CONGENITAL PULMONARY STENOSIS IN IDENTICAL TWINS

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The occurrence of congenital heart disease in twins is of value in the consideration of the relative importance of hereditary and environmental factors in the aetiology of congenital malformations. Monozygotic and dizygotic twin pairs may be either concordant (both co-twins affected) or discordant (only one twin of the pair affected) for congenital heart disease. Discordance, even in monozygotic twins, is apparently far commoner, though when concordance for congenital heart disease does occur the type of cardiac lesion is usually the same in both members of the pair.

In this paper a pair of monozygotic twin boys with congenital pulmonary valvular stenosis is described.

CASE REPORTS

The twins (Fig. 1), aged 5 years, were first seen in this Cardiac Clinic in April 1962. The mother had been well

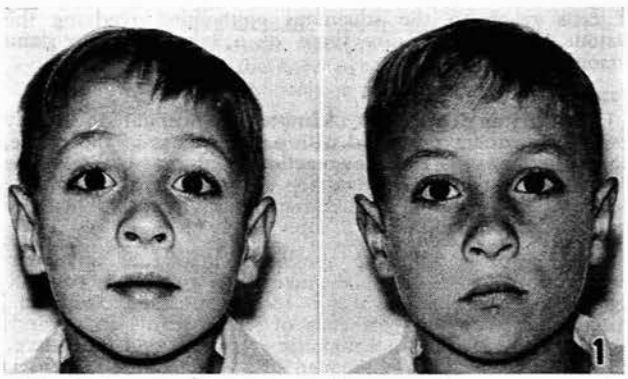

Fig. 1. Showing the very similar general appearances of the twins. Lionel, who had the severer pulmonary stenosis, is on the left.

during the pregnancy. There were no other siblings and no familial history of congenital heart disease. The parents were not related.

Case 1—Alfred

This boy had progressed normally from birth and neither cyanosis nor breathlessness had ever been observed. He was very active and the parents had regarded him as a normal child.

On examination he was found to be well built and had no external congenital stigmata. The pulses were all normal and the blood pressure 100/70 mm.Hg. The jugular venous pressure was not raised, though a slightly prominent 'a' wave was present. There was no clinical evidence of hypertrophy of either ventricle. On auscultation there was a grade-3 pulmonary ejection systolic murmur over the praecordium, which was loudest at the pulmonary area. A physiological third heart sound was present at the apex. The second heart sound was widely split and the pulmonary component was slightly decreased in intensity. A phonocardiogram (Fig. 2) showed this splitting to be 0·03 seconds in expiration and 0·05 seconds in inspiration.

The electrocardiogram (Fig. 2) indicated some right axis deviation (+90°). Upright T waves and an R/S ratio of 1 in leads V_1 and V_4R were compatible with early right ventricular hypertrophy. On fluoroscopic examination some prominence of the main pulmonary artery and slight pulmonary oligaemia were seen. The clinical impression of isolated mild pulmonary valvular stenosis was confirmed at cardiac catheterization, at which the right ventricular pressure was 47/0 mm.Hg, and the pulmonary-artery pressure 22/10 mm.Hg.

Case 2—Lionel

This child had also been regarded as normal until the age of 3 years, when the parents had noted mild cyanosis after exercise. He tired more easily than his brother and was more breathless on effort.

On examination he was extremely similar in general appearances to his twin, but was a pound lighter and an inch shorter. The pulse was normal and the blood pressure 90/60 mm.Hg. A prominent 'a' wave was present in the jugular venous pulse. Palpation of the praecordium revealed a systolic thrill at the second left intercostal space and a heave at the lower sternal area compatible with right ventricular hypertrophy. On auscultation there was a long grade-5 pulmonary ejection systolic murmur and a delayed soft pulmonary component of the second heart sound. Phonocardiography (Fig. 3) showed that the systolic murmur extended past the aortic closure sound and that the soft pulmonary component was delayed 0·05 seconds in expiration and 0·06 seconds in inspiration after the aortic component.

An electrocardiogram (Fig. 3) revealed more right axis deviation (+ 105°) than in his brother and fairly marked right ventricular hypertrophy. A prominent main pulmonary artery and oligaemic lungs were seen on radiological examination. At cardiac catheterization the right ventricular and pulmonary-artery pressures were 100/0 and 18/12 mm.Hg respectively. No intracardiac shunts were demonstrated.

In view of the significant degree of pulmonary stenosis Lionel was referred for surgery in August 1962. At operation (Mr. Paul Marchand), the pulmonary valve orifice was found to be narrowed to ¼ inch in diameter. The commissures were successfully split to the valve ring and, after an uneventful postoperative course, the patient was discharged 3 weeks later.

Evidence for Monozygosity

Apart from the slight disparity in size, which can be accounted for by the severer degree of pulmonary stenosis in case 2 (Lionel), the twins were identical in their general appearances. Blood groupings (total of 8) were performed by Dr. A. Zoutendyk, of the South African Institute for Medical Research, and were the same in both boys. A dermal ridge count of the finger-tip patterns showed a correlation coefficient of 0·92, which is again suggestive of monozygosity.[1]

COMMENTS

Studies of the families of patients with congenital heart disease have shown a greater incidence of congenital cardiac malformations among the siblings or other relatives than in the general population.[2-5] The cardiac malformation has usually been the same or similar in the affected members of the family,[2,4-8] and thus genetic factors have been suggested.[4-6,8] Contrary, however, to what might have been expected as further evidence for a genetic cause, congenital heart disease in twins, whether monozygotic or dizygotic, has much more frequently involved only one member of the pair.[3,9-14] Thus Campbell,[14] discussing the

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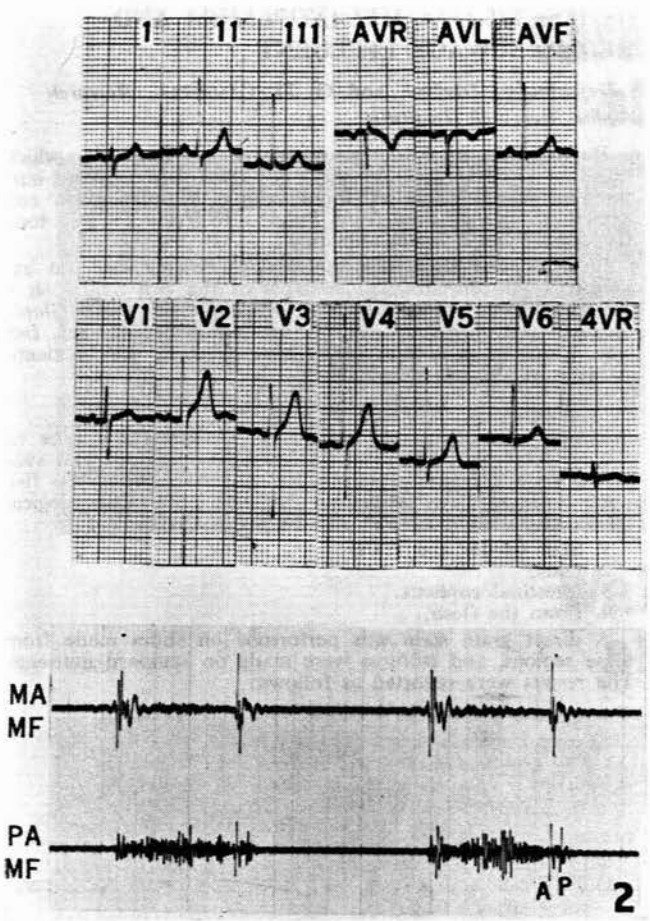

Fig. 2. Electrocardiogram and phonocardiogram of case 1 (Alfred). MA=mitral area; MF=medium frequency; PA=pulmonary area; A=aortic component of second heart sound; P=pulmonary component of second heart sound.

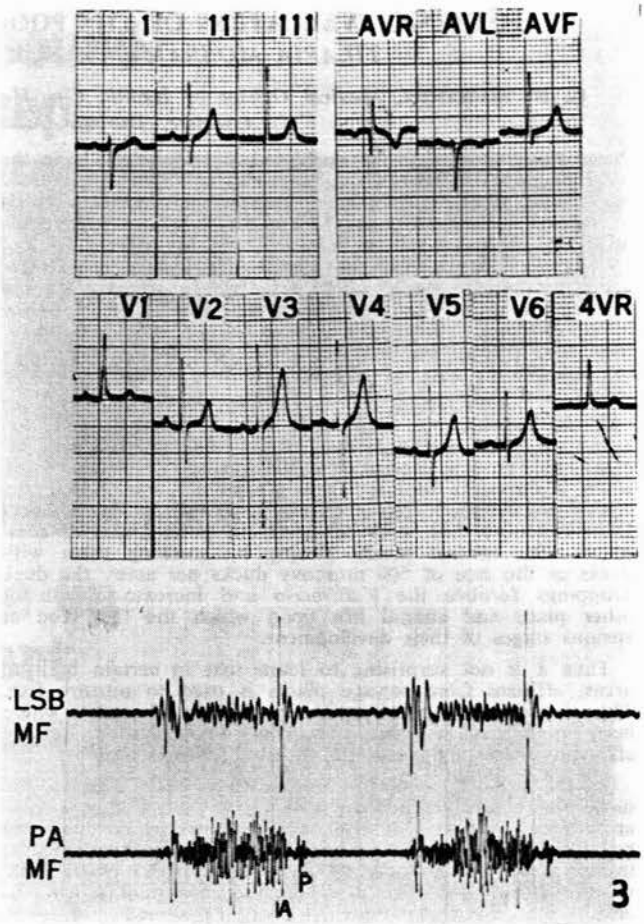

Fig. 3. Electrocardiogram and phonocardiogram of case 2 (Lionel). Interpretation as for Fig. 2. LSB=left sternal border.

factors important in the aetiology of congenital heart disease in twins, concluded as follows:

'The immediate cause of the malformation of the heart must be environmental, though probably there is some genetic predisposition. A disturbance of the foetal circulation to the affected twin because of a single placenta and chorion seems to be the most likely environmental cause.'

Congenital heart lesions occurring in both members of monozygotic twins have occasionally been reported. They include ventricular septal defect,[15] patent ductus arteriosus,[4,12,16] persistent truncus arteriosus,[17] and cor biloculare.[18] Concordant heart lesions in these few instances of monozygotic twins do not, however, necessarily favour a genetic cause, for extrinsic factors acting on both may be responsible. In our twins there was no obvious factor, either genetic or environmental, which we could incriminate as a possible cause of the cardiac anomaly.

SUMMARY

The clinical, electrocardiographic, radiological and catheterization findings of a pair of monozygotic twins with congenital pulmonary valvular stenosis are described. The significance of discordance for congenital heart lesions in monozygotic twins, which has been much more frequently reported than concordance, is briefly discussed.

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