Original articles

Discrete sub-aortic stenosis and ventricular septal defect

L D R SMITH, C CHARALMBOPOULOS, M L RIGBY, S PALLIDES, S HUNTER, C LINCOLN, AND E A SHINEBOURNE

Departments of Paediatric Cardiology, Brompton Hospital, London, and Freedom Hospital, Newcastle

SUMMARY We present details of 15 children, aged 3 months to 11 years, with discrete sub-aortic stenosis and ventricular septal defect. We emphasise a high index of clinical suspicion and echocardiography as the best means of diagnosing this dangerous combination. Physical signs were those of ventricular septal defect in all patients, with auscultatory evidence of additional sub-aortic stenosis in only one. Five patients had a short early diastolic murmur of mild aortic incompetence. The electrocardiograph showed isolated left ventricular hypertrophy in eight patients. Cardiac catheterisation and angiography identified the ventricular septal defect in all cases but detected the sub-aortic stenosis in only eight. Cross sectional echocardiography showed both lesions in all 11 patients to whom it was available.

Ventricular septal defect is reported as occurring in between 6 and 25% of patients with sub-aortic stenosis.^{1–3} Clinical diagnosis of the combination of lesions may be difficult as the physical signs of each may mask the other. At cardiac catheterisation the presence of a ventricular septal defect may obscure haemodynamic evidence of sub-aortic stenosis by producing a lower left ventricular outflow tract gradient.^{4–6} Angiography may fail to disclose the sub-aortic stenosis as the offending membrane or fibro-muscular band may be very thin, and may be obscured by flow of contrast medium through the ventricular septal defect. Cross sectional echocardiography, while probably the optimal diagnostic technique, may still miss discrete fibrous bands unless particular views are taken.⁷

Omitting to make the diagnosis of sub-aortic stenosis in the presence of ventricular septal defect has serious consequences as the effect of the stenosis is likely to progress and may lead to left ventricular hypertrophy, myocardial damage, damaged aortic valve leaflets, heart failure, and death.^{1–3 6 8–10} For these reasons we report the clinical, haemodynamic, angiographic, echocardiographic, and operative findings in a group of patients with discrete sub-aortic stenosis and ventricular septal defect, with particular emphasis on how the diagnosis may best be made before surgery.

Patients and presentation

We present data on 15 children, 9 boys and 6 girls, admitted to the Brompton Hospital, London and the Freeman Hospital, Newcastle. Evidence of congenital heart disease was noted in all patients within the first year of life, and in 13 within the first 3 months, yet the age when diagnosis was confirmed varied from 3 months to 11 years. The mean age at the time of cardiac catheterisation was 3.8 years. The clinical presentation was congestive cardiac failure in 10, murmur in an asymptomatic child in four, and supraventricular tachycardia in one.

Physical findings

All 15 patients had a pansystolic murmur maximal at the left sternal edge in the third to fourth intercostal space; associated with a thrill in 12. In five patients an early diastolic murmur was present, or developed between the initial presentation and the time of diagnosis. Only one patient had an ejection systolic murmur maximal at the second right interspace—the usual murmur of aortic stenosis.

In all 15 patients the clinical features were thought to be typical of a ventricular septal defect but in five the addition of an early diastolic murmur suggested

aortic incompetence. In only six patients was there an increased apical (left ventricular) impulse and in none was the character of the peripheral pulses felt to be abnormal. One of the three patients with additional right ventricular outflow tract obstruction had cyanosis.

Investigations

Radiography. The chest radiograph showed cardiomegaly and pulmonary plethora in 12 patients, isolated cardiomegaly in two, and normal sized heart with normal lungs in one. Atrial situs solitus, laevocardia, and left aortic arch were found in all cases.

Electrocardiography. Sinus rhythm was noted in 14 patients; one patient had intermittent supraventricular tachycardia. The mean frontal QRS axis ranged between -30° and $+100^\circ$. Seven patients had isolated left ventricular hypertrophy, five biventricular hypertrophy, two (with associated right ventricular outflow tract obstruction) isolated right ventricular hypertrophy, and one was normal. Flat left sided T waves were present only in one patient who had right bundle branch block after a previous operation for repair of a ventricular septal defect.

Echocardiography. Cross sectional echocardiography was recorded in 11 patients using an Advanced Technology Laboratories mechanical sector scanner. In each case a sub-aortic ridge or diaphragm was identified together with a ventricular septal defect. Four patients were seen before the availability of cross sectional echocardiography. The sub-aortic stenosis was best identified using left parasternal and apical long axis views of the left ventricular outflow tract and aortic root. In all 11 patients sub-aortic stenosis was seen in the apical long axis view (Fig. 1). In 10 patients a distinct linear echo, parallel and close to the aortic valve, was shown extending from the ventricular septum to the anterior mitral valve leaflet (Fig. 2). In the 11th patient the sub-aortic stenosis was seen to be caused by posterior deviation of the interventricular septum. In this patient the ventricular septal defect was muscular while in the other 10 it was perimembranous.

Cardiac catheterisation.

Haemodynamics

Fourteen patients underwent cardiac catheterisation and angiography—six twice and one three times. In all patients the ventricular septal defect was confirmed at the first or only catheterisation. In contrast, in only four patients was sub-aortic stenosis detected at the first study, while in four others

Fig. 1 Cross sectional echocardiogram using off axis apical four chamber view. The sub-aortic membrane is arrowed.

AO aorta; LA left atrium; LV left ventricle; RV right ventricle; S ventricular septum; VSD ventricular septal defect

Fig. 2 Cross sectional echocardiogram using left parasternal long axis view. The sub-aortic membrane is arrowed. Abbreviations as for Fig. 1.

the sub-aortic stenosis was confirmed at a subsequent investigation. In six patients sub-aortic stenosis was never diagnosed at cardiac catheterisation. The peak systolic gradient across the left ventricular outflow tract at the time of the most recent cardiac catheterisation was available in 12 patients and ranged from 0 to 68 mm Hg. A gradient of 10 mm Hg was recorded in eight patients and in four of these who underwent repeat catheterisation the gradient had increased from a mean of 2.5 mm Hg to a mean of 45.5 mm Hg. A gradient of less than 10 mm Hg was found in two patients and no gradient in

Fig. 3 Aortogram performed in modified long axis projection (see text). Contrast is seen to pass across a small ventricular septal defect situated immediately underneath the aortic valve cusps (↓) and above a discrete sub-aortic membrane (↑).

the remaining two, yet all had surgical findings of a sub-aortic ridge.

The pulmonary to systemic flow ratio was available in 12 patients and ranged from 1.7:1 to 3.8:1. In those patients in whom an increased left ventricular outflow tract gradient was noted at the second cardiac catheterisation, the pulmonary to systemic flow ratio had decreased.

Angiography

An aortogram was performed in 12 of the 14 patients who were catheterised. In three of the four with an early diastolic murmur, aortic regurgitation was detected.

For optimal angiographic visualisation of the sub-aortic membrane and ventricular septal defect, we used a modified long axial view (Fig. 3), with 20° craniocaudal tilt at 65° left anterior oblique.

An apparently discrete sub-aortic obstruction was shown in seven patients, while in one, left ventricular outflow tract obstruction was caused by leftward and posterior deviation of the infundibular septum. In the four patients in whom sub-aortic stenosis was not detected angiographically conventional frontal and lateral projections had been used.

A bicuspid aortic valve was seen in two patients both of whom were also noted to have an aneurysm of the ventricular septum.

Surgery and outcome

Seven of 10 patients who underwent surgery had

closure of ventricular septal defect and resection of sub-aortic stenosis at the same operation. One patient had the sub-aortic stenosis resected four years after closure of ventricular septal defect and this patient had the highest left ventricular outflow tract gradient. The remaining two patients who underwent surgery each had associated coarctation. One has had this resected and is awaiting further surgery, the other presented in infancy with heart failure, underwent resection of coarctation and pulmonary artery banding, and died six months later after repair of ventricular septal defect and debanding of the pulmonary artery. Sub-aortic stenosis was diagnosed at necropsy. This was the only patient who died.

Operative findings. The sub-aortic stenosis found at operation or necropsy (one patient) was membranous in three patients, fibromuscular in six, and was caused by a deviated infundibular septum in one (muscular malalignment defect).

The ventricular septal defect found at operation was perimembranous in eight and muscular in one. The diameter varied from 0.5 cm to the diameter of the aortic annulus. In eight patients the ventricular septal defect was above the sub-aortic stenosis and in one, below. In one patient a large parachute type aneurysm of the membranous septum in close approximation with the ventricular septal defect was imbricated.

An abnormal aortic valve was noted in five patients. Bicuspid valves were present in two but no valvar stenosis was found.

Discussion

Various types of sub-aortic stenosis have been described including membranous, caused by a thin fibrous diaphragm just below the aortic valve; fibromuscular, where a fibromuscular ring is found lower in the left ventricular outflow tract; tunnel stenosis; and that found in hypertrophic obstructive cardiomyopathy. The first two types are considered to constitute discrete sub-aortic stenosis. The variability in the reported incidence of discrete sub-aortic stenosis in isolation and in association with ventricular septal defect is due partly to lack of consistency in the definition of discrete. Variability in the age of populations studied compounds this since a large proportion of ventricular septal defects will close spontaneously.^{1-3 5-6 11} Of 14 000 case notes on patients who have been catheterised at the Brompton Hospital there are no cases of ventricular septal defect and sub-aortic stenosis occurring in the over 18 years age group. In comparable studies in infants, ventricular septal

defect occurs in approximately 20% of patients with discrete sub-aortic stenosis.¹⁻³

Unrelieved sub-aortic stenosis may lead to permanent myocardial damage and left ventricular hypertrophy.² The jet of blood streaming from the sub-aortic stenosis may damage the aortic valve leaflets and lead to regurgitation,¹² and conduction defects may also develop.² The importance of making the diagnosis in association with accompanying ventricular septal defect is thus stressed but failure to do so is well documented.^{1 5 6 13 14}

Clinical diagnosis may be difficult but certain findings or combinations of findings should engender a high index of suspicion. Once suspected clinically, in our view, echocardiographic proof of the diagnosis is readily achieved. The clinical features to stress in an asymptomatic patient with the signs of a small ventricular septal defect, that is a pansystolic murmur at the fourth left intercostal space at the sternal edge and a normal second heart sound, are disproportionate voltage criteria of left ventricular hypertrophy on electrocardiogram or a short early diastolic murmur at the second or third interspace indicating mild aortic regurgitation. The typical ejection systolic murmur of aortic stenosis, maximal at the second right intercostal space, is not usually present, being found in only one patient in our series.

The differential diagnosis in those patients with left ventricular hypertrophy detected on electrocardiogram includes hypertrophic cardiomyopathy. Those patients with early diastolic murmurs should be distinguished from patients with a high ventricular septal defect and aortic incompetence due to prolapse of the right coronary cusp into the defect. Echocardiography will confirm or refute these diagnoses. Cross sectional echocardiography, which is preferable to M mode echocardiography,^{9 10 15 16} can be used to visualise the left ventricular outflow tract throughout the cardiac cycle and may show clear images of the discrete sub-aortic stenosis. The left ventricular outflow tract is best viewed with left parasternal and apical long axis views,⁷ the latter showing the site of sub-aortic stenosis in 100% of our patients to whom it was available.

Cardiac catheterisation and angiography (in long axis projection)¹⁷ are useful to confirm the diagnosis, using the latter to exclude additional septal defects.

An aortogram shows aortic regurgitation and sometimes facilitates the visualisation of the discrete sub-aortic stenosis.

We conclude that while sub-aortic stenosis and ventricular septal defect together are not common, the gravity of overlooking sub-aortic stenosis

warrants constant awareness of this possible combination. Once suspected clinically, however, the diagnosis may be confidently confirmed by the combination of cross sectional echocardiography, cardiac catheterisation, and angiography, perhaps all of which should be used.

References

- Newfeld EA, Muster AJ, Paul MH, Ideiss FS, River WL. Discrete subvalvar aortic stenosis in childhood. Study of 51 patients. *Am J Cardiol* 1976;**38**:535-61.
- Somerville J, Stone S, Ross D. Fate of patients with fixed sub-aortic stenosis after surgical removal. *Br Heart J* 1980;**43**: 629-47.
- Reis RL, Paterson LM, Mason DT, Simon AL, Morrow AG. Congenital fixed subvalvular aortic stenosis. An anatomical classification and correlations with operative results. *Circulation* 1971;**45** Suppl 1:11-8.
- Ben-Schachar G, Moller J, Castaned-Zuniga W, Edwards JE. Signs of membranous sub-aortic stenosis appearing after correction of persistent common atrioventricular canal. *Am J Cardiol* 1981;**47**:340-4.
- Dirksen T, Moulart AJ, Buis-heim TN, Brom AG. Ventricular septal defect associated with left ventricular outflow tract obstruction below the defect. *J Thorac Cardiovasc Surg* 1978;**75**: 688-94.
- Vogel M, Freedom RM, Brand A, Trusler GA, Williams WA, Rowe RD. Ventricular septal defect and sub-aortic stenosis. An analysis of 41 patients. *Am J Cardiol* 1983;**52**:1258-63.
- Di Sessa TG, Hagan AD, Isabel-Jones JB, Ti CC, Mercier JC, Friedman WF. Two-dimensional echocardiographic evaluation of discrete sub-aortic stenosis from the apical long axis view. *Am Heart J* 1981;**101**:773-82.
- Champsour G, Trusler GA, Mustard WT. Congenital discrete subvalvar aortic stenosis. Surgical experience and long-term follow-up in 20 paediatric patients. *Br Heart J* 1973;**35**:443-6.
- Khan MM, Varma MPS, Cleland, J, et al. Discrete sub-aortic stenosis. *Br Heart J* 1981;**46**:421-31.
- Surg CS, Price EC, Cooley ZA. Discrete sub-aortic stenosis in adults. *Am J Cardiol* 1978;**42**:283-90.
- Kelly DT, Wulfsberg E, Rowe RD. Discrete sub-aortic stenosis. *Circulation* 1972;**46**:309-22.
- Billeden LC, Allen HD, Moller JH. Natural history and operative results in discrete membranous sub-aortic stenosis. Surgical experience and long-term follow-up in 20 paediatric patients. *Br Heart J* 1983;**35**:443-6.
- Lauer RM, Du Shane JW, Edwards JE. Obstruction of left ventricular outlet in association with ventricular septal defect. *Circulation* 1960;**22**:110-25.
- Fisher DJ, Snider AR, Silverman NH, Stanger P. Ventricular septal defect with silent discrete subaortic stenosis. *Pediatr Cardiol* 1982;**2**:265-9.
- Berry TE, Aziz KV, Paul MH. Echocardiographic assessment of discrete sub-aortic stenosis in childhood. *Am J Cardiol* 1979;**43**:957-61.
- Kruger SK, French JW, Forkeer AD, Caudill CC, Popp RL. Echocardiography in discrete subaortic stenosis. *Circulation* 1979;**59**:506-13.
- Barger LM, Elliot LP, Soto B, Bean PR, Cowy GG. Axial cine-angiography in congenital heart disease. Section 1. Concept technical and anatomical considerations. *Circulation* 1977;**56**: 1075-83.

Correspondence to Dr E A Shinebourne, Brompton Hospital, London SW3 6HP

Received 3 December 1984