ENDOCARDIAL FIBRO-ELASTOSIS IN IDENTICAL TWINS

BY

J. L. GREAVES, P. S. W. WILKINS and S. PEARSON

From the Stockton Children's Hospital and the Central Pathological Laboratory, Middlesbrough

(RECEIVED FOR PUBLICATION MAY 3, 1954)

Fibro-elastosis of the endocardium in siblings has been reported on only two occasions, first by Ullrich (1938), who described it in two monovular offspring of male triplets, and secondly by Weinberg and Himelfarb (1943), who described it in two siblings. A further unrecorded example in siblings has also been mentioned to the authors (Bonham Carter, 1954). The condition in identical female twins therefore seems worthy of record.

Case Reports

J.C. and E.C. were identical female twins, blood group O, Rh positive, born normally at home on July 30, 1953. The parents were healthy, the mother aged 25 years and the father aged 28 years. There was one other healthy girl born one year before. The twins were admitted at the age of 18 hours because of poor housing conditions. Birth weights were: J.C., 4 lb. 8 oz. and E.C., 4 lb. 12 oz. No abnormality could be found in either on examination. They thrived satisfactorily. J.C. weighed 5 lb. 1 oz. when discharged at the age of 4 weeks on August 28 and E.C. 5 lb. 9 oz. on discharge on September 14 at 6½ weeks. At no time during this admission was any cardiac anomaly suspected or detected. The babies were seen at out-patients at the age of 12 weeks and were thriving well.

Case 1. J.C. was re-admitted on December 9, 1953, aged 19 weeks, with a history of vomiting feeds for one week following an upper respiratory infection with a running nose and cough. Difficulty in respiration, increased sweating and cyanosis around the mouth had been noticed. On examination the baby was pale and dyspnoeic, with a running nose. The temperature was 100° F. and the pulse 180 per minute, regular. The apex beat could not be located, the heart sounds were muffled but no murmurs were heard. The liver was enlarged three fingerbreadths below the costal margin and there was some rib recession but no sign of pneumonia or oedema. A diagnosis of cardiac failure of uncertain causation was made and the baby was put in an oxygen tent and given digoxin, 0.1 mg. intravenously, and then 0.1 mg. every eight hours orally for 24 hours, followed by 0.1 mg. b.d. for five days. Considerable improvement resulted.

Blood examination showed: R.B.C. 4.5 m. per c.mm., Hb 76% (11.2 g. per 100 ml.), colour index 0.9, W.B.C. 6,300 per c.mm., with a normal differential count. An electrocardiogram showed the heart to be in an indeterminate position with no special ventricular preponderance.

The baby began to improve and was discharged on December 24, 15 days after admission. She was feeding well, and the weight had risen to 10 lb. 8 oz. Ten days later, on January 3, 1954, she had to be re-admitted on account of the recurrence of cardiac failure; temporary relief from digoxin was obtained but she failed to gain weight and refused some of her feeds. Respirations were always rapid and cyanosis frequent. Radiographs of the chest showed a very large globular heart and a barium swallow showed the oesophagus to be displaced posteriorly and to the right due to enlargement of the left atrium.

On February 3, 31 days after her last admission, the baby died in heart failure.

NECROPSY.—The only striking abnormalities were those in the chest. There was a little serous fluid in

FIG. 1.—Case 1: left side of the heart showing white thickened endocardium and dilated chambers.

FIG. 2.—Case 1: right ventricle showing relatively normal appearance of endocardium.

FIG. 3.—Left ventricle from Case 1, $\times 110$, stained with Verhoeff's stain, showing marked thickening and fragmentation of elastic tissue of endocardium.

each pleural sac and also in the pericardial sac. Both lungs were congested and there was a small area of bronchopneumonia at the apex of the right lung. The heart was enlarged and globular (maximum length 5.8 cm., maximum width 5.5 cm.) and weighed 52 g. (normal is 31 g., Coppoletta and Wolbach, 1933). The left ventricle (Fig. 1) was markedly dilated and hypertrophied. The thickness of its wall was 9 mm. and the right ventricle was hypertrophied but not obviously dilated (Fig. 2). Its wall thickness was 3 mm. The foramen ovale and ductus arteriosus were closed and there was no abnormality of the valves. The endocardium of the left side of the heart, both auricle and ventricle, was white and thickened.

HISTOLOGY. Sections of left ventricular muscle and of kidney, liver and spleen were stained for increased glycogen or fat with negative result. The heart muscle fibres were normal and there was no cellular infiltration. The elastin stains revealed a great increase in the number of elastic fibres in the thickened endocardium of the left ventricle, the elastic fibres being short and fragmented (Fig. 3). This appearance was even more striking in the endocardium of the left auricle (Fig. 4). There was also a patchy thickening of the endocardium with increase in elastic tissue in a section taken from the right ventricle.

FIG. 4.—Left auricle from Case 1, $\times 110$, stained to show elastic thickening of endocardium.

Portions of aorta, kidney and spleen were similarly examined to exclude a generalized affection of the vascular tree, but no abnormality was found.

FIG. 5.—Case 2: left side of the heart showing white thickened endocardium.

Case 2. E.C. was admitted on December 28, 1953, with a history of upper respiratory infection for two weeks; dyspnoea and refusal of feeds had been noted for two days. As had been expected from the findings in her twin, cardiac failure was present. She was pale and dyspnoeic, with cyanosis of the mucous membranes. There was some rib recession, the breath sounds being harsh and vesicular, but no râles were heard. The neck veins were slightly distended and the pulse rate was 200 per minute. No cardiac murmurs were heard. The liver was enlarged two fingerbreadths below the costal margin and there was no oedema.

Again there was a good response to digoxin with temporary improvement.

Blood examination showed: Hb 78% (11.5 g. per 100 ml.), W.B.C. 7,400 per c.mm. with a normal differential count. The E.C.G. on admission showed ventricular extra-systoles and that the heart was in an indeterminate position. An E.C.G. three weeks later showed no special ventricular preponderance. Radiographs of the chest and a barium swallow showed an identical appearance to that found in the other twin.

The baby had two further episodes of cardiac failure with temporary improvement from digoxin before dying in cardiac failure at the age of 26½ weeks on February 1, 1954.

NECROPSY. As before, the significant findings were in the chest. The heart was enlarged, its greatest vertical extent being 6 cm. and its greatest width being 5.5 cm. It weighed 48 g. (normal = 32 g.). The foramen ovale and ductus arteriosus were closed. There was no valvular abnormality. Both the ventricles were dilated and hypertrophied, the left ventricular muscle being

10 mm. thick (Fig. 5) and that of the right ventricle 3.5 mm. thick (Fig. 6). The endocardium of the left side of the heart was of a dense white appearance and seemed thickened.

HISTOLOGY. Extensive examination showed that the abnormalities were confined to the endocardium of the left side of the heart which showed a great increase in the number of elastic fibres, the fibres being short and fragmented. Again the left auricular endocardium was that most markedly involved.

Comment

Endocardial fibro-elastosis is reported to be one of the commoner causes of otherwise unexplained cardiac failure and idiopathic cardiac hypertrophy in infants. It is at least as common as idiopathic myocarditis of the Fiedler group and far commoner than glycogen storage disease and anomalies of the coronary arteries (Rosenbaum, Nadas and Neuhauer, 1953). In our experience it is by no means a rare event and we have seen four other cases in the past nine months. Gowing in a recent review confirms this opinion (Gowing, 1953).

The diagnosis should therefore be suspected in any infant presenting with otherwise unexplained cardiac failure, particularly if no murmurs are heard. Partial confirmation can often be obtained by finding left auricular enlargement on x-ray screening. This has already been noted by others (Freer and

FIG. 6.—Case 2: right ventricle showing relatively normal endocardium.

Matheson, 1953). The differentiation from idiopathic myocarditis may, however, be impossible during life. In our experience electrocardiographic examination has not proved as diagnostically helpful as has been suggested (Adams and Katz, 1952).

Once cardiac failure due to endocardial fibro-elastosis has occurred, the clinical course seems to be almost invariably downhill, with temporary remissions, the total duration of symptoms in the majority of cases being under six months (Blumberg and Lyon, 1952).

The concept of the disease has been ably reviewed by others (Gross, 1941; Gowing, 1953). Both writers agree that a developmental anomaly is the most likely cause of the condition and that there is no evidence that infection plays a causative role, although an attack of respiratory or other infection may herald the first onset of cardiac failure. Johnson (1952) suggested that anoxia of the left side of the heart due to premature closure of the foramen ovale accounted for the left side being that mainly involved, and our findings would agree with this idea but cannot be said to prove it since the patients died so long after birth. In our cases, however, there was no evidence of valvular deformity or abnormality of the coronary arteries. In neither of our cases and in none of the other cases seen in this area has there been any evidence of endocarditis or myocardial infection. We feel

that the striking similarity shown by the lesions in these twins' hearts, together with the lack of any signs of inflammation, strongly supports a developmental cause for this condition.

Summary

Endocardial fibro-elastosis in identical twins is reported. Previous reports of its incidence in siblings are referred to.

The condition does not seem so uncommon as has been thought.

We wish to thank Dr. Doyle for the photographs of the hearts, Dr. Stanley Wray for the photomicrographs, Dr. David Glass for x-raying the patients, Dr. J. J. Tillie for permission to mention other cases admitted under his care and Mr. B. Isserlin for kindly translating the German reference.

REFERENCES

- Adams, F. H. and Katz, B. (1952). *J. Pediat.*, **41**, 141.
 Blumberg, R. W. and Lyon, R. A. (1952). *Amer. J. Dis. Child.*, **84**, 291.
 Bonham Carter, R. E. (1954). Personal communication.
 Coppoletta, J. M. and Wolbach, S. B. (1933). *Amer. J. Path.*, **9**, 55.
 Freer, J. L. and Matheson, W. J. (1953). *Archives of Disease in Childhood*, **28**, 284.
 Gowing, N. F. C. (1953). *J. Path. Bact.*, **65**, 13.
 Gross, P. (1941). *Arch. Path., Chicago*, **31**, 163.
 Johnson, F. R. (1952). *Ibid.*, **54**, 237.
 Rosenbaum, H. D., Nadas, A. S. and Neuhauser, E. B. D. (1953). *Amer. J. Dis. Child.*, **86**, 28.
 Ullrich, O. (1938). *Z. menschl. Vererb.-u. KonstitLehre*, **21**, 585.
 Weinberg, T. and Himelfarb, A. J. (1943). *Bull. Johns Hopk. Hosp.*, **72**, 299.