Rhabdomyoma of the heart in a newborn infant

Diagnosis by echocardiography

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SUMMARY An intracavitary right ventricular tumour was identified echocardiographically before operation in an infant with a rhabdomyoma of the heart. Necropsy showed associated tuberous sclerosis.

Cardiac tumours occur very rarely in infancy. When present, the commonest of these are rhabdomyoma.¹ Until now, only a few cases of rhabdomyoma have been diagnosed echocardiographically.²⁻⁴ Affected infants with severe involvement usually die as a result of obstruction to blood flow, and thus operation may be undertaken in an attempt at palliation. Tuberous sclerosis is often associated with rhabdomyoma and must be sought.

We are reporting here an infant presenting with right ventricular outflow obstruction caused by rhabdomyoma which was diagnosed by echocardiogram in the first instance. In addition to a discussion of the echocardiographic diagnosis, we also draw attention to the accompanying conduction disturbance, the angiocardigraphic features, and the pathology, including the ultrastructure of this tumour.

Case report

This infant girl was born at term by elective caesarean section. A hydatidiform mole had been

removed from the mother four months before conception on this occasion. During the first three months of pregnancy she was taking dihydroprogesterone. The parents and two sibs were otherwise normal. There was no history of any fits, rashes, or mental retardation in the family. Birthweight was 2920 g. On the second day of life the infant had a cyanotic spell which responded to oxygen. Duskiness was observed at times during the next few days and because further cyanotic attacks occurred, she was referred to the Transvaal Memorial Hospital for Children for assessment. Abnormal physical findings at 9 days of age, apart from a right hip click, were confined to the cardiovascular system. She was moderately cyanosed at rest. The pulses were normal and the heart rate 140 per minute. A parasternal heave of right ventricular hypertrophy was present. The second heart sound was single and was preceded by a grade 2/6 systolic murmur, heard maximally at the second left interspace. A continuous murmur was audible under the left clavicle. There were no signs of heart failure.

An electrocardiogram showed the features of

Fig. 1 Vectorcardiogram (upper panel) and electrocardiogram showing type A Wolff-Parkinson-White syndrome.

H, horizontal; RS, right sagittal; F, frontal.

Note: Standardisation of electrocardiogram 1 cm = 1 mV (V5 and V6 are half standard).

Fig. 2 Scan from tricuspid valve (TV) to aorta (Ao). Tumour mass is seen within area usually occupied by tricuspid valve, and extends into right ventricular outflow tract (RVOT). Left atrium (LA) is normal in size.

Wolff-Parkinson-White syndrome type A (Fig. 1). A vectorcardiogram confirmed this diagnosis and showed poor rightward forces (Fig. 1). The chest x-ray film showed normal situs, right atrial enlargement, a pulmonary bay and lung fields that were possibly oligaeic.

A provisional diagnosis of right ventricular outflow obstruction was made. An echocardiogram (Fig. 2) showed a tumour mass which appeared in the area where the tricuspid valve is usually identified, and which extended into the right ventricular outflow tract. The mass was also seen in the right ventricular cavity. The left ventricle and aorta appeared normal. The septum was not clearly seen because of the probable tumour mass.

At cardiac catheterisation, a gradient of 20 mmHg was found across the right ventricular outflow tract. Right-to-left shunting at atrial level was demonstrated on oximetry. Systemic arterial saturation was 75 per cent (Table). On angiocardiography

(Fig. 3), a mass was seen to indent the right ventricular outflow tract. Left ventriculography also showed some indentation of the outflow tract from the septum. The aorta was seen to feed pulmonary arteries via a persistent ductus arteriosus.

Fig. 3 Right ventriculogram (frontal projection) shows mass indenting posteromedial aspect of ventricle and reducing size of outflow tract.

Table Cardiac catheterisation findings

Site	Pressure (mmHg)	O ₂ saturation (%)
SVC		67, 67
IVC		74
RA	<i>a</i> = 12 <i>v</i> = 10 (<i>m</i> = 10)	71, 72
RV	high	64
	apex	75/0–10
MPA	55/20 (<i>m</i> = 40)	—
PV	—	84, 88
LA	<i>a</i> = 11, <i>v</i> = 11 (<i>m</i> = 8)	—
LV	78/0–10	75

IVC, inferior vena cava; PV, pulmonary vein; LA, left atrium; MPA, main pulmonary artery; RA, right atrium; RV, right ventricle; SVC, superior vena cava.

A right atrial injection confirmed shunting from right to left atrium.

At operation the same evening, on opening the right atrium a large white lobulated tumour mass arising from the right ventricle was seen to involve the tricuspid valve and to bulge into the right atrium. It appeared to originate from the ventricular septum. The tumour was removed piecemeal, with resultant destruction of the tricuspid valve mechanism. The foramen ovale was sutured. Surprisingly the infant came off bypass, but succumbed the following day in a low output state with evidence of gross tricuspid regurgitation.

PATHOLOGY

Fragments of tumour tissue removed at operation

showed the histological features of a cardiac rhabdomyoma.

NECROPSY

The heart was slightly enlarged and deformed by a bulging tumour mass 3 cm in diameter (Fig. 4) which involved the posterior walls of both atria and extended on to the posterior ventricular surface. Microscopy showed that this crossed the atrio-ventricular junction near the atrioventricular node. A second tumour 1 cm in diameter involved the apex of the left ventricle. In addition, many other smaller tumours were present throughout the myocardium. On incision of these masses they were white in colour and were clearly demarcated from the rest of the muscle. Histological section (Fig. 4)

Fig. 4 Histology of the rhabdomyoma (upper panel) showing 'spider cells'. The arrow points to cross-striations seen mainly in the periphery of the cells. Frontal view of the heart (lower left) which has a window cut into the right ventricle. The destroyed tricuspid valve is evident. The arrow points to a tumour mass visible on the surface. The right lower panel shows the posterior aspect of the heart. Double arrows indicate tumour at the apex of the heart, while a single arrow points to tumour that was present on the posterior surface of the atria and which was shown by sectioning to cross the atrioventricular groove close to the atrioventricular node.

showed these areas to consist of discrete masses of large rounded vacuolated cells predominantly with peripherally placed nuclei ('spider cells'). Glycogen was present in the cytoplasm of the 'spider cells' and was concentrated towards the periphery. Cross striations were observed in only a few cells, and when present, were also situated at the periphery of the cells. Electron microscopy showed myofibrils with z-bands, and clusters of leptofibrils with striations at a periodicity of approximately 1600 nm. Glycogen granules were observed in the background.

The brain appeared normal by external appearance, but numerous 'tuberosities' were palpable within the cortex, which on microscopical examination showed diffuse infiltration by large cells with eosinophilic-staining cytoplasm, which were mainly gemistocytic astrocytes, characteristic of tuberous sclerosis.

Apart from the spleen which showed abnormal histiocytes in the red pulp, and the liver, which showed severe portal condensation of collagen containing large abnormal vascular channels, other organs appeared normal.

Discussion

The echocardiographic diagnosis of tumour was easy in this case. Like myxomas, which may have a similar laminar appearance on echocardiography,^{5,6} rhabdomyomas appear to be intracavitary but do not prolapse into the ventricle. This patient and other previously reported cases of right ventricular rhabdomyoma²⁻⁴ all showed a static laminar mass related to the ventricular septum and the cavity of the right ventricle.

Other electrocardiographic abnormalities are common, but the Wolff-Parkinson-White syndrome is unusual in patients with rhabdomyomas,¹ though this abnormality was shown in a case illustrated in a standard paediatric cardiology textbook.⁷ It appears that a glycogen-containing tumour extending from the atria to the ventricles, as in our case, may conceivably constitute an accessory pathway. Paroxysmal atrial tachycardia did not occur in our infant.

Operation in our case was intended to effect a

cure, but as in other cases the tumour was multicentric.^{8,9} Palliation may then be all that is possible. Furthermore, many cases have associated tuberous sclerosis and will succumb to the effects of this anomaly.⁹ In the absence of a family history, the diagnosis of tuberous sclerosis was not made in our case, until the postmortem examination. Even after death, careful examination of the brain may be necessary to establish the diagnosis. Had our patient survived a successful operation, her prognosis would have been determined by the associated tuberous sclerosis.

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