# Supravalvular aortic stenosis in the adult

## A case presentation with unique associated features

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### Summary

A 42-year-old symptomatic woman with possible adult Williams's syndrome (mental retardation, 'elfin facies' and supravalvular aortic stenosis (SVAS)) is documented. This patient displayed many unique features in addition to the severe SVAS (peak systolic gradient 96 mmHg): there was an associated persistent left superior vena cava draining into the coronary sinus, mitral valve prolapse (Barlow's syndrome) and complete right bundle-branch block on ECG. Peripheral pulmonary artery stenosis was absent. M-mode echocardiography in the adult with SVAS is described for the first time in the literature, as is the use of the 60° cross-sectional scan. Both these non-invasive procedures proved of value in the diagnosis. The degree of subendocardial ischaemia, as determined by the 'endocardial viability ratio', was calculated and the possible mechanisms producing angina pectoris with 'coronary artery hypertension' in this condition are discussed. The literature on associated mitral valve abnormalities in SVAS is reviewed. A successful surgical result was obtained; the various forms of operation are outlined.


*S. Afr. med. J.*, **59**, 796 (1981).

## Clinical presentation

The patient, a 42-year-old White woman, had first been seen some 6 years previously and diagnosed as having aortic stenosis without symptoms. In 1975 she was stated to have infective endocarditis on the basis of cardiac lesions, splenomegaly, and culture of a single blood specimen for *Streptococcus faecalis*, and was apparently treated with success. In 1978 she underwent investigation for refractory anaemia and macrocytosis, as well as a possible Fanconi syndrome. Intensive investigation led to a final diagnosis of 'pre-leukaemia'. At that stage she was also treated for possible brucellosis with osteitis. The patient was then referred to the Cardiac Clinic of Tygerberg Hospital in March 1980. She claimed that she had been 'born with a leaking heart'. It was rather difficult to obtain a concise history, but this was most suggestive of angina pectoris over the preceding few months, with fatigue and shortness of breath on moderate exertion. The patient initially resided in an institution for people of low intelligence, but was later transferred to Stikland Hospital as she apparently attempted suicide. She had three healthy brothers and there was no indication of hereditary disease. Her medication consisted only of tranquillizers.

On examination she was obviously mentally retarded with a uniformly short stature and 'elfin facies' (Fig. 1). No other abnormalities were noted on general examination.

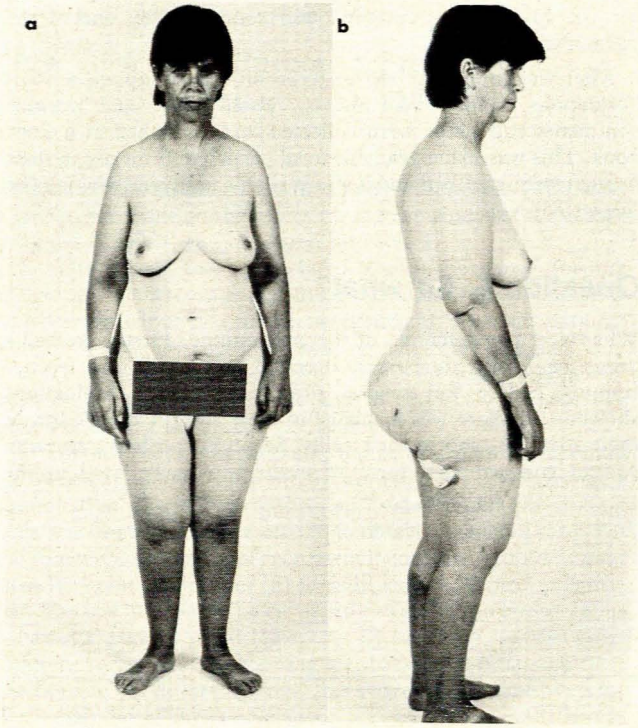


**Fig. 1. Body habitus of patient showing 'elfin facies', short stature and steatopygia.**

Examination of the cardiovascular system revealed a normal radial pulse, with all pulses present and no radiofemoral delay. The jugular venous pressure was not elevated and the blood pressure was 150/80 mmHg in both upper limbs. The apex beat was impalpable and there was no evidence of ventricular enlargement. A systolic thrill at the base of the heart was also evident over the carotid arteries, especially the right. On auscultation over the mitral area the first and second heart sounds were normal and there were no additional sounds. A grade 1/6 pansystolic murmur was audible which radiated to the axilla and up the left sternal edge to the base of the heart. There were no diastolic murmurs. The same features were detected in the tricuspid area. In the pulmonary area a grade 3/6, late-peaking, ejection systolic murmur was audible of the same quality as that heard at the apex. The aortic component of the second heart sound was easily audible and the pulmonic

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Date received: 4 July 1980.

component was not accentuated. There were no ejection clicks. The aortic area demonstrated similar features except that the systolic murmur had an intensity of 4-5/6 and also radiated up both sides of the neck. Examination of the respiratory, gastro-intestinal and central nervous systems showed no abnormality.

A chest radiograph (Fig. 2) showed a fairly prominent main pulmonary artery segment as well as good demarcation from the aortic knuckle. The ascending aorta was dilated or unfolded but the left atrial appendage was prominent. There was no obvious ventricular enlargement and the lung fields were normal.

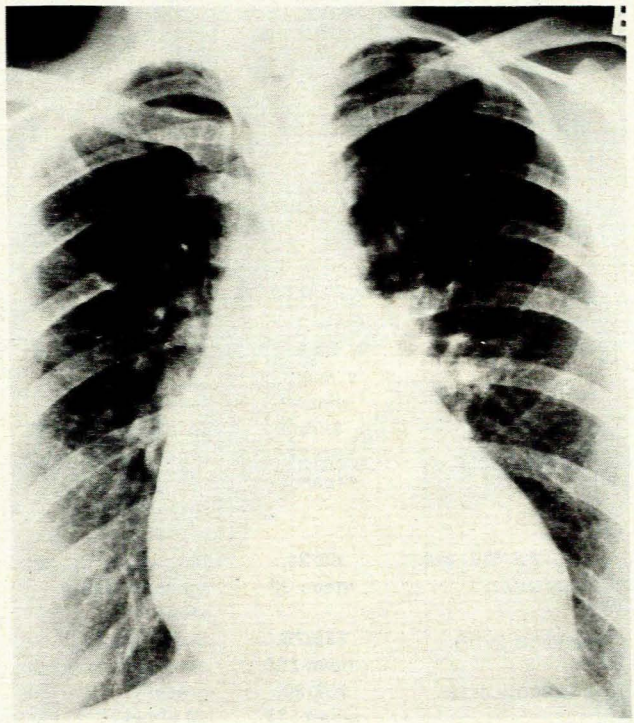

**Fig. 2. Postero-anterior chest radiograph demonstrating prominent main pulmonary artery segment and left atrial appendage.**

A resting ECG (Fig. 3) revealed sinus rhythm of 81/min, a mean QRS axis of plus 104°, and a PR interval of 0,20 second. There was evidence of complete right bundle-branch block and the atria did not appear hypertrophied.

Urine, haemoglobin value, white blood cell count and erythrocyte sedimentation rate were normal, as were serum electrolytes, fasting glucose values, liver function, haematological values, and renal function.

A skeletal survey revealed normal bone maturation. The skull

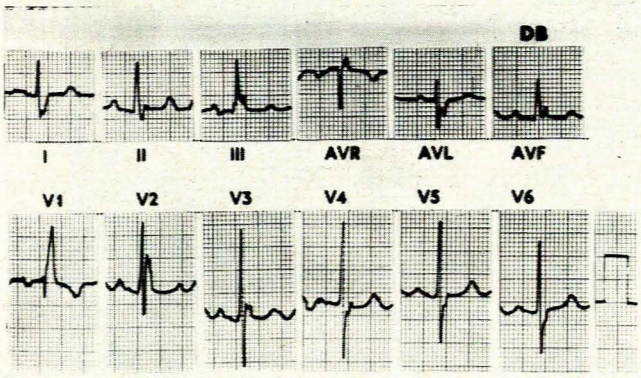


**Fig. 3. Resting ECG showing complete right bundle-branch block without any atrial hypertrophy.**

radiograph demonstrated definite thickening of the bones of the vault with increased density, a feature suggestive of some hyperplastic marrow condition or previous hypercalcaemia. There were no dental anomalies. Chromosome studies were entirely negative.

A phonocardiogram (Fig. 4) showed a single second heart sound, absence of any ejection or non-ejection clicks, and the ejection systolic murmur as well as a normal-looking carotid arterial pulse.

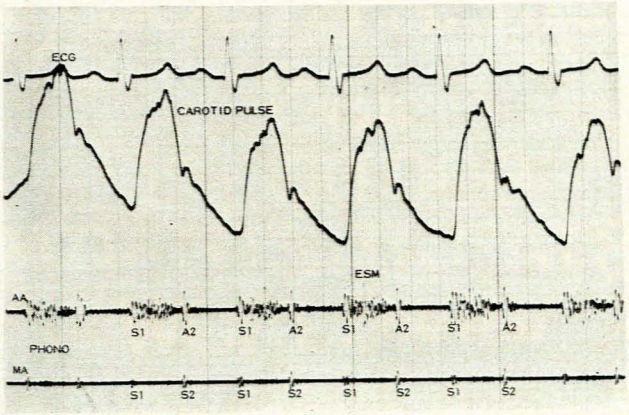


**Fig. 4. Phonocardiogram with normal carotid pulse tracing, and ejection systolic murmur seen at aortic area (AA), but less evident at mitral area (MA).**

An M-mode echocardiogram (Fig. 5) showed a decrease in the internal aortic diameter (measured just proximal to the aortic valve cusps) from 21 mm to 14 mm at the area of supravalvular aortic stenosis (SVAS), a 33% (7 mm) reduction. This compared quite favourably with the reduction of 42% (from 16 mm to 9 mm) calculated on cine angiography, and was in keeping with previous findings[1] that echocardiography tends to underestimate the severity of the obstruction. Prolapse of the posterior mitral leaflet was also clearly demonstrated. The left atrium was enlarged and significant symmetrical left ventricular hypertrophy was seen (Table I).

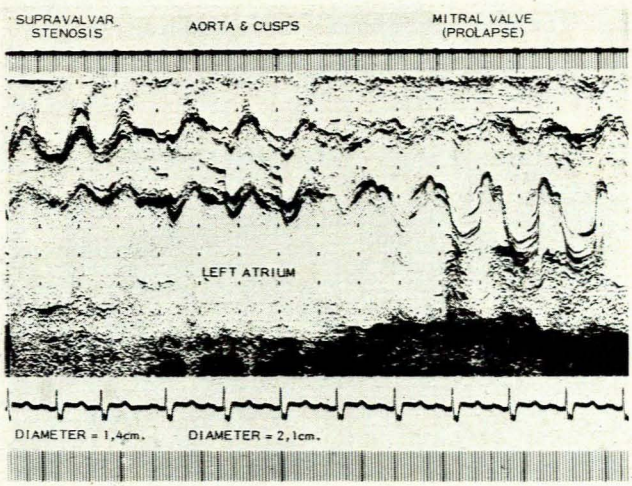


**Fig. 5. M-mode echocardiogram demonstrating supravalvular stenosis with 7 mm reduction in aortic diameter. Prolapse of posterior mitral leaflet into dilated left atrium is clearly visualized.**

Cross-sectional echocardiography (Fig. 6) demonstrated the area of SVAS as well as the normal mobility of the aortic cusps. The left atrium also appeared enlarged. These features confirmed the findings of M-mode echocardiography.

**TABLE I. ECHOCARDIOGRAPHIC MEASUREMENTS**

| | Result | Normal (mean) |
|---|---|---|
| Left ventricular end-diastole | 44 mm | 35-56 (46 mm) |
| Left ventricular end-systole | 27 mm | |
| Interventricular septum thickness (IVS) | 16 mm | 7-11 ( 9 mm) |
| Left ventricular posterior wall (LVPW) | 17 mm | 7-11 ( 9 mm) |
| IVS/LVPW ratio | 0,94 | < 1,3 |
| Left atrium dimension | 52 mm | 19-40 (29 mm) |
| Right ventricular end-diastole | 13 mm | 10-26 (17 mm) |
| Left ventricular end-diastolic volume | 88 ml | 130 ml/m$^2$ |
| Left ventricular end-systolic volume | 27 ml | 25 ml/m$^2$ |
| Ejection fraction | 69% | > 60% |
| Shortening fraction | 39% | 28-38% |
| Systemic isovolumic contraction time | 24 ms | 28-38 ms |
| Systemic pre-ejection period (PEP) | 81 ms | — |
| Left ventricular ejection time (LVET) | 326 ms | — |
| PEP/LVET ratio | 0,25 | < 0,28-0,38 |

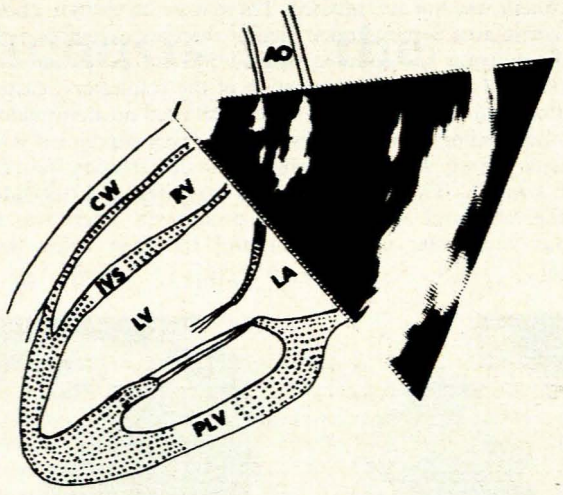


Fig. 6. Cross-section echocardiogram. Area of SVAS and aortic valve cusps are clearly seen.

**TABLE II. INTRACARDIAC PRESSURES**

| Catheter position | Pressure (mmHg) | Comment |
|---|---|---|
| Right atrium | 'a' wave 10, 'v' wave 7, mean 5 | Large 'a' wave<br>Normal |
| Right ventricle | 50/5-7 | Raised systolic |
| Main pulmonary artery | 50/24, mean 33 | Moderate pulmonary hypertension, no pulmonary stenosis |
| Right and left pulmonary artery | 50/24, mean 33 | No peripheral pulmonary branch stenosis |
| Ascending aorta | 136/76, mean 100 | Supra-aortic stenosis with peak systolic gradient (PSG) of 96 mmHg |
| Supra-aortic area | 232/80, mean 112 | |
| Left ventricle | 232/4-23 | Raised pressures. No aortic stenosis |
| dp/dt | (mm/s) 2 409 | Normal |
| Pulmonary capillary wedge | 'a' wave 24, 'v' wave 34, mean 21 | Moderately elevated. No mitral stenosis |

Cardiac catheterization was performed using the standard percutaneous Seldinger technique via the right femoral vein and artery. A 7F Goodale-Lubin and pigtail catheter was used to measure intracardiac pressures on the right and left sides, as well as oxygen saturation in the main pulmonary artery and central aorta (Tables II and III). The right-sided catheter was also advanced into a persistent left superior vena cava (SVC) via the coronary sinus into which contrast medium was injected (Fig. 7), using a 7F Rodrigues-Alvarez catheter. Pressure withdrawal from the left ventricle to the ascending aorta indicated severe SVAS (Fig. 8). Left ventricular cine angiography (right anterior oblique view) demonstrated a normally contracting ventricle but there was obvious prolapse of the posterior leaflet of the mitral valve (non-calcified) accompanied by mild mitral insufficiency (Fig. 9, left). Aortic cine angiography (left anterior oblique view) showed asymmetrical SVAS distal to the origins of the coronary arteries (Fig. 9, right). The aortic valve had three cusps, was not calcified and was competent. Selective coronary arteriography

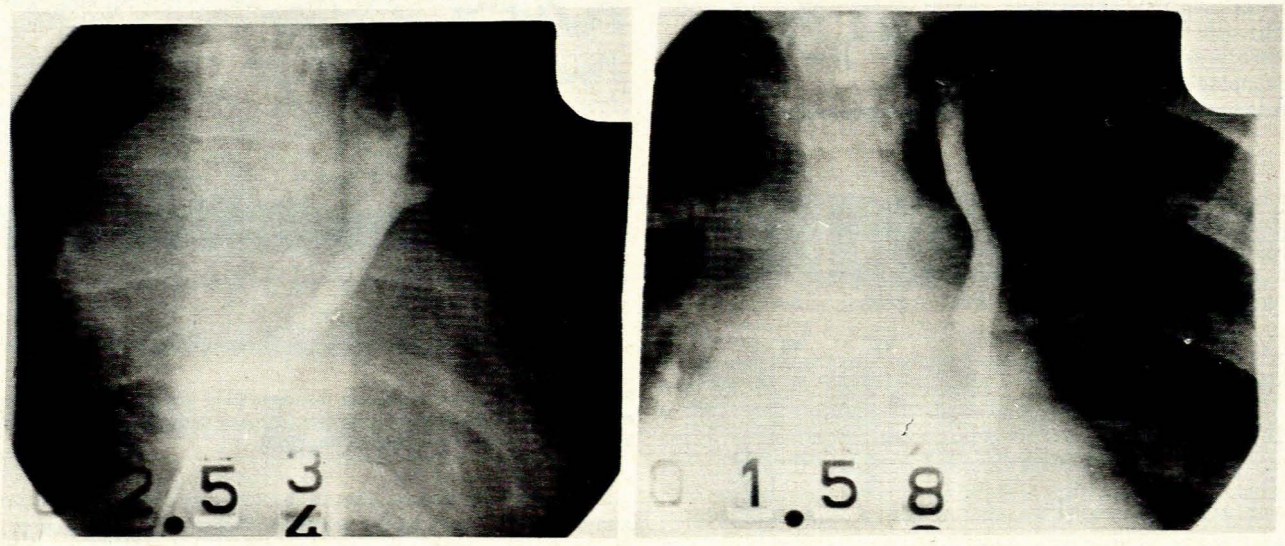

Fig. 7. Cine angiograms showing persistent left SVC draining into enlarged coronary sinus.

**TABLE III. HAEMODYNAMIC CALCULATIONS**

| | Results |
|---|---|
| Oxygen consumption (ml/min) | 240 |
| Arteriovenous $O_2$ difference (vol %) | 3,0 |
| Cardiac output (Fick) (l/min) | 8,3 |
| Cardiac index (Fick) (l/min/m²) | 5,6 |
| Pulmonary vascular resistance (U) | 1,4 |
| Index (U/m²) | 2,1 |
| Systemic vascular resistance (U) | 11,4 |
| Index (U/m²) | 16,9 |
| Pulmonary/systemic resistance ratio (%) | 12 |
| Stroke volume (ml/beat) | 101 |

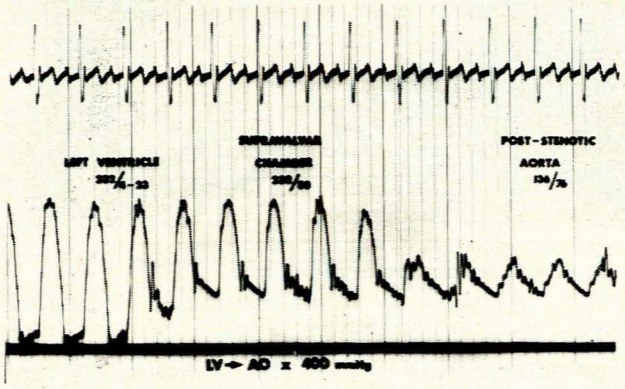


**Fig. 8. Pressure withdrawal from left ventricle (LV) to ascending aorta (AO) demonstrating severe SVAS.**

demonstrated normal coronary arteries (dominant right coronary). Pulmonary arteriography (cut films) in the anteroposterior view showed the absence of peripheral pulmonary branch stenosis as well as normally draining pulmonary veins (Fig. 10). In retrospect the supravalvular membrane was clearly visible on angiography. The procedure was completed without complication.

## Operative findings and surgical correction

A median sternotomy approach was used and venous cannulas were inserted into the superior and inferior vena cava. Moderate body hypothermia to 28°C was employed during cardiopulmonary bypass. Cold cardioplegic solution (500 ml) was then rapidly infused into the aortic root for myocardial protection.

A transverse left atriotomy, behind the interatrial groove, gave excellent exposure to the mitral valve. A small cleft in the anterior leaflet of the mitral valve near the posteromedial commissure was repaired. The degree of posterior leaflet prolapse could not be determined at surgery.

The left superior vena cava drained into the coronary sinus but did not communicate with the right SVC. The left ventricle demonstrated concentric hypertrophy, and slight dilatation of the left atrium and enlargement of the left atrial appendage were noted. There was quite marked post-stenotic dilatation of the ascending aorta with an 'hourglass' narrowing some 2,5 cm above the aortic valve annulus. An oblique aortotomy into the non-coronary sinus of Valsalva, extending from the base of the innominate artery to just above the aortic annulus, was then carried out. This incision revealed a fibrous membrane with a central opening about 0,5 cm² in size. The aortic valve appeared normal, but both coronary ostia were enlarged, the remainder of the vessels appearing macroscopically normal. The fibrous ring was excised and the aortic root enlarged with a large elliptical gusset of woven Dacron. An extended aortoplasty as described by Doty *et al.*[2] was not deemed necessary as adequate symmetrical enlargement of the aortic root was achieved with the elliptical patch. The patient made an uneventful recovery. It is planned to repeat cardiac catheterization at some later date.

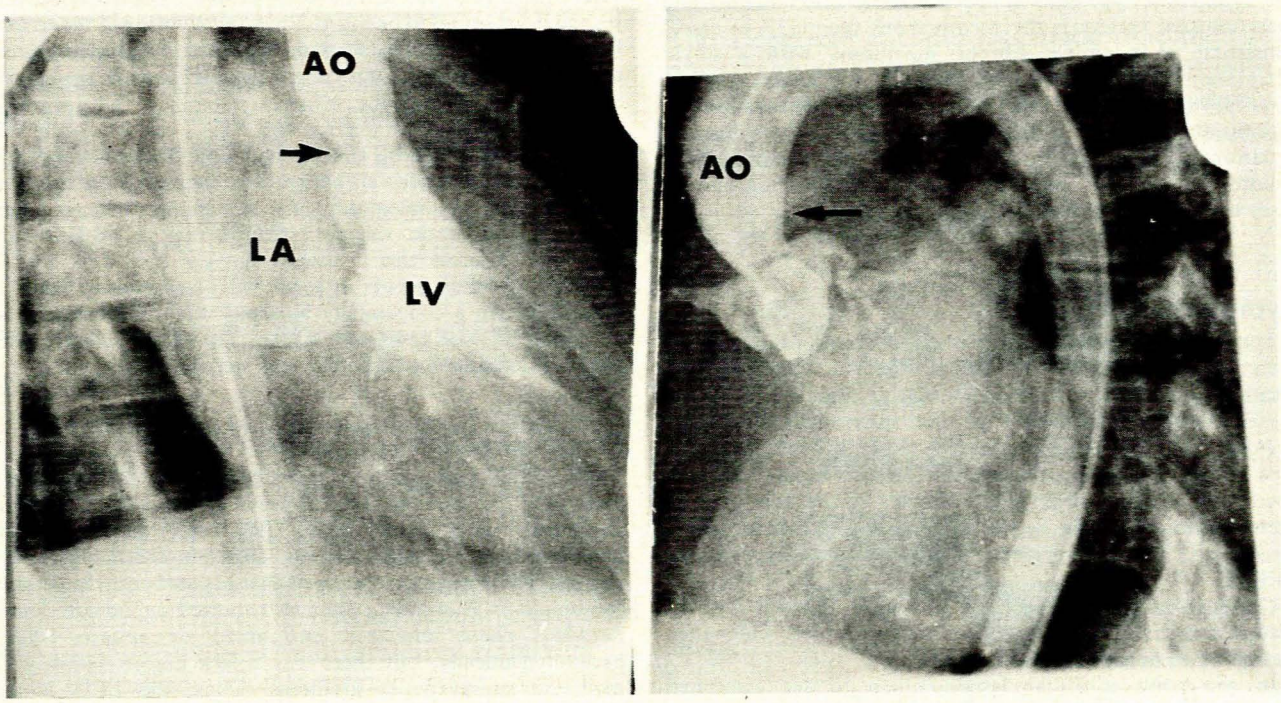


**Fig. 9.** *Left:* **Left ventricle (LV) cine angiogram (right anterior oblique projection) showing mitral valve prolapse and dye-filled left atrium (LA) secondary to mitral insufficiency (AO = ascending aorta);** *right:* **aortic cine angiogram (left anterior oblique view). Supravalvular stenosis (*arrowed*) is clearly demonstrated with proximal dilatation of the coronary arteries. The fibrous membrane above the aortic valve is present (*arrowed*).**

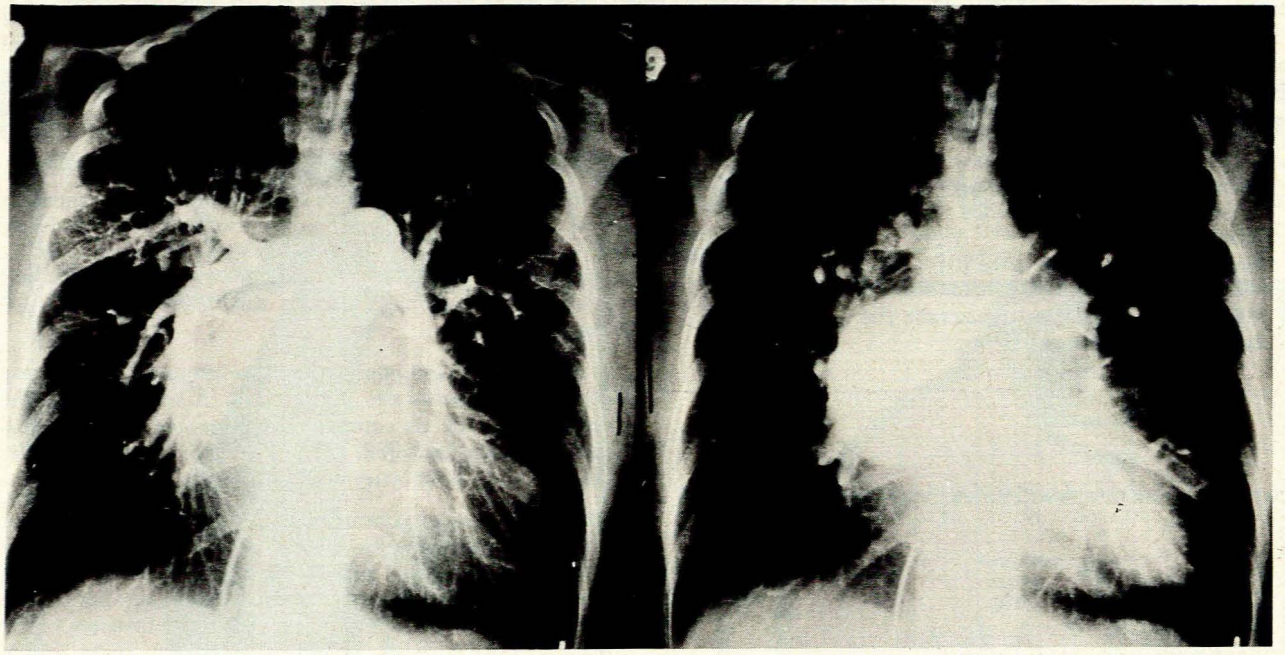

**Fig. 10. Pulmonary arteriogram showing normal pulmonary artery tree (*left*) and normal pulmonary venous phase (*right*).**

## Discussion

SVAS, a congenital abnormality with the obstruction just cephalad to the origin of the coronary arteries, was originally described by Mencarelli in 1930[3] and is seen in between 0,6% and 6% of cases of left ventricular outflow tract obstruction.[4,5] There are three recognized varieties, the most common type being the hourglass (segmental) variety which occurs in some 66% of cases,[4] and in which there is gross thickening of the media often associated with intimal fibrous hyperplasia. Next in frequency is the hypoplastic type seen in approximately 21% of cases. The most unusual variety is the membranous type (13% of cases) consisting of a simple fibrous membrane with a single perforation allowing for blood flow. In the adult SVAS is classically not associated with elfin facies, mental retardation and significant peripheral pulmonary branch stenosis (Williams's syndrome).[6,7] Although over 150 cases have been documented in children, this lesion is considered rare in adults, in whom only 26 cases have been described.[4,8-11]

## Symptoms and signs

Symptoms of SVAS are not really different from those in aortic valvular stenosis or subaortic stenosis. Classic angina pectoris is the most common complaint and is usually the presenting symptom (refer to 'Pathophysiology of angina pectoris').[5,11-13] In our patient this was rather difficult to assess in view of her mental retardation, and because angina may well have been due to mitral valve prolapse. By the time patients present with angina the degree of stenosis is significant and is usually associated with evidence of left ventricular hypertrophy on ECG.

**Congestive cardiac failure** is a most unusual form of presentation and often accompanies angina pectoris, which then also becomes decubitus in character.

In the adult it is far less common to see associated hypercalcaemia, peculiar facies, mental retardation, stenosis of the branches of the aortic arch, and peripheral pulmonary artery stenosis. Our patient's form was similar to that usually seen in juveniles, apart from the absence of stenosis of the pulmonary and aortic branches. Our patient, at 42 years of age, is also the oldest patient with SVAS in the English literature, the second oldest being 37 years of age when reported by Page *et al.*[9]

Bacterial infection at the site of SVAS with the well-known features of infective endocarditis has been described.

**Differentiation from aortic valvular stenosis** is often somewhat difficult. In SVAS an ejection click is usually absent; the rough ejection systolic murmur has a maximum intensity at the suprasternal notch (as opposed to the aortic area) and radiates well into the neck and sometimes into the interscapular region. The more severe the SVAS, the less likely is the aortic component of the second heart sound to be heard. Reversed splitting of the second heart sound and the presence of third and/or fourth heart sounds are often encountered. With aortic valvular stenosis the carotid arterial pulse is anacrotic, whereas it tends to be normal in SVAS. Aortic valvular stenosis and SVAS may be combined, making clinical diagnosis almost impossible. Diastolic murmurs of aortic insufficiency can be heard sometimes with either of these lesions. Radiologically, the ascending aorta is often prominent in aortic valvular stenosis due to post-stenotic dilatation, whereas this is usually not the case with SVAS. Calcification often occurs in aortic valvular stenosis, which is usually due to a congenitally bicuspid valve, but can also be visualized in some cases with SVAS. The ECG is of very little use in differentiating aortic valvular stenosis from SVAS.

## Electrocardiographic features

The ECG findings in supravalvular aortic stenosis[14] are essentially the same as in aortic valvular stenosis,[15,16] with an abnormally wide frontal plane QRS angle, left ventricular hypertrophy by voltage criteria, and left ventricular strain pattern. These three categories of changes may occur in varying combinations. Maron and Sissman[14] showed a good correlation between these changes and mild or severe SVAS haemodynamically, but there was a lack of correlation with moderate supravalvular gradients. Thus, the ECG is not clinically reliable in estimating the severity of the left ventricular outflow obstruction.

Our patient's ECG showed complete right bundle-branch block and a mean frontal plane QRS axis of plus 104° (Fig. 3).

This was despite the marked symmetrical hypertrophy of the left ventricle found on echocardiography and the severe SVAS demonstrated on cardiac catheterization. Apart from one unproven case of a 30-year-old asymptomatic woman,[17] in whom there was 'right bundle-branch block and probable left ventricular hypertrophy' on ECG, there is no record in the literature of complete right bundle-branch block occurring with SVAS. The significance of this finding is not understood, but it may be due to the mitral valve prolapse in which this conduction abnormality is known to occur.[18] On the other hand, the complete left bundle-branch block has been described in SVAS.[8]

## Echocardiography in diagnosis

Since the clinical features of left ventricular outflow tract obstruction do not always define the anatomical type of obstruction (subvalvular, valvular and supravalvular) echocardiography has proved a valuable non-invasive adjunct to diagnosis. Others have documented the echocardiographic characteristics seen in muscular subaortic stenosis,[19,20] fixed subaortic stenosis,[21,22] and aortic valvular stenosis.[23] It is only fairly recently that attempts have been made to document the echocardiographic features seen in supravalvular aortic stenosis.

### *M-mode echocardiography (single-dimensional)*

Usher *et al.*[24] were the first to demonstrate the presence of SVAS by a continuous recording obtained by directing the transducer sequentially from the left ventricle to the mitral valve and then the aortic root distal to the stenosis. Their patient was a mentally retarded 9-year-old White boy with elfin facies in whom cardiac catheterization confirmed mild SVAS of the segmental or hourglass type with a peak systolic gradient of 15 mmHg.

Nasrallah and Nihill[25] described the echocardiographic features in 2 children with segmental SVAS associated with aortic valvular stenosis. In 1 patient the echocardiogram showed a decrease in the narrowing postoperatively. These workers also noted a close correlation as regards percentage narrowing of the stenotic area between echocardiographic and angiographic findings.

Bolen *et al.*[1] described echocardiographic findings in 6 children with segmental SVAS; echocardiography tended to underestimate the degree of stenosis when compared with angiography. This constant finding was attributed to the echo beam striking the ascending aorta obliquely with angulation of the transducer cephalad from the aortic sinuses, thus causing a falsely increased internal aortic diameter.

In the presence of SVAS, the sinuses of Valsalva are usually more dilated than normal, and thus this underestimation of stenosis tends to be more pronounced. These workers demonstrated a non-linear underestimation of the severity of the narrowing.

Table I shows marked concentric (symmetrical) left ventricular hypertrophy in our patient as evidenced by the increased thickness of both the IVS and LVPW, a decreased PEP/LVET ratio, as well as an increased ejection and shortening fraction secondary to the left ventricular outflow tract obstruction. The internal diameter of the ascending aorta decreases from 21 mm at the aortic valve cusps to 14 mm at the segment of stenosis (Fig. 5). The left atrium is enlarged on account of the mitral insufficiency secondary to prolapse of the posterior mitral leaflet (Barlow's syndrome) as is clearly seen in Fig. 9 (left).

This appears to be the first description of M-mode echocardiographic features seen in the adult with SVAS. It also appears that associated Barlow's syndrome has never been documented previously. This association thus raises the possibility of a common pathogenetic factor.

### *Cross-sectional echocardiography (two-dimensional)*

Weyman *et al.*[26] were the first to document the use of this technique in the diagnosis of SVAS. They described the features in 4 children (1 with hypoplastic and 3 with the hourglass or segmental variety) and 1 adult (segmental type). These workers used a 30° cross-sectional scan with the patients either in the 30° left lateral or supine position, and utilized long axis recordings of the ascending aorta. In each of their cases the echocardiographically determined diameter of the obstruction was within 3 mm of the comparable angiographic value. They also noted a good correlation between the peak systolic aortic gradient found at cardiac catheterization and the percentage decrease in diameter echocardiographically. Thus, because it provides a spatial orientation,[27] this technique allows for more accurate definition of SVAS and complements the M-mode assessment.

In our patient we utilized a 60° cross-sectional scan and thus were able to visualize a larger area. The region of obstruction in the ascending aorta is clearly seen (Fig. 6), as well as the large left atrium. This appears to be the first report documenting the use of this wider cross-sectional scan in diagnosis.

## Pathophysiology of angina pectoris

SVAS differs from aortic valvular stenosis in that the obstruction in the former condition occurs proximal to the origin of the coronary arteries. Therefore, the coronary arteries are exposed to the markedly elevated ventricular systolic pressure and their flow is impeded during systole. Because of the resultant wide pulse pressure in the coronary arteries these vessels are subjected to a significant distending force which causes important structural alterations. Peterson *et al.*[4] described three different types of coronary artery abnormality seen in 20 out of 33 cases of SVAS. The first type was characterized by mild narrowing at or just distal to the coronary ostia[28] and was independent of the lesion causing the SVAS. The second type consisted of a generalized increased tortuosity of the coronary arteries, as well as an increased external diameter caused by hypertrophy of the media which also diminished the internal lumen diameter.[6,11,12] Intimal fibrous thickening, sometimes resembling atherosclerosis, also tended to occur in this group. In the third type fusion was visualized between the area of SVAS and the free margin of a non-fenestrated aortic valve cusp, so as to exclude the related coronary artery from its aortic communication.[29] This vessel was thin-walled, undilated, and normal histologically, whereas the remaining artery showed compensatory hypertrophy, tortuosity and even atherosclerosis.

Vincent *et al.*[30] believe that the mechanism of left ventricular subendocardial ischaemia is the same in both severe valvular stenosis and SVAS. They have shown that raised systolic pressure in the supravalvular chamber and the coronary arteries, seen in SVAS, may increase only systolic (subepicardial) flow. These alterations are inadequate in response to the increased metabolic requirements. Since left ventricular subendocardial coronary flow occurs primarily during diastole a diastolic pressure-time index (DPTI) can be used to measure it. Likewise, the systolic pressure-time index (SPTI) gives an estimation of left ventricular oxygen requirements.[31] Thus, the adequacy or otherwise of subendocardial blood flow can be determined from the ratio DPTI/SPTI (also termed the endocardial viability ratio) which signifies the supply/demand ratio. Vincent *et al.*[30] have shown that if the DPTI/SPTI ratio fell below 0,7 subendocardial ischaemia was present. In our patient (Fig. 11) the relevant values were as indicated below:

| | | | |
|---|---|---|---|
| DPTI | 0,18 | 0,20 | 0,19 |
| SPTI | 0,38 | 0,42 | 0,40 |
| $\frac{\text{DPTI}}{\text{SPTI}}$ | 0,47 | 0,48 | 0,48 |

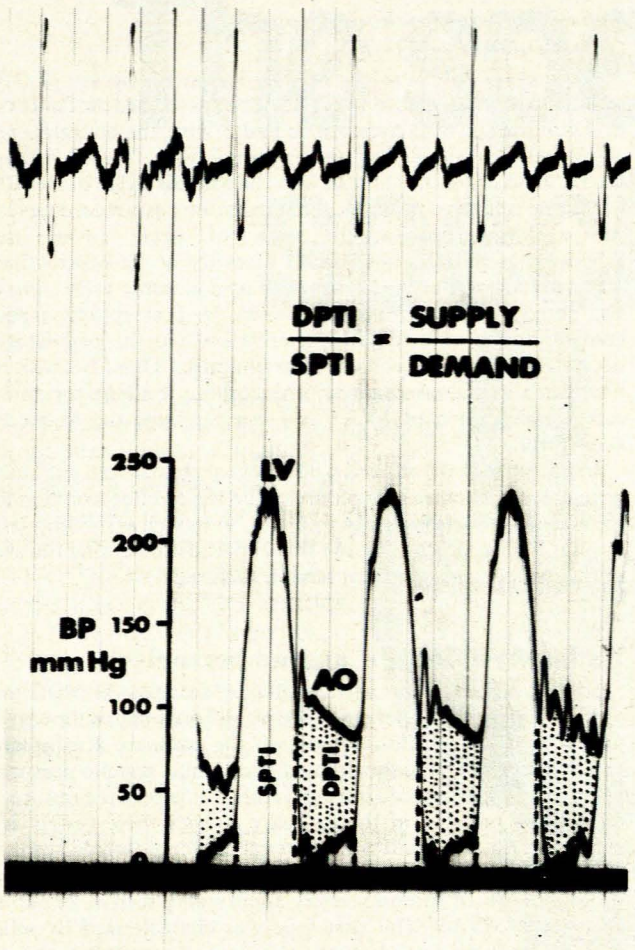


**Fig. 11. Left ventricular (LV) and aortic (AO) pressure trace showing evidence of subendocardial ischaemia (see text) (SPTI = systolic pressure-time index; DPTI = diastolic pressure-time index).**

These calculations demonstrated the presence of significant subendocardial ischaemia. It is interesting that there were no definite features of ischaemia on the resting ECG, although interpretation can be difficult in the presence of complete right bundle-branch block. Our patient also had coronary arteries which were normal apart from localized dilatation near their ostia, as shown angiographically.

More recently, Baller *et al.*[32] have disputed the accuracy of the DPTI/SPTI ratio as an index of imbalance between myocardial oxygen supply and demand. Further studies in patients with SVAS need to be undertaken to resolve this important issue.

## Associated mitral valve abnormalities

Mitral valve lesions have rarely been reported in association with SVAS, and it is uncertain whether this is just coincidental or whether there is a true association. Sissman *et al.*[33] were the first to report a 'somewhat thickened, but otherwise unremarkable' mitral valve at autopsy of a 70-year-old woman with membranous SVAS. Neufeld *et al.*[34] noted fibrous thickening of the mitral valve leaflets as well as the posterior commissure at autopsy on a 2-year-old boy with hypoplastic SVAS. Page *et al.*[9] described angiographic evidence of mitral insufficiency in a 37-year-old man with segmental SVAS. Autopsy showed extensive calcification of the mitral valve annulus without any evidence of rheumatic or congenital deformity of the valve leaflets. In none of these 3 cases was the functional significance of this involvement determined. Other workers[35-37] reported on 3 further cases in which clinical mitral insufficiency of unknown relevance was encountered. In another patient left atrial enlargement was noted.[38]

Becker *et al.*[39] documented autopsy findings of collagenous thickening of the mitral leaflets in an infant and a child with hypoplastic SVAS and in an adult with the hourglass type. These authors also described varying degrees of protrusion of segments of the mitral leaflets toward the left atrium, a feature which they termed 'hooding'. In the 2 older patients there were also endocardial fibrous lesions of the left ventricle directly related to thick chordae attached to the posterior leaflets. These latter lesions were thought to be the result of friction caused by the hooding.[40] The left atrium was enlarged only in the adult, suggesting that there might have been significant clinical mitral insufficiency, and there were also autopsy features to suggest possible healed infective endocarditis in this case.

Prolapse of the posterior mitral leaflet (Barlow's syndrome) and mild mitral insufficiency were demonstrated in our patient. This condition does not appear to have been described in association with SVAS. Mitral valve prolapse has been described in association with secundum atrial septal defect,[41] ventricular septal defect,[42] patent ductus arteriosus,[43] pulmonary valvular stenosis,[42] Fallot's tetralogy,[42] Epstein's anomaly,[44] congenital bicuspid aortic valve stenosis,[45] and some other miscellaneous conditions. It is difficult to be sure whether the 'hooding' of the mitral leaflets described by Becker *et al.*[39] at autopsy could have in fact been proved as prolapse clinically, echocardiographically and angiographically. These workers did not demonstrate the 'myxomatous degeneration' seen in mitral valve prolapse, which makes this association unlikely. 'Hooding' has been described in association with Marfan's syndrome (arachnodactyly),[10] and this syndrome has been found in association with SVAS.[13,28,46] It is also interesting to note that mitral valve prolapse has been documented in association with Marfan's syndrome.[47] Therefore, an association between SVAS and mitral valve prolapse should not be particularly surprising.

## Persistent left superior vena cava

The continued existence of the left brachiocephalic or left SVC is the most common malformation of the systemic intrathoracic veins.[48] This can occur in isolation or in association with other cardiovascular malformations. There are no definite clinical signs or symptoms to suggest its presence but sometimes it may be suspected radiologically by a prominent shadow to the left of the superior mediastinum.

In our patient, a persistent left SVC draining into the right atrium via the coronary sinus was clearly demonstrated (Fig. 7). There was no communication between the persistent left SVC and the normally placed and functional right SVC, a situation which occurs in some 60% of cases.[48] The inferior vena cava was seen to drain normally into the right atrium and had no connection with the persistent left SVC which is sometimes encountered in complex anomalies of the abdominal viscera, for example polysplenia.[49] A possible associated atrial or ventricular septal defect[50] was excluded by oximetry and angiography, as was anomalous pulmonary venous drainage.

Persistent left SVC has been found in association with Fallot's tetralogy,[51] patent ductus arteriosus, coarctation of the aorta, partial or total anomalous pulmonary venous drainage, and a few other cardiovascular anomalies. However, this appears to be the first case in the English literature of SVAS associated with a persistent left SVC, as well as with mitral valve prolapse. The surgeon had to take care not to ligate the persistent left SVC as it drained into the coronary sinus and had no communication with the normal right SVC.

## Surgical treatment

Successful surgical repair of SVAS was first carried out at the

Mayo Clinic by McGoon *et al.*,[12] who widened the aortic segment involved with a diamond-shaped patch graft made of such material as woven Teflon. Others[10,52] used the same technique with good long-term results.

In 1977, Doty *et al.*[2] described an improved technique for correction of severe cases. They successfully operated upon 8 patients, 6 of whom had the hourglass (segmental) type of defect, and the remaining 2 the diffuse hypoplastic variety. These workers used an 'extended aortoplasty' repair in that the fibrotic supravalvular ring was incised at two sites in the right coronary and non-coronary sinus of Valsalva. They claimed that this resulted in a more symmetrical repair. In addition to the use of a tubular Dacron prosthesis fashioned so as to provide a wide aortic cross-sectional area, they lengthened the aortic valve cusps in order to make it function more satisfactorily. This extended aortoplasty made it less likely that the aortic valve cusps would obstruct the coronary ostia, and that there would be resulting aortic insufficiency. Previous workers[4,53] also demonstrated that some 50% of patients operated upon by the original McGoon[12] technique had a left ventricular/aortic peak systolic gradient in excess of 30 mmHg after operation.

Extended aortoplasty has also been used with some success in the diffuse hypoplastic variety, although many authors have advised against any form of surgery on account of the lack of reduction of the pressure gradient, as well as the high operative risk.[36,54,55] Various procedures creating a bypass between the left ventricular apex or ascending aorta and the abdominal aorta or another distal systemic artery have been devised, with varying results.[53,56,57]

More recently, Landes *et al.*[58] reported experience with 9 patients who had the hourglass type of SVAS, and 5 patients suffering from the diffuse hypoplastic variety. They concluded that the latter type of defect was more sinister in that the results of aortic patch angioplasty were poorer. Most of their patients had associated cardiovascular anomalies, such as aortic valvular stenosis, aortic insufficiency, or pulmonary artery stenosis, as well as a variety of coronary ostial abnormalities. In these cases, the authors suggest that the SVAS should be corrected first, as well as any coronary ostial obstruction; only then should the remaining defects be operated upon.

The surgical findings in our case were quite unusual in that, in addition to the segmental ('hourglass') variety of SVAS, a thick fibrous membrane with a single perforation in the centre was present. Surgical repair was carried out using the original method described by McGoon *et al.*[12] as it was thought unnecessary to employ the 'extended aortoplasty' of Doty *et al.*[2] No complications were encountered during the operation and the patient's postoperative course was entirely uneventful.

We wish to sincerely thank Miss H. Weymar of the Cardiac Clinic, Tygerberg Hospital, for her diligent preparation of the manuscript as well as the production of most of the photographs. We should also like to thank Dr N. J. Serman of the Faculty of Dentistry for the dental assessment, and Dr A. E. Retief of the Department of Cytogenetics for carrying out the chromosomal studies.

Finally, thanks are also due to Dr H. Segal, Superintendent of Stikland Hospital, and Dr C. Vivier, Medical Superintendent of Tygerberg Hospital, for permission to publish.

REFERENCES

1. Bolen, J. L., Popp, R. L. and French, J. W. (1975): Circulation, **52,** 817.
2. Doty, D. B., Polansky, D. B. and Jensen, C. B. (1977): J. thorac. cardiovasc. Surg., **74,** 362.
3. Mencarelli, L. (1930): Ann. ital. anat. Ist., **1,** 829.
4. Petersen, T. A., Todd, D. B. and Edwards, J. E. (1965): J. thorac. cardiovasc. Surg., **50,** 734.
5. Hancock, E. W. (1961): Guy's Hosp. Rep., **110,** 1.
6. Williams,. J. C. P., Barratt-Boyes, B. G. and Lowe, J. B. (1961): Circulation, **24,** 1311.
7. Beuren, A. J., Schulze, C., Eberle, P. *et al.* (1964): Amer. J. Cardiol., **13,** 471.
8. Pansegrau, D. G., Kioshos, J. M., Durnin, R. E. *et al.* (1973): *Ibid.*, **31,** 635.
9. Page, H. L., Vogel, J. H. K., Pryor, R. *et al.* (1969): *Ibid.*, **23,** 270.
10. Nordström, S. and Silander, T. (1962): Thorax, **17,** 154.
11. Morrow, A. G., Waldhausen, J. A., Peters, R. L. *et al.* (1959): Circulation, **20,** 1003.
12. McGoon, D. C., Mankin, H. T., Vlad, P. *et al.* (1961): J. thorac. cardiovasc. Surg., **41,** 125.
13. Denie, J. J. and Verheugt, A. P. (1958): Circulation, **18,** 902.
14. Maron, B. J. and Sissman, N. J. (1971): Amer. Heart J., **82,** 300.
15. Hugenholtz, P. G., Lees, M. M. and Nadas, A. S. (1962): Circulation, **26,** 79.
16. Brann, I. (1965): Cardiologia, **46,** 3.
17. Underhill, W. L., Tredway, J. B., D'Angelo, G. J. *et al.* (1971): Amer. J. Cardiol., **27,** 560.
18. Barlow, J. B. and Pocock, W. A. (1975): Amer. Heart J., **90,** 636.
19. Popp, R. L., and Harrison, D. C. (1969): Circulation, **40,** 905.
20. Shah, P. M., Gramiak, R. and Kramer, D. H. (1969): *Ibid.*, **40,** 3.
21. Popp, R. L., Silverman, J. F., French, J. W. *et al.* (1974): *Ibid.*, **49,** 226.
22. Davis, R. H., Feigenbaum, H., Change, S. *et al.* (1974): Amer. J. Cardiol., **33,** 277.
23. Gramiak, R. and Shah, P. M. (1970): Radiology, **96,** 1.
24. Usher, B. W., Goulden, D. and Murgo, J. P. (1974): Circulation, **49,** 1257.
25. Nasrallah, A. T. and Nihill, M. (1974): Brit. Heart J., **37,** 662.
26. Weyman, A. E., Caldwell, R. L., Hurwitz, R. A. *et al.* (1978): Circulation, **57,** 491.
27. Weyman, A. E., Feigenbaum, H., Hurwitz, R. A. *et al.* (1976): Amer. J. Med., **60,** 33.
28. Perou, M. L. (1961): Arch. Path. **71,** 453.
29. Bourassa, M. G. and Campeau, L. (1963): Circulation, **28,** 572.
30. Vincent, W. R., Buckberg, G. D. and Hoffman, J. I. E. (1974): *Ibid.*, **49,** 326.
31. Sarnoff, S. J., Braunwald, E., Welch, G. H. *et al.* (1958): Amer. J. Physiol., **192,** 148.
32. Baller, D., Sigmund-Duchanova, H., Zipfel, J. *et al.* (1979): Basic Res. Cardiol., **74,** 378.
33. Sissman, N. J., Neill, C. A., Spencer, F. C., *et al.* (1959): Circulation, **19,** 458.
34. Neufeld, H. N., Wagenvoort, C. A., Ongley, P. A. *et al.* (1962): Amer. J. Cardiol., **10,** 746.
35. Farrehi, C., Dotter, C. T. and Griswold, H. E. (1964): Amer. J. Dis. Child., **108,** 335.
36. Rastelli, G. C., McGoon, D. C., Ongley, P. A. *et al.* (1966): J. thorac. cardiovasc. Surg., **51,** 873.
37. Takekawa, S. D., Kincaid, O. W., Titus, J. L. *et al.* (1966): Amer. J. Roentgenol., **98,** 800.
38. Shumacker, H. B. and Mandelbaum, T. (1965): Circulation, **31,** suppl. 1, p. 36.
39. Becker, A. E., Becker, M. J. and Edwards, J. E. (1972): Amer. J. Cardiol., **29,** 90.
40. Salazar, A. E. and Edwards, J. E. (1970): Arch. Path. **90,** 364.
41. Pocock, W. A. and Barlow, J. B. (1971): Amer. Heart J., **81,** 720.
42. Rippe, J. M., Sloss, L. J., Angoff, G. *et al.* (1979): Amer. Heart J., **97,** 561.
43. Devereux, R. B., Perloff, J. K., Reichek, N. *et al.* (1976): Circulation, **54,** 3.
44. Roberts, W. C., Glancy, D. L., Seningen, R. P. *et al.* (1976): Amer. J. Cardiol., **38,** 377.
45. Jeresaty, R. M. (1973): Progr. cardiovasc. Dis., **15,** 623.
46. Burry, A. F. (1958): Brit. Heart J., **20,** 143.
47. Barlow, J. B., Bosman, C. K., Pocock, W. A. *et al.* (1968): Brit. Heart J., **30,** 203.
48. Winter, F. S. (1954): Angiology, **5,** 90.
49. Campbell, M. and Deuchar, D. C. L. (1954): Brit. Heart J., **16,** 423.
50. Karnegis, J. N., Wang, Y., Winchell, P. *et al.* (1964): Amer. J. Cardiol., **14,** 573.
51. Rao, B. N. S., Anderson, R. C. and Edwards, J. E. (1971): Amer. Heart J., **81,** 361.
52. Starr, A., Dotter, C. and Griswold, H. (1961): J. thorac. Surg., **41,** 134.
53. Keane, J. F., Fellows, K. E., LaFarge, C. G. *et al.* (1976): Circulation, **54,** 112.
54. Weisz, D., Hartmann, A. F. and Weldon, C. S. (1976): Amer. J. Cardiol., **37,** 73.
55. Berhard, W. F., Keane, J. F., Fellows, K. E. *et al.* (1973): J. thorac. cardiovasc. Surg., **66,** 404.
56. Cooley, D. A., Norman, J., Mullins, C. *et al.* (1975): Bull. Texas Heart Inst., **2,** 376.
57. Merin, G., Copperman, I. J. and Borman, J. B. (1976): Chest, **70,** 546.
58. Landes, R. G., Zavoral, J. H., Emery, R. W. *et al.* (1978): J. thorac. cardiovasc. Surg., **75,** 80.