Unusual angiographic appearances of the left ventricle in 2 cases of Pompe's disease (glycogenosis type II)

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SUMMARY The angiographic and haemodynamic findings in 2 cases of Pompe's disease (glycogenosis type II) indicated an abnormal trabecular pattern, not previously reported, on the left ventricular angiogram of both patients. This feature may be helpful in distinguishing Pompe's disease from other forms of myocardial abnormality.

Pompe's disease (glycogenosis type II) is an uncommon inherited abnormality of carbohydrate metabolism, characterised by the accumulation of glycogen in skeletal and cardiac muscle. Though several cases have now been reported, and characteristic electrocardiographic findings have been described, there are relatively few reports of the haemodynamic and angiographic features of the disease.

In this report, 2 cases are described with distinctive angiographic features which may be of value in the differentiation from other forms of myocardial disease.

Case reports

CASE 1

The patient was the fourth child in a family with no history of previously affected children. Poor muscle tone was apparent in the neonatal period and the child was seen at the age of 3 months in a neurology clinic because of delay in gross motor milestones. On examination no abnormality was present in the

central nervous system, apart from severe hypotonia. The heart sounds were normal and there was no cardiac bruit. Subcostal and intercostal recession was present and shortly after admission the child developed signs of congestive cardiac failure. The chest x-ray film showed cardiomegaly (cardiothoracic ratio 0.6) with normal pulmonary vascularity, and the electrocardiographic findings reported in the Table were consistent with those previously reported in this condition (Caddell and Whittemore, 1962; Hohn and Lambert, 1968).

Echocardiogram

The echocardiogram showed considerable thickening of the interventricular septum (1.4 cm) and posterior left ventricular wall (1.0 cm) compared with the normal values (Meyer, 1977). Motion of the interventricular septum and anterior mitral valve leaflet was normal. The left ventricular end-diastolic and end-systolic dimensions were within the normal range at 2.0 cm and 1.4 cm, respectively, and the percentage change in left ventricular dimension in systole calculated by the method of Gutgesell *et al.* (1977) was 30 per cent. This falls at the

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Table Electrocardiographic and haemodynamic data

Electrocardiographic data						Catheterisation data			
Case	Rhythm	P wave	PR(s)	QRS	T wave	RV (mmHg)	MPA (mmHg)	LV (mmHg)	Aorta
1	Sinus	Normal	0.06	Very high voltages across praecordium; Q wave 14 mm in V6, R wave 66 mm	Biphasic in V5 and V6	30/5	30/10	85/7	—
2	Sinus	Very low voltages in all leads	0.06	Very high voltages across praecordium; Q wave 20 mm in V6, R wave 96 mm	Biphasic in V5 and V6	25/3	25/12	70/10	70/40

Fig. Left ventricular angiographic appearances. (A) Left ventricular angiogram in case 1. The unusual trabeculation of the left ventricle is seen. There is no evidence of left ventricular outflow obstruction. (B) Left ventricular angiogram in case 2. The trabeculation at the apex of the left ventricle is unusually prominent, whereas the septal surface is smooth. No evidence of left ventricular outflow obstruction is present.

lower end of the normal range quoted by these authors.

Cardiac catheterisation and angiography

The investigation was carried out at the age of 4

months with local anaesthesia. No evidence of left-to-right shunting was present but the foramen ovale was patent. A continuous withdrawal tracing across the right ventricular outflow tract showed no pressure gradient, and the left ventricular pressure was not raised. Cineangiograms were obtained from the left and right ventricles. The left ventricular wall was considerably thickened and an abnormally prominent trabecular pattern was present (Fig. A). There was no evidence of left ventricular outflow tract obstruction and the coronary arteries were normal. The right ventricle showed a similar abnormal trabecular pattern. Though the end-diastolic volume of the left ventricle was high, in this patient left ventricular function assessed angiographically was only moderately impaired.

The diagnosis of type II glycogenosis was confirmed by assay of leucocyte α 1-4 glucosidase (acid maltase).

CASE 2

The patient is the second child of healthy parents with no history of previously affected children. At the age of 4 months the child was admitted to hospital because of poor feeding and respiratory problems. Physical examination showed obvious hypotonia but no other abnormality in the central nervous system. A grade 2/6 systolic murmur was present at the left sternal border and a third sound was heard at the apex. Evidence of congestive heart failure was present. The chest x-ray film showed considerable cardiomegaly (cardiothoracic ratio 0.7) and the electrocardiographic findings are shown in the Table.

Cardiac catheterisation and angiography

The investigation was performed at the age of 4 months under local anaesthesia. No evidence of left-to-right shunting was found but the foramen ovale was patent. Continuous pressure recording during withdrawal showed no gradients across either the left or right ventricular outflow tracts. Cineangiography from the left ventricle (Fig. B) showed the ventricle to be dilated and poorly functioning. The abnormal trabecular pattern seen in case 1 was again apparent, and the ventricular wall was considerably thickened. No evidence of subaortic stenosis was seen and the coronary arteries were normal.

The diagnosis of type II glycogenosis was confirmed by assay of leucocyte α 1-4 glucosidase.

Discussion

Relatively few patients with type II glycogenosis have been investigated by cardiac catheterisation

and angiography and, among those studied, varying patterns of abnormality have been recorded. Obstruction to left or right ventricular outflow caused by hypertrophy of the interventricular septum has been reported in several cases (Hohn *et al.*, 1965; Rees *et al.*, 1976) but is not universally present. When present, left ventricular outflow obstruction may be severe and surgical treatment has been attempted in 1 case (Ehlers *et al.*, 1962).

The angiographic features of the condition have been described in 6 patients. Ruttenberg and colleagues (1964) described the findings in 2 patients studied by venous angiography, and contrasted the poor left ventricular contractility seen in 1 patient with the relatively good ventricular function seen in the second patient. At necropsy the first patient proved to have associated endocardial fibroelastosis. Hohn *et al.* (1965), Hernandez *et al.* (1966), and Rees *et al.* (1976) each studied single cases in which left ventricular contractility was good. The sixth case, reported by Nihill *et al.* (1970), had a poorly functioning left ventricle, assessed by follow up after a right ventricular angiogram.

Our 2 cases are of particular interest, since an abnormally prominent trabecular pattern was present on the left ventricular angiogram of both patients. Prominent trabeculation has not previously been reported as a feature of the left ventricular angiogram in this condition, but was noted on the right ventricular angiogram of the case described by Hernandez *et al.* (1966) and appears to be present to a lesser extent in the left ventricular angiogram illustrated in the report of Nihill *et al.* (1970). Pathological studies have shown a high incidence of endocardial fibroelastosis in cases of Pompe's disease (Dincsoy *et al.*, 1965). Review of the published cases suggests that those cases with a dilated non-compliant left ventricle are likely to have associated endocardial fibroelastosis, whereas when the ventricle is contracting effectively outflow tract obstruction is more frequent.

During the investigation of infants with pronounced hypotonia and evidence of cardiac abnormality, the presence of distinct trabeculation in the left ventricle should alert the investigator to the possibility of glycogen storage disease of the heart.

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