients affected with tuberous sclerosis. Tuberous sclerosis is an autosomal dominant condition with high penetrance and an estimated prevalence between 1 in 15,000 and 1 in 30,000 with a slight male preponderance, 50 to 80 percent of those affected seem to have resulted from a new mutation.

Cardiac rhabdomyomas are frequently multiple and lobulated. On echocardiography they are seen as one or more echogenic masses intimately related to the cardiac musculature. They arise most often in the interventricular septum but some 30 percent will have involvement of the atrial wall [4].

Histological proof of the diagnosis of rhabdomyoma is no longer considered necessary in the presence of characteristic echocardiographic findings [5]. The advent and widespread use of 2D echocardiography and more recently the increasing application of US for screening of foetal cardiac anomalies have undoubtedly led to earlier and more frequent diagnosis of cardiac rhabdomyoma in the asymptomatic

Fig. 1 : Antenatal ultrasound of the thorax of one of the twins showing a 3.5 x 3.3 cm size echogenic mass in the region of the interventricular septum in four chamber view of the foetal heart.

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cases [5,6].

Symptoms resulting from cardiac rhabdomyoma are largely due to effects of tumour size or location within the heart. Inflow or outflow obstruction may lead to myocardial dysfunction with resultant congestion or cyanosis. Refractory cardiac arrhythmias and systemic embolisation are other complications. There are conflicting views about the advocacy of early and aggressive surgery in asymptomatic cases by different authors [5,7].

Spontaneous regression of this tumour has been reported [5,8]. Cardiac tumours have been detected antenatally, the earliest recorded being at 20 weeks gestation [9]. To the best of our knowledge no case of cardiac tumour in one of the twins has been reported in the English medical literature.

References

1. Nadas A, Ellison R. Cardiac tumours in infancy. *Am J Cardiol* 1968;21:363-6.
2. Heath D. Pathology of cardiac tumours. *Am J Cardiol* 1968;21:315-27.
3. Saher R, Mintzerr J, Farina M, Alley R, Bishop M. Clinical presentation of rhabdomyoma of the heart in infancy and childhood. *Am J Cardiol* 1972;30:95-103.
4. Fenoglio J, McAllister H, Ferrans V. Cardiac rhabdomyoma: a clinicopathologic and electron microscopic study. *Am J Cardiol* 1976;38:241-51.
5. Alakalay A, Ferry D, Lin B, Fink B, Pomerance J. Spontaneous regression of cardiac rhabdomyoma in tuberous sclerosis. *Clin Ped* 1987;26:532-5.
6. Bini R, Westaby S, Bargerion L, Pacifico A, Kirklin J. Investigation and management of primary cardiac tumours in infants and children. *J Am Coll Cardiol* 1983;2:351-7.
7. Houser S, Forbes N, Stewart S. Rhabdomyoma of the heart: a diagnostic and therapeutic challenge. *Ann Thorac Surg* 1980;29:373-7.
8. John FS, John DD, Jeffrey FS, Robert MF. Natural history of cardiac rhabdomyoma in infancy and childhood. *Am J Cardiol* 1990;66:1247-9.
9. Wallace G, Smith HC, Watson GH, Rimmer S, D'Souza SW. Tuberous sclerosis presenting with fetal and neonatal cardiac tumours. *Archives of disease in childhood* 1990;65:377-9.