Congenital aortic stenosis—an unusual form

Consideration of surgical management

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Three patients with severe congenital aortic stenosis in whom there was unusual thickening of the aortic valve associated with supra-aortic stenosis and aortic hypoplasia are described. The clinical presentation suggested the diagnosis of organic subaortic stenosis and surgical management presented problems. It is suggested that the aortic stenosis in these patients may be part of a more diffuse abnormality.

Congenital aortic valve stenosis in young patients usually takes the form of a mobile dome with a varying sized central orifice that may be eccentric. The obstruction is not often critical enough to require relief in the first decade of life, though after the period of rapid body growth at puberty aortic valve stenosis frequently requires surgical attention. Out of 32 patients under 20 years with congenital aortic valve obstruction who have had operations at the National Heart Hospital in the past 5 years, 3 children with an unusual type of aortic valve abnormality associated with a supra-aortic obstruction have been treated. Attention is drawn to this variant as it presents difficulty in bedside diagnosis as well as in surgical management.

Reports

Case 1 A girl aged 9 years was the product of a normal pregnancy with a birthweight of 3.17 kg (7 lb). The mother was known to have asymptomatic mitral stenosis during the pregnancy. During the first 7 months the child fed poorly, was slow to gain weight, and had many respiratory infections. At age 10 months a moderate sized persistent duct was ligated and the child improved after this. Cardiac murmurs persisted and the child was followed up regularly at outpatients; at 7 years she was noted to be breathless on effort and the electrocardiogram showed severe left ventricular hypertrophy with sharp T inversion and ST depression in all left ventricular leads.

On examination she appeared to be small and was below the tenth percentile in height but she was mentally normal. There were multiple telangiectases over the face and body. Her eyes were widely spaced and the hairline was low on the neck (Fig. 1). The carrying angle of the elbows

was wide on both sides, there was a slight protuberant sternum, and her appearance resembled the mosaic form of Turner's syndrome.

The pulse was slow rising in the neck where there was an intense systolic thrill. The left ventricular impulse was powerful. On auscultation there was an intense long systolic ejection murmur, single second sound, and a moderate immediate diastolic murmur; a short presystolic

FIG. 1 (a) Face and upper chest of Case 1 showing widely spaced eyes, widely spaced nipples, and right chest protuberance. (b) Back of neck of Case 1 showing low hairline.

FIG. 2 Phonocardiogram from Case 1 showing long systolic ejection, systolic murmur in mitral area (MA). A presystolic murmur is recorded at MA; aortic valve closure is not clearly delineated; the carotid pulse appears anacrotic.

apical murmur was present but no ejection sound was heard or recorded (Fig. 2). The physical signs suggested severe subaortic stenosis with some aortic regurgitation. The chest radiograph confirmed slight cardiomegaly with prominent upper pulmonary vein shadows but the aorta was small and there was no evidence of poststenotic dilatation.

Investigation with aortography showed an unusually small aorta with a slight supra-ventricular narrowing, and a grossly disordered thickened aortic valve with aortic regurgitation into a curious sac-like structure (Fig. 3). The catheter could not be passed across the supra-ventricular and valve obstruction in spite of numerous attempts. A gradient of 200 mmHg was recorded across the pulmonary valve. Late renal films showed a left-sided double kidney with two ureters.

In view of the severity of electrocardiographic changes and the presence of dyspnoea, an operation was recommended, which was undertaken on 28 February 1969 (D.N.R.). The external appearance of the heart was abnormal with a sharp pointed apex and a disproportionately large left coronary artery. A gradient of 120 mmHg was found across the left ventricular outflow tract. An incision into the narrow ascending aorta re-

FIG. 3 Aortogram from Case 1. (a) Anteroposterior view. (b) Lateral view. Thickened aortic valve cusp remnants are shown by arrow. The left coronary artery is large and a supra-aortic valve narrowing is present. Considerable aortic regurgitation into a left ventricular pouch and into the body of the left ventricle has occurred. A small subvalvar shelf subsequently resected is visible under the valve in the anterior part of the lateral view.

(a)

(b)

vealed a small orifice about 4 mm in diameter surrounded by a pink thick endothelium. Excision of this ridge of tissue exposed a blind area leading to the left coronary artery. This area was further opened by resecting a lump of thick tissue presumed to be an analogue of the left coronary cusp, and this gave access to the right coronary sinus giving off the right coronary artery which was covered by another rudimentary cusp. Fused to this was a third lump assumed to be the non-coronary cusp which was also excised. Resection of a slight subvalvar fibrous thickening and a myotomy into severe subvalvar hypertrophy was done. A 1.6 cm reconstituted frozen aortic homograft was put in below the coronary arteries and below the origin of the curious lumps of tissue. A Dacron patch was used to close the aortotomy, to widen the hypoplastic aorta and prevent residual supra valve obstruction.

The postoperative course was uncomplicated until the fifth day when she developed aortic regurgitation and gross left ventricular failure. Her condition remained poor in spite of antifailure therapy.

On 7 March 1969 it was decided to re-examine the homograft. After intravenous induction of anaesthesia, bradycardia developed which was untreated, and by the time she was brought into the operating theatre the heart was arrested. She was rapidly put on bypass and the aortotomy was reopened. Inspection of the valve showed no evidence of structural failure or suture dehiscence. The only possibility was that the right coronary cusp was seated too high and not apposing. This was detached and reattached lower down. The valve then appeared to be competent and the left ventricular end-diastolic pressure was now 15 mmHg and the heart came off bypass without difficulty.

The cardiovascular state remained stable throughout her postoperative course apart from transient heart block requiring pacing; no diastolic murmur was heard. Unfortunately she remained with a spastic quadriplegia and has not regained consciousness but is still alive.

Case 2 A boy aged 7 years was the third child of a normal pregnancy. The birthweight was 2.72 kg (6 lb) at term.

A murmur was heard at age 3 years when he was found to have signs of aortic stenosis. Fatigue and effort dyspnoea which coincided with electrocardiographic deterioration were noted at age 6 years. He was investigated and referred for an operation. While awaiting operation he was admitted 6 months later because of two syncopal attacks which occurred when playing with his friends.

Examination showed a small child below the normal percentiles for height and weighing 16 kg. He had a pointed pixie face resembling his mother but no other unusual stigmata. The pulse was small and slow rising in the neck where an intense systolic thrill was evident. A powerful left ventricular thrust was palpable. On auscultation there was a long ejection systolic murmur and

soft delayed aortic closure, but no ejection sound could be heard. A soft immediate diastolic murmur was constant. The electrocardiogram confirmed the presence of gross left ventricular hypertrophy with sharp T inversion in all left ventricular leads. Chest radiograph showed small aortic knuckle without poststenotic dilatation of the ascending aorta and slight cardiac enlargement.

The clinical diagnosis was severe subvalvar aortic stenosis but angiocardiology showed a small ascending aorta with a supra-aortic narrowing and a curious thickened stenotic-aortic valve which was regurgitant (Fig. 4). There was a 100 mmHg gradient across the left ventricular outflow tract.

On 29 October 1969 (D.N.R.) with cardiopulmonary bypass the aortic valve was explored. The findings resembled those in Case 1 and the preoperative gradient across the outflow was 145 mmHg. In this patient the three lumps of valve tissue were fused, knobby, and grossly thickened. An attempt was made to sculpture the valve rather than replace it in such a young child. The commissures were divided and this left a more regurgitant but less stenotic valve with a gradient of 40 mmHg across it. A myotomy incising into the hypertrophied subvalvar region was made. A Dacron patch was used to close and widen the narrow aorta.

The child made an uneventful recovery from operation though a diastolic murmur was always

FIG. 4 *Angiocardigram from Case 2. Left ventricular injection (anteroposterior) showing thick aortic valve, small aorta, and supra valve narrowing.*

FIG. 5 Longitudinal section of base of aorta, aortic valve cusp, and ventricular septum from Case 2. The gross thickening of the aortic valve cusp (arrow) in comparison with the mitral valve is obvious. (Given by Dr. W. Scarrett.)

signs or blood picture during the next fortnight, but he then developed fever, splinter haemorrhage, showers of red spots on legs and feet, and was admitted looking very toxic. The retinae showed petechiae but no evidence of excessive pigmentation. Blood cultures continued to be negative but he was then treated for bacterial endocarditis with 12 million units penicillin in a drip and 0.5 g streptomycin daily. There was a slight response to treatment, but 10 days later the fever increased and more spots appeared including a classical Osler node on the little toe. The penicillin was increased with slight improvement for 2 weeks, but once again he worsened. There was no change in his cardiovascular signs and no evidence of localized pus. After several relapses which failed to respond to massive doses of intravenous penicillin he developed the signs of acute intracerebral damage and was transferred to the Middlesex Hospital under the care of Mr. John Andrew for evacuation of a cerebral abscess or haemorrhage. Burr holes were made and ventricular fluid was found at high pressure containing red cells and 170 white cells/mm³ but he never regained consciousness and died a few days later.

At necropsy by Dr. W. Scarrett at the Middlesex Hospital, the source of the infection was found to be an abscess full of Gram-positive cocci under the aortic arch and by the side of the top of the ascending aorta which had tracked down into the aorta and was communicating with a false aneurysm behind the Dacron patch. There was no infection on the three aortic 'cusps' which were grossly thickened (Fig. 5). The 'cusps' were in the form of lumps without any evidence of the usual curved cusp. Histology showed that the cusps were made up of excessive amounts of avascular hyaline connective tissue containing scattered fibroblasts with areas of patchy mucoid degeneration. The ascending aortic wall was thin with generalized hypoplasia. Death was due to a subarachnoid haemorrhage from a mycotic weakness in the right middle cerebral artery.

Case 3 A boy aged 10 years was the product of a normal pregnancy when the mother was 31 years. The birthweight was 4.08 kg (9 lb). There were 2 other normal sibs. The child developed normally but was always smaller than his brothers though his birthweight was greater. A murmur was heard at age 18 months. He noted effort dyspnoea during games and there was a story of angina after running since aged 8 years but his physical activity had never been restricted. After demonstration of a gradient of 85 mmHg across the left ventricular outflow tract and pulmonary artery angiocardiology, he was referred by Dr. E. Coleman for resection of subaortic stenosis as the electrocardiogram was showing deterioration with T inversion in the left ventricular leads.

On examination he was seen to be a small boy with a pointed pixie face unlike his parents or sibs; he was below the third percentile for height. The pulse was small with a sharp upstroke, more conspicuous in the right than the left arm where the pulse felt anacrotic. There was the usual

present and louder than before operation. By the third week he still had a low grade fever with occasional sudden spikes to 39°C. No cause for this could be found and in spite of splenomegaly he was gaining weight with good appetite, was happily active, and maintaining the haemoglobin at 89 per cent; he was discharged after 5 weeks for weekly examination and full blood picture at outpatients. There was no change in the clinical

thrusting apex, a systolic thrill at the base and neck, a long ejection systolic murmur, an immediate aortic diastolic murmur, and soft delayed aortic valve closure. No ejection sound was heard or recorded. The chest x-ray showed slight cardiac enlargement, small aortic knuckle, and no prominence of the ascending aorta. Because of the policy of clearly outlining the anatomy of all forms of congenital aortic stenosis before operation and the similarity to Cases 1 and 2, left ventricular angiocardiology and retrograde aortography were done. The aortic valve was thick and mildly regurgitant and appeared to be adherent to a supravalue narrowing. The aorta looked small and hypoplastic (Fig. 6).

At operation (D.N.R.) the findings were similar to those in Case 2. The aortic valve cusps were poorly formed, thick and nodular, and shelf-like with 3 fused commissures. The aorta was opened through an oblique incision which opened through a supra-aortic ridge at the level of the commissural attachments. The non-coronary sinus was fully opened down to the base of its sinus, and the ridges of the left and right coronary sinuses were excised giving full access to both coronary artery orifices. The valve was trimmed and one com-

missure opened. Inspection of the subvalvar region showed no evidence of localized obstruction though septal hypertrophy was obvious. No myotomy was done. An elliptical Dacron patch was fixed in the non-coronary sinus as part of the closure of the aortotomy.

The postoperative course was uncomplicated. Aortic regurgitation persisted but the blood pressure was maintained at 125/70 mmHg.

Discussion

Congenital aortic valve stenosis, before calcification occurs, typically appears as a pliable dome made up of two or three fused cusps with a rudimentary commissure. The valve cusps may be thickened with small avascular lumps which sometimes contain calcium granules in late adolescence or in the third decade. Usually, under 20 years the stenosed aortic valve is mobile, opens with an ejection sound, and is associated with poststenotic dilatation of a normally developed ascending aorta. In the 3 patients described the valve

FIG. 6 Angiocardiology from Case 3. (a) Anteroposterior view of left ventricular angiocardiology in systole showing thickened aortic valve cusps possibly fusing with a supra-aortic stenosis and hypoplasia of the aorta. (b) Anteroposterior aortogram showing mild aortic regurgitation through left coronary cusp which has a squared-off appearance. The supra-aortic narrowing is obvious.

(a)

(b)

cusps were not only very thick and badly formed but two were shown to be made up of curious fibrohyaline material unlike the usual valve histology; it is not surprising that they did not open with a click. In Case 1 there was little attempt at differentiation into cusps which were represented by tissue 'blobs'. Present in all these patients were hypoplasia of the aorta and a true supra-aortic valve obstruction which are not found in association with the usual congenital aortic valve stenosis.

This hypoplasia of the whole aorta suggests that this may be part of a diffuse arteriopathy similar to that found with hypercalcaemic supra-aortic stenosis (Garcia *et al.*, 1964). However, there were none of the other stigmata generally associated with that syndrome, such as mental deficiency, large ridged teeth, and prominent supraorbital ridges (Williams, Barratt-Boyes, and Lowe, 1961), nor evidence of pulmonary or other arterial stenoses (Beuren *et al.*, 1964). Aortic valve stenosis is not associated with the hypercalcaemic problem though clinical features in such patients at first suggested this possibility (Black and Bonham-Carter, 1963); subsequent investigation demonstrated the supra-aortic obstruction. It appears that the cases described here represent a different problem. It is known that the diffuse arteriopathy with supra-aortic stenosis can occur without hypercalcaemia and present as a familial problem (McDonald, Gerlis, and Somerville, 1969) or as a sporadic occurrence. The sibs of Cases 2 and 3 were examined and found to be normal; Case 1 was an only child and there was no history of heart disease in other members of the family.

The pulmonary and aortic valves, when congenitally stenosed in Turner's syndrome and its mosaic forms, are frequently very thick and lumpy containing material which histologically resembles the valve structure shown in Case 3. None of these patients had true Turner's syndrome though Case 1 had some of the features seen in the mosaic form and also the pulmonary valve was found to be abnormal at operation. The sex chromatin was normal in all, and there was no evidence of chromosome abnormality from blood culture in Cases 1 or 2. However, the small stature of all three patients in spite of normal birthweight and normal-sized sibs, as well as the curious hypoplasia of the aorta with unusual change in the valve, suggests that there is something common to these 3 patients with aortic stenosis, but they do not fit into any of the described syndromes. It is difficult to know whether the facies is outside the normal variation. Congenital abnormalities outside

the cardiovascular system in Case 1 and the general physical abnormalities of all 3 patients suggest there may be a diffuse congenital abnormality, but it is impossible to say whether this disturbance in foetal life is infective, genetic, metabolic, or endocrine. There was no history of rubella in any of the pregnancies of these patients nor did the retinal fundi suggest this aetiology, but theoretically this could cause these types of associated lesions (Rowe, 1963). Possibly some other virus or infective agent was involved.

The clinical presentation in these patients with the absence of both dilatation of the ascending aorta and an opening ejection sound in combination with an immediate diastolic murmur suggested the diagnosis of fixed organic subaortic stenosis rather than aortic valve stenosis. Subvalvar stenosis was the clinical diagnosis in all 3 patients and was considered to be confirmed in Case 3 after pulmonary artery angiocardiology and gradient measurement by left ventricular puncture. The need for good aortography and left ventricular angiocardiology is obvious when the patient presents with these features of congenital aortic stenosis and should be routinely performed in patients considered for operation.

Recognition of this type of lesion is important before an operation is planned, as this is a more formidable surgical problem than either organic subvalve or supravalue stenosis or the usual form of congenital aortic valve obstruction in which simple aortic valvotomy may be adequate to relieve temporarily the left ventricle of its burden. Though valvotomy and preservation of the patient's own valve should be attempted, it may not always achieve a good haemodynamic result with this type of valve abnormality, as was shown in Case 2. Thus, some form of valve replacement as in Case 1 may be required to restore good function and to prevent aortic regurgitation. In view of this we believe that operation should be undertaken only when damage to the left ventricle appears, preferably at the first sign of ST-T changes in left ventricular leads, and is not indicated necessarily by the finding of a high outflow tract gradient. It is considered better to restrict the patient from excessive physical activity, thereby abolishing symptoms if present, than to undertake this difficult surgical proposition prematurely.

The best type of aortic valve replacement in this and other young patients has yet to be evaluated over a long term. Pulmonary autograft (Ross, 1967) has shown promising results over a 3-year period, but this operation may be inadvisable in these patients owing to

associated abnormality of the pulmonary valve. A cadaver aortic homograft valve is sited with difficulty owing to the distortion of the narrow aortic root, and the future of these valves after 7 years is also unknown (A. Yates and D. N. Ross, personal communications). A plastic prosthesis is undesirable in so young a patient and with such a small aortic root the appropriate sized prosthesis would be stenotic. Similarly, a frame mounted fascia lata valve would probably leave important obstruction. At the present time it appears that replacement with a fresh homograft is the best form of valve replacement in these circumstances. The type of valve replacement employed also depends on availability of material, and experience in this series cannot necessarily be applied or recommended to others.

The purpose of this report is to emphasize the existence of this uncommon form of aortic stenosis which poses more difficult problems in surgical management compared with other forms of congenital aortic stenosis amenable to simple valvotomy, and inevitably has a worse surgical prognosis. It is also possible that there is a different and as yet unidentified factor in the aetiology of this form of congenital aortic stenosis which appears to be part of a more diffuse abnormality.

Addendum

Recently Dr. R. E. Bonham-Carter has referred a boy aged 7 years with pulmonary valve stenosis and the same hypoplasia of the ascending aorta

associated with supra-aortic valve, and probable subvalve stenosis; this appeared to be associated with the rubella syndrome with a confirmatory high titre of rubella antibodies in the blood and typical retinal pigment.

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