Hypertrophic cardiomyopathy complicated by complete heart block

Case report and review of the literature

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Summary

A 48-year-old man with symptoms of presyncope and congestive cardiac failure had hypertrophic cardiomyopathy (HCM) without obstruction. Complete heart block (CHB), a rare complication of this disease, was preceded by complete left bundle-branch block. Right ventricular (RV) heart failure was a dominant clinical feature but improved dramatically after temporary transvenous RV pacing prompting the insertion of a permanent RV inhibited pacemaker. Repeated ventricular fibrillation was successfully controlled by amiodarone. This is the seventh case of HCM complicated by CHB reported in the literature, and the first in which RV endomyocardial biopsies were undertaken. Two other patients reported in the literature had RV inhibited permanent pacemakers implanted, and a further 2 had atrioventricular sequential pacemakers.

S Afr Med J 1984; **66**: 847-855.

Case report

A 48-year-old White man noticed the onset of effort-related shortness of breath, dizziness and some tightness in the chest, but no angina. He began to reduce his physical activity drastically, gained weight, and markedly increased his smoking since he was under increasing stress at work. There was no history of hypertension or a previous myocardial infarction, or of hyperlipoproteinaemia, diabetes mellitus, acute rheumatic fever, or gout. The patient's general practitioner referred him to a private cardiologist in Johannesburg. In February 1982 he was normotensive with no evidence of cardiomegaly or cardiac failure; at that time he had a 'short aortic ejection murmur' over the precordium 'compatible with mild aortic stenosis'. A chest radiograph was normal but a resting electrocardiogram (ECG) demonstrated complete left bundle-branch block with quite marked clockwise rotation. On treadmill exercise testing (Bruce protocol) he was unable to manage even stage one; the response of both his blood pressure and pulse rate to exercise were poor. The post-exercise ECG showed no evidence of myocardial ischaemia, although it was difficult to be certain of this in view of the bundle-branch block. Lung function tests revealed some small airways obstruction and features compatible with chronic obstructive pulmonary disease. The cardiologist concluded that there was 'an element of myocardial ischaemia', but was more impressed with the degree of pulmonary disease, obesity and lack of exercise. The patient was prescribed nifedipine 10 mg 3 times daily in addition to a bronchodilator, and advised to discontinue smoking and exercise regularly.

The patient's symptoms remained unchanged and he returned after 6 weeks, when it was decided to undertake cardiac catheterization. This investigation established the presence of moderately severe biventricular cardiac failure. The venous oxygen saturation was 54% and that in the arterial blood 88%. This hypoxia was attributed to chronic obstructive pulmonary disease. Left ventricular (LV) cine angiography showed a 'normal contracting left ventricle and mild mitral regurgitation'. An aortic cine angiogram excluded aortic regurgitation and a pulmonary angiogram delineated a normal pulmonary vasculature. Selective coronary angiography revealed normal coronary arteries. The patient's clinical picture was 'compatible with chronic obstructive pulmonary disease and cor pulmonale'; there was 'no significant left-sided disease, the murmurs being of minimal haemodynamic significance'. The patient was given digoxin and diuretics in addition to nifedipine and the bronchodilator.

During the succeeding year he stopped smoking and continued taking his medication, but there was no really significant improvement in symptoms and progressive swelling of the ankles and abdomen appeared. His right hypochondrium became rather tender and at the beginning of February 1983 he began to have severe dizzy spells, mainly on effort, without actual syncope or shortness of breath. Occasional palpitations were present. A specialist physician again noted complete heart block (CHB) with an idioventricular rate of 42/min (Fig. 1). The patient was then referred to Tygerberg Hospital and admitted to the intensive coronary care unit (ICCU) on 22 February 1983. On admission he had severe oedema of both lower limbs with prominent varicosities, and was centrally and peripherally cyanosed; however, there was no clubbing. The heart rate was about 40/min and the blood pressure 130/80 mmHg. The jugular venous pressure was raised beyond the angle of the jaw with prominent cannon 'a' waves and a positive hepatojugular reflux. All peripheral pulses were palpable and there were no audible bruits. The apex beat was not palpable but there was a most prominent left parasternal heave and epigastric pulsation indicative of right ventricular (RV) enlargement. There was varying intensity of the first heart sound in keeping with CHB, an exceptionally loud pulmonic component of the second heart sound at the pulmonary area, and a prominent RV third sound. A grade 3/6 pansystolic murmur was best heard at the apex and radiated well to the axilla with no variation on respiration, and appeared to be due

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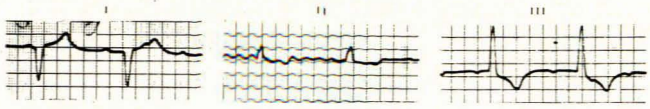

Fig. 1. Resting ECG (leads I, II and III) showing complete atrioventricular block with an idioventricular rate of 42/min.

to mitral regurgitation. There were no diastolic murmurs at the mitral area, but there was a high-frequency grade 2/4 decrescendo murmur audible at the fourth left intercostal space, suggesting aortic regurgitation. The lungs were clinically normal but a tender and soft, non-pulsatile 6 cm hepatomegaly was easily palpable and moderate ascites was evident. There was no splenomegaly.

The differential diagnosis at this stage was of a probable chronic rheumatic mitral and aortic regurgitation haemodynamically aggravated by the onset of CHB about 1 year earlier. There was also the possibility of recurrent pulmonary emboli giving rise to cor pulmonale. The results of sideroom investigations were normal. The blood urea and serum creatinine levels were slightly elevated, possibly through prerenal failure secondary to congestive cardiac failure. The hepatic enzyme and bilirubin values were also elevated and the uric acid level was raised; this was attributed to the chronic diuretic usage as well as possible presymptomatic gout. Another interesting feature was a hypogammaglobulinaemia. Results of haematological investigations were within normal limits. Blood tests for syphilis were all negative, as were the autoagglutination tests. Blood gas values were normal, as were pulmonary function tests. A chest radiograph showed some cardiomegaly with upper lobe pulmonary venous congestion. The ventilation-perfusion scintiscan was negative, making pulmonary embolism most unlikely. Digoxin levels were persistently within the therapeutic range.

At this stage the patient was treated with standard anticardiac failure medication including subcutaneous heparin. An M-mode echocardiogram carried out while the patient was in CHB demonstrated symmetrical ventricular hypertrophy with a hypercontractile LV, but no evidence of systolic anterior movement of the anterior mitral valve leaflet (Fig. 2a). This investigation was repeated with the temporary RV pacemaker switched on and no significant change in haemodynamic parameters could be demonstrated (Fig. 2b). A two-dimensional echocardiogram revealed marked symmetrical LV hypertrophy with massive septal hypertrophy just below the aortic valve, causing 'obstruction' at the end of systole (Fig. 3). A high LV ejection fraction of 77% and signs of LV hypertrophy were detected by means of a gated blood pool technetium-99m scintiscan. Two days later full cardiac catheterization was decided upon. He was in CHB but had had a pervenous prophylactic temporary RV bipolar pacing electrode inserted (Fig. 4, top). While the patient was in the cardiac catheterization laboratory he went into spontaneous ventricular fibrillation (Fig. 4, centre) and had to be electrically defibrillated. Several such episodes responded to conventional therapy. The temporary RV pacemaker was switched on with satisfactory capture (Fig. 4, bottom). Cardiac catheterization was then proceeded with; the intracardiac pressures and haemodynamic indices initially measured with the temporary pacemaker switched off showed markedly elevated pressures and subnormal indices. The temporary RV pacemaker was then switched on, at a rate of 75/min, and the intracardiac pressures and haemodynamic indices were measured again after 10 minutes (Fig. 5). These values were a great improvement on the pre-pacing values. Left ventricular cine angiography in the right anterior oblique (RAO) projection with the temporary pacemaker switched off delineated a hypercontractile and markedly hypertrophied ventricle with apparent hypertrophy

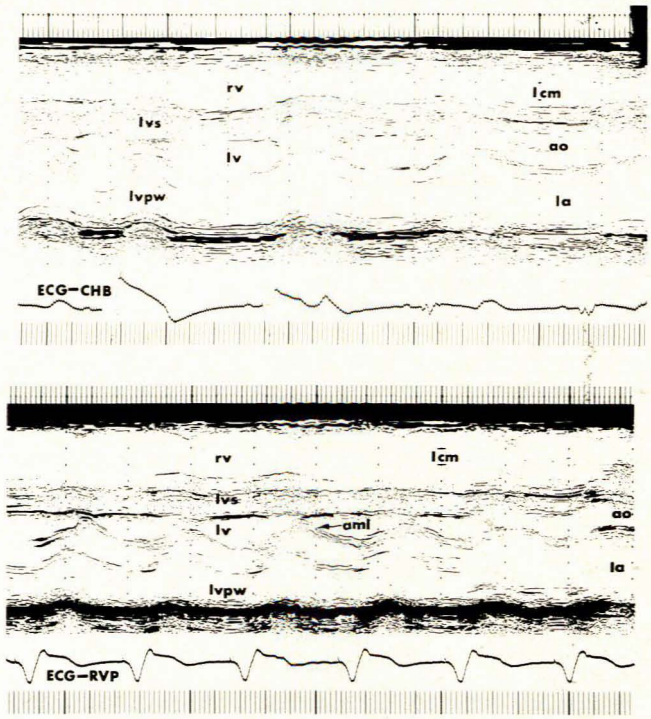

Fig. 2. a — M-mode echocardiogram taken during CHB showing symmetrical ventricular hypertrophy and a supernormal LV ejection fraction; the RV ventricular ejection fraction is also normal. b — M-mode echocardiogram taken during regular temporary RV pacing, showing that no significant haemodynamic change is evident (la = left atrium; ao = aorta; lvpw = LV posterior wall; aml = anterior mitral leaflet; RVP = right ventricular pacing).

of the interventricular septum (IVS) in its mid-portion (Fig. 6). There was an additional mild-to-moderate, non-calcific, mitral regurgitation. RV cine angiography in the shallow RAO view demonstrated a markedly hypertrophied ventricle with somewhat increased generalized contractility. In view of all this the diagnosis of hypertrophic non-obstructive cardiomyopathy (HCM) was now strongly suspected. Multiple RV endomyocardial biopsy specimens were obtained from the apex, free wall and IVS. During catheterization the patient became less co-operative and difficult to manage, probably because of repeated episodes of ventricular fibrillation as well as the significant congestive cardiac failure.

The RV endomyocardial biopsy specimens were processed for light and electron microscopic examination. Myocardial hypertrophy was confirmed by measuring the maximal cell diameter of longitudinally sectioned cells by means of an ocular micrometer. The average cell diameter was 21 μm (normal 10 - 12 μm). On light microscopy one could also see a slight increase in the interstitial connective tissue. On electron microscopy myocytes with a normal parallel orientation of myofibrils were seen (Fig. 7, top), but many cells displayed mild myofibrillar disarray (Fig. 7, bottom), favouring a diagnosis of HCM. Other features included mitochondriosis, prominent lipofuscin granules and very occasional lipid droplets.

In view of the dramatic haemodynamic improvement demonstrated by temporary RV pacing, it was decided to leave the pacemaker switched on at a rate of 75/min. The medication was continued and the patient went on to improve clinically. Another gated blood pool scintiscan within the next 2 days with the temporary pacemaker functioning showed an improved LV ejection fraction of 88% (compared with 77% while he was in CHB). The RV ejection fraction assessed on 24 February was much lower, at 56%. His only setback was temporary loss of short-term memory for a few days after catheterization.

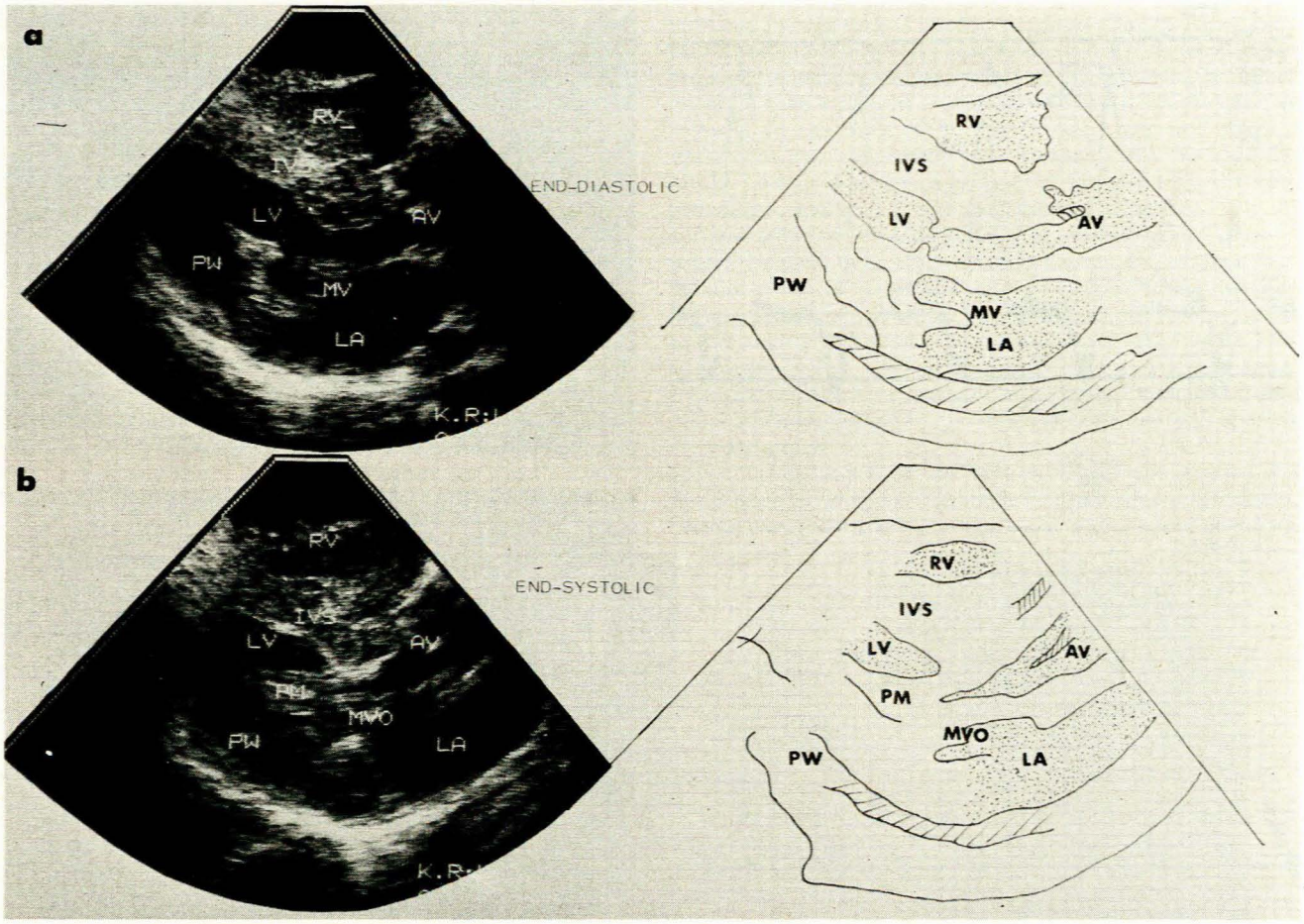

Fig. 3. Two-dimensional (cross-sectional) echocardiogram in (a) end-diastole, and (b) end-systole. Marked symmetrical LV hypertrophy with massive septal hypertrophy just below the aortic valve causing 'obstruction' at end-systole is present (PW = posterior wall of LV; AV = aortic valve; MV = mitral valve; LA = left atrium; MVO = mitral valve orifice; PM = papillary muscle).

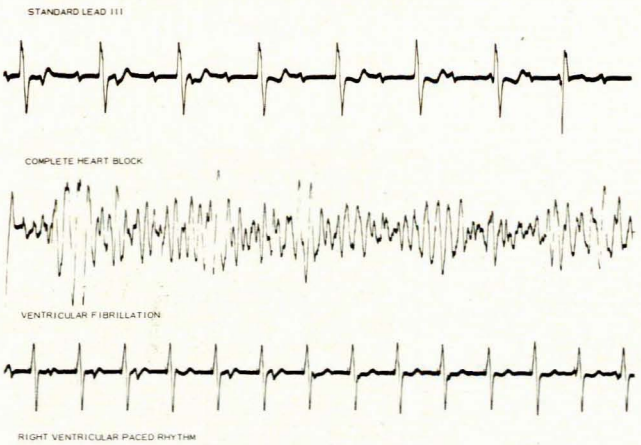

Fig. 4. Rhythm strips (standard lead III) showing (top) CHB; (centre) ventricular fibrillation; and (bottom) RV paced rhythm.

This temporary amnesia was almost certainly related to the resuscitation attempts on the day of cardiac catheterization.

The RV endomyocardial biopsy specimens showed features compatible with HCM which was patchy in distribution (Fig. 7, bottom) since there were areas of apparently normal myocardium (Fig. 7, top). In view of these findings and the episodes of ventricular fibrillation it was decided to prescribe amiodarone (Cordarone-X) 200 mg twice daily after normal thyroid screening tests and slit-lamp examination of the corneas. Diuretic therapy as well as a uricosuric drug were also administered. Repeated 24-hour Holter monitoring failed to show any arrhythmias. Since the patient reverted to CHB when the temporary RV pacemaker was switched off, and since pacing had clearly demonstrated quite a dramatic haemodynamic improvement, on 25 February 1983 it was decided to insert a bipolar permanent pacemaker in the right subclavian region, connected to a RV electrode. ECG showed RV rhythm, as did a rhythm strip. The patient continued to improve clinically and was discharged on 8 March 1983. Later, at the cardiac clinic outpatients' department, he claimed to be feeling markedly improved, with no further dizzy episodes. Holter monitoring sessions demonstrated no arrhythmias. Repeated clinical examination revealed a high-frequency early diastolic murmur heard maximally at the fourth left intercostal space, the cause of which had not been adequately explained.

The patient was readmitted to the ICCU on 26 April 1983 for reassessment after permanent pacemaker insertion and amiodarone therapy for 2 months. New complaints were recurrent superficial skin sepsis and carbuncles. His effort tolerance had improved steadily, as had the swelling of his ankles. He had no palpitations, dizziness or chest pain. Clinical examination revealed very mild ankle oedema, the varicosities on his legs being less prominent. Koilonychia was marked, as were the superficial carbuncles in various stages of development. Cardiovascular examination documented a regular full-volume and rather jerky pulse of about 70/min. The jugular venous pressure was raised to 6 cm and the blood pressure was

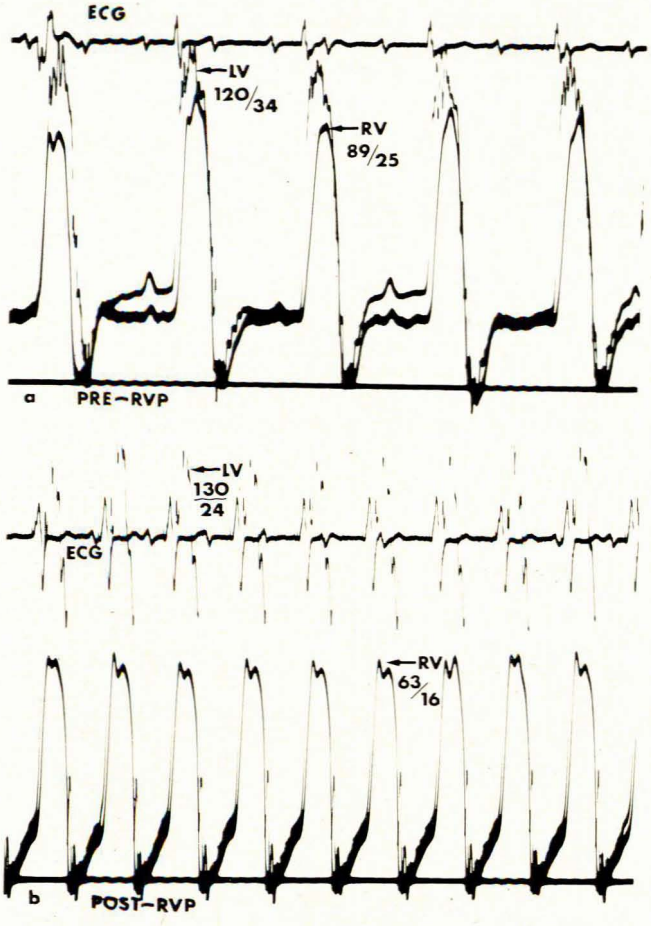

Fig. 5. Simultaneous LV and RV intracardiac pressures: (a) during CHB before RV pacing (pre-RVP) and (b) after RV pacing (post-RVP). A dramatic fall in both RV peak-systolic and end-diastolic pressure is seen, as well as a slight rise in LV peak-systolic pressure and a fall in LV end-diastolic pressure (all pressures measured in mmHg).

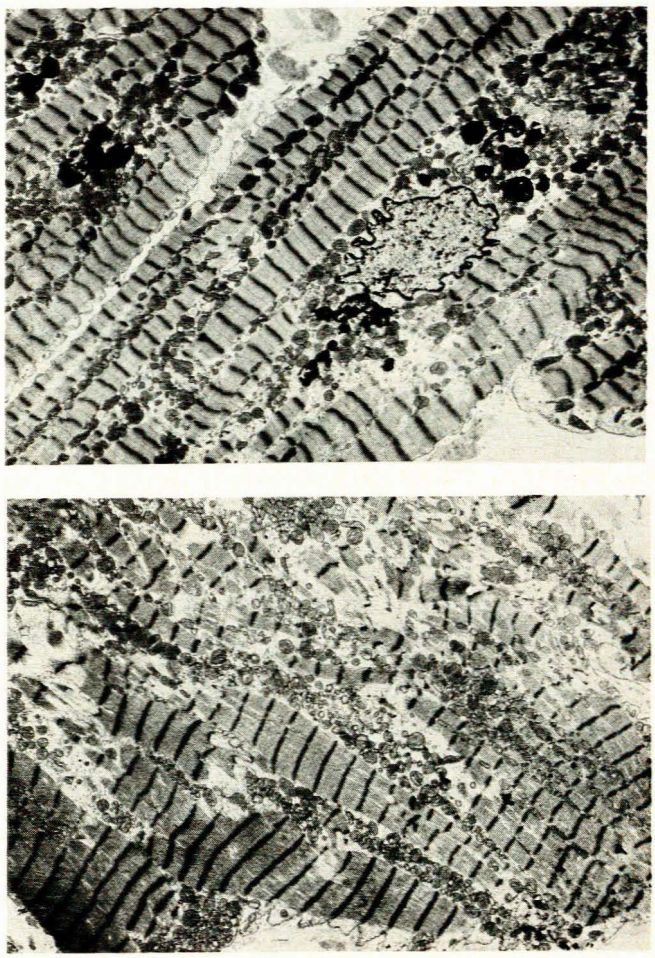

Fig. 7. Electron photomicrographs of RV endomyocardial biopsies. Top: myocardial cell with normal myofibrillar orientation; note mitochondriosis, lipofuscin and crenated nucleus (x 6 400). Bottom: myocardial cell with mild myofibrillar disarray (x 6 200).

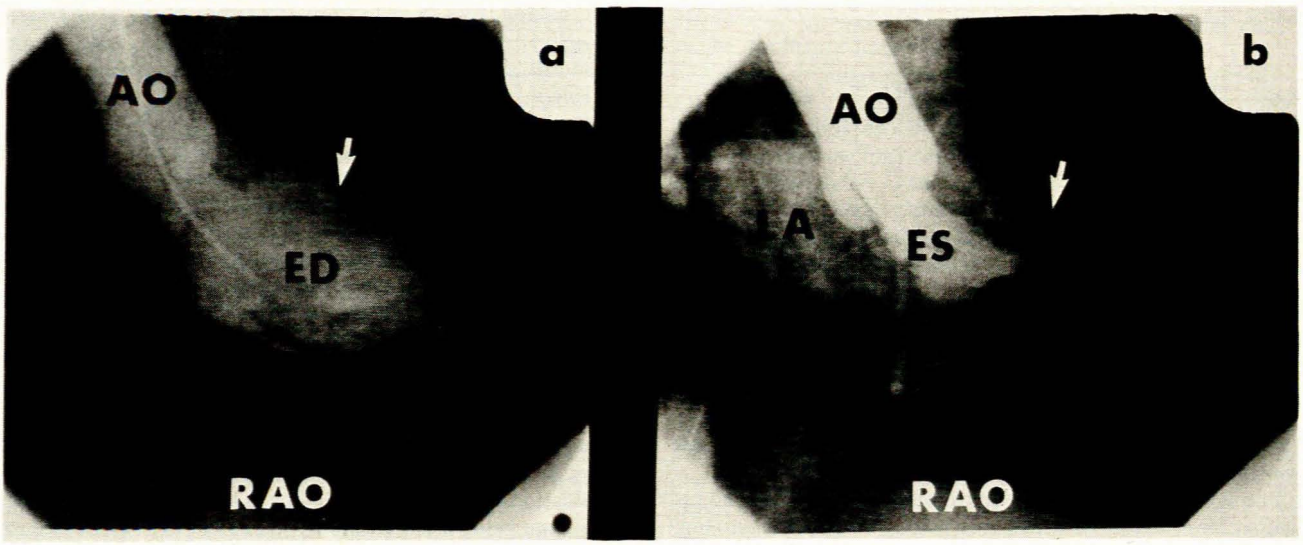

Fig. 6. LV cine angiograms in the RAO projection delineating marked hypertrophy with cavity obliteration at the end of systole. The RV pacing electrode is *in situ* (arrowed). Mild-to-moderate mitral regurgitation is visualized. a — end-diastole (ED); b — end-systole (ES) (LA = left atrium; AO = aorta).

100/60 mmHg. The apex beat was not palpable but there was prominent RV enlargement. A grade 3/6 pansystolic murmur of mitral regurgitation was audible. The grade 2/4 intensity high-pitched early diastolic decrescendo murmur at the fourth left intercostal space was still prominent. Pulmonary hypertension was also clinically detectable. The lungs were clear and examination of the abdomen revealed a 7 cm non-pulsatile and slightly tender hepatomegaly. The rest of the clinical examination was normal.

A chest radiograph confirmed the good position of the permanent pacing electrode and pacemaker function was normal. Holter monitoring did not detect any arrhythmias. Since the patient was on amiodarone, thyroid function tests were carried out and were found to be normal, and a slit-lamp examination failed to show any possible corneal deposits. Repeat technetium-99m scintigraphy documented a LV ejection fraction of 75% (previously 88%), and a RV ejection fraction of 69% (previously 56%). A phonocardiogram recorded on high frequency at the fourth left intercostal space verified the presence of the early diastolic murmur. On 28 April 1983 cardiac catheterization was again undertaken. The intracardiac pressures demonstrated moderately severe biventricular failure no worse than 2 months previously. However, LV function as assessed on the basis of the dp/dt was worse and the RV dp/dt was also abnormally low. Despite these findings the cardiac output was very significantly improved by the permanent pacemaker. LV cine angiography in the RAO (Fig. 8) and left anterior oblique (LAO) (Fig. 9) projections demonstrated marked mid-cavity systolic obstruction without any definite evidence of systolic anterior motion of the anterior leaflet of the mitral valve. Mild-to-moderate, non-calcific, mitral regurgitation was again visualized. An aortic cine angiogram in the LAO projection failed to show any aortic regurgitation. Selective coronary angiography documented only minor internal luminal irregularities of both the right and left coronary artery.

At this stage it was decided that the mitral regurgitation was not of such severity to necessitate a mitral valve replacement,

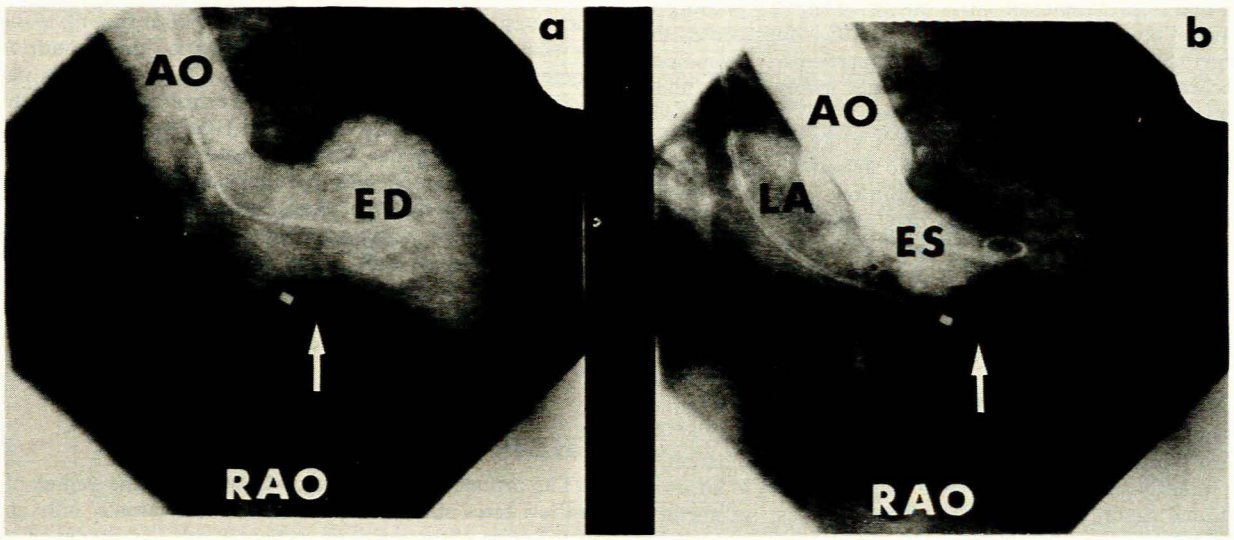

Fig. 8. LV cine angiograms in the RAO projection showing marked hypertrophy with mid-cavity obstruction. There is moderately severe mitral regurgitation. The permanent RV bipolar pacing electrode is arrowed. a — end-diastole (ED); b — end-systole (ES) (LA = left atrium; AO = aorta).

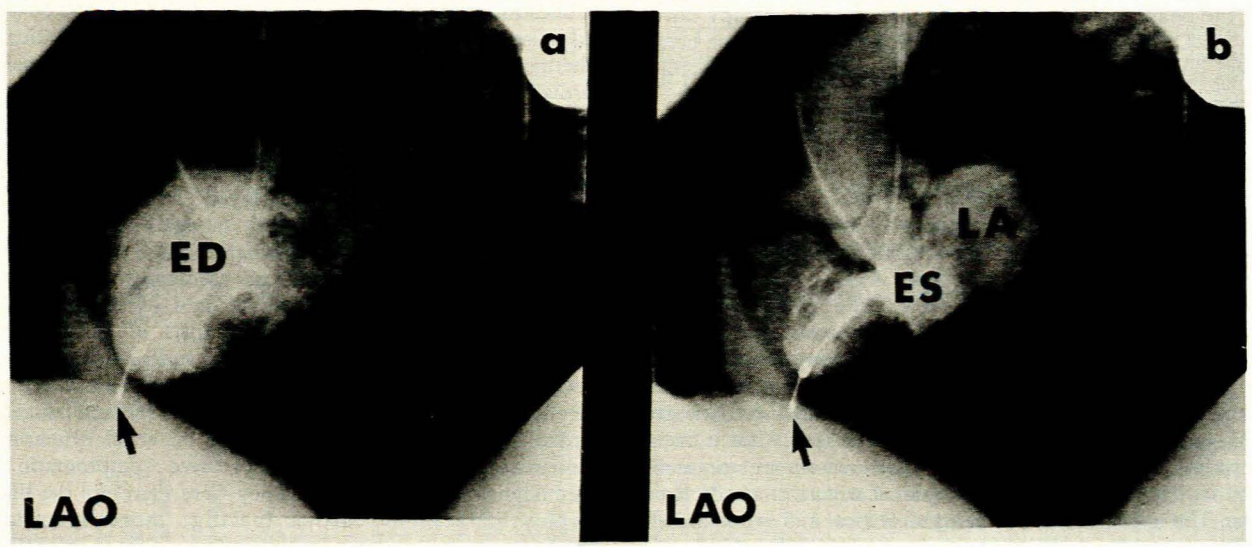

Fig. 9. LV cine angiograms in the LAO view demonstrating severe symmetrical hypertrophy with end-systolic cavity obliteration. There is no systolic anterior motion of the anterior mitral valve leaflet but moderately severe mitral regurgitation is present (LA = left atrium). The permanent RV bipolar pacing electrode is arrowed. a — end-diastole (ED); b — end-systole (ES).

and the patient was discharged on amiodarone, diuretics and a uricosuric agent. Since then he has been seen on several occasions and is well enough to return to work.

Discussion

General aspects

Idiopathic HCM[1-3] has multiple guises,[4,5] and there are still several unanswered questions, particularly relating to the natural history,[6-8] correct form of therapy and prognosis.[9,10] So-called 'familial' and 'sporadic' types are recognized. It is generally accepted that there are two haemodynamic types, the non-obstructive form (referred to as 'hypertrophic cardiomyopathy' or HCM) and the obstructive variety (termed 'hypertrophic obstructive cardiomyopathy' or HOCM). Goodwin prefers to use the term 'hypertrophic cardiomyopathy with or without obstruction'.[3] Some patients with HOCM have a resting gradient, but in others this may have to be provoked by procedures such as the Valsalva manoeuvre and drugs like isoproterenol or amyl nitrite which can produce a 'functional obstruction'.[11,12] Classic HOCM is represented by asymmetrical septal hypertrophy (ASH), particularly prominent just below the aortic valve but sometimes involving the mid-portion of the IVS or even the apical part.[13,14] Varying degrees of hypertrophy of the free wall of the LV are often present,[15] especially if there is concomitant aortic stenosis.[16] Involvement of the RV and obstruction to RV outflow have been documented.[17,18] Mitral regurgitation may aggravate the haemodynamic findings and lead to LV dilatation which can sometimes resemble dilated (congestive) cardiomyopathy.

Thus, there is a wide pathological spectrum. Symptoms are numerous: lassitude, dyspnoea, chest pain (both atypical and typical angina pectoris), palpitations, dizziness and syncope, or the patient may be completely asymptomatic. One complication is sudden death, almost certainly due to a ventricular arrhythmia.[19-21] Drug therapy has essentially consisted of ß-blockers[22] and calcium antagonists, alone or in combination.[23] More recently amiodarone has been used, primarily as an anti-arrhythmic agent, with what appears to be most satisfactory results.[24] Disopyramide can diminish the resting gradient in HOCM and may be beneficial over the long term.[12,25] Digoxin is contraindicated because it is known to increase the gradient across the LV outflow tract. Surgical intervention has involved such procedures as LV myotomy and myectomy.[26,27] Nevertheless, the Mayo Clinic results with this form of therapy have been most gratifying.[28] Mitral valve replacement has also been carried out when the mitral regurgitation has been of significance.

Echocardiographic controversy

In the past M-mode echocardiography has often been cited as pathognomonic of HCM, particularly if it was obstructive in nature.[29] Diagnostic features were ASH, systolic anterior movement of the anterior mitral valve leaflet and mid-systolic closure of the aortic valve,[30] but it soon became apparent that hypertensive heart disease and ischaemic heart disease could also produce some of these features. Some workers have attempted to use echocardiography to distinguish between HCM and hypertensive heart disease, but results have been uncertain.[31] The inaccuracy of this non-invasive technique in diagnosis has been shown by occasional negative echocardiograms. The appearance of both asymmetrical and symmetrical LV hypertrophy in families with known HCM[32] adds more confusion to its definition; there is in fact a wide spectrum of genetically transmitted HCM.[4,5,32] The ultrasound beam in M-mode echocardiography sometimes does not traverse the thickest portion of the IVS, thus giving a falsely reduced IVS:LV posterior wall ratio and leading to a diagnosis of symmetrical (or concentric) hypertrophy, as in our patient. However, two-dimensional (cross-sectional) echocardiography clearly showed the disproportionate ventricular septal hypertrophy just below the aortic valve; others have emphasized this discrepancy.[15,33]

Endomyocardial biopsy assessment

The role of endomyocardial biopsy in diagnosis has been controversial, and most researchers believe that there are no pathognomonic features.[34] Nevertheless, interpretation of histological findings becomes more meaningful if the clinical, ECG, echocardiographic, haemodynamic and cine angiographic features are considered. Light microscopy usually shows markedly hypertrophied and irregularly arranged myocardial fibres ('myofibrillar disarray'), which contain bizzarely shaped nuclei often surrounded by a clear area ('perinuclear halo').[35] Varying amounts (usually minor) of interstitial fibrosis are also encountered. A further nonspecific histological finding is that of an increase in the number of mitochondria ('mitochondriosis'). Some authors have claimed that in HCM there is a patchy distribution of the myocardial abnormality, whereas this is usually confined to the IVS in the HOCM variety;[36,37] others disagree.[38] There has been controversy about accuracy of results of RV endomyocardial biopsies in diagnosis, due to a common belief that the disease only involves the LV. This misconception is highlighted by the fact that RV obstruction has been demonstrated,[6,17,18] as in our case. Most workers only perform RV biopsies in suspected cases and find the same histopathological features as seen on LV biopsy.[39] Biopsy findings cannot, however, be interpreted in isolation since they are insufficiently specific.

CHB in HCM

The influence of varying cycle length on the degree of LV outflow tract obstruction in HOCM was initially only appreciated after observations in patients with low grades of atrioventricular heart block.[40-42] The appearance of third-degree block or CHB is rare, and only 6 previously documented cases have been found in the literature (Table I). The first description was of a 10-year-old boy[43] (1965), followed in 1973 by the publication by Matlof *et al.*[40] of a report of CHB in a 60-year-old man with HOCM. Matlof *et al.*[40] gave no indication whether their patient was treated by insertion of a permanent cardiac pacemaker. The third case was in a 71-year-old man, described in 1975 by Johnson and Daily.[44] He had right bundle-branch block, left axis deviation and first-degree atrioventricular block pre-operatively. A temporary transvenous ventricular pacemaker was employed, but this resulted in cardiogenic shock. During surgery he again went into shock with the onset of acute atrial fibrillation; with digoxin administration further deterioration in his cardiovascular status took place. Further evaluation (clinically and by M-mode echocardiography) established the diagnosis of HOCM. The patient's condition deteriorated during ventricular pacing and it was therefore decided not to insert a permanent ventricular pacemaker; 6 months later he returned in CHB and congestive cardiac failure. The authors inserted an atrial synchronous ventricular pacemaker with impressive symptomatic and haemodynamic improvement. They were therefore the first to employ permanent cardiac pacing, more specifically synchronous atrioventricular pacing, in a case of HOCM complicated by CHB.

Chmielewski *et al.*[45] in 1977 described a fourth case in a 35-year-old man who presented with recurrent syncope. A

TABLE I. CHB IN HCM

Author	Year	Age of the patient (yrs)	Sex	Symptoms	Conduction before CHB	Permanent pacemaker inserted	Resting gradient (mmHg)	Provoked gradient (mmHg)	Coronary angiogram	Surgery	Outcome
Luisada[43]	**1965**	**10**	**M**	**Nil**	**Normal**	**Nil**	**Nil**	**27**	**Not done**	**No**	**Alive**
Matlof *et al.*[40]	**1973**	**60**	**M**	**Syncope, fatigue**	**Unknown**	**Unknown**	**33**		**Not done**	**No**	**Alive**
Johnson and Daily[44]	**1975**	**71**	**M**	**CCF**	**RBBB, LAD and 1° AVB**	**AVS**	**Nil**	**95**	**Not done**	**No**	**Alive**
Chmielewski *et al.*[45]	**1977**	**35**	**M**	**Syncope**	**Intermittent LBBB and Mobitz II 2° AVB**	**AVS**	**Nil**	**76**	**Normal**	**No**	**Alive**
Spilkin *et al.*[46]	**1977**	**20**	**M**	**Presyncope**	**RBBB, LAD and 1° AVB**	**Ventricular**	**64**	**—**	**Normal**	**No**	**Alive**
Koide *et al.*[5]	**1982**	**49**	**F**	**CCF**	**A. fib.**	**Ventricular**	**Unknown**	**Unknown**	**Not done**	**No**	**Dead**
Przybojewski *et al.* (present study)		**48**	**M**	**Presyncope, CCF**	**LBBB**	**Ventricular**	**Nil 11 (RV)**	**Not done**	**Normal**	**No**	**Alive**

CCF = congestive cardiac failure; RBBB = right bundle-branch block; LAD = left axis deviation (possible left anterior hemiblock); 1° AVB = first-degree atrioventricular block; LBBB = left bundle-branch block; 2° AVB = second-degree atrioventricular block; A. fib. = atrial fibrillation; AVS = atrioventricular sequential.
Gradients refer to the LV outflow tract unless otherwise indicated.

permanent sequential atrioventricular cardiac pacemaker was inserted, after which the patient remained asymptomatic at 6-months' follow-up. In the same year Spilkin *et al.*[46] documented the case of a 20-year-old man who had increasing dyspnoea on effort and significant precordial pain. The patient had clinical signs of HOCM confirmed on M-mode echocardiography. A resting ECG revealed first-degree atrioventricular block, right bundle-branch block and marked left axis deviation (presumably left anterior hemiblock). The patient responded well to a ventricular inhibited pacemaker but these workers suggest that an atrial synchronous ventricular pacemaker would be more beneficial in cases with a poorly compliant LV, as seen in elderly patients with chronic disease. Koide *et al.*[5] documented the case of a 49-year-old woman who had had cardiomegaly for 15 years and died from a cerebral embolism 6 years after going into atrial fibrillation, at which stage an M-mode echocardiogram revealed moderate symmetrical LV hypertrophy as well as dilatation of most of the heart chambers. About 6 months before her death she went into CHB which necessitated the insertion of a permanent ventricular inhibited pacemaker. Autopsy documented all the features of HCM.

Our patient had features of HCM. This diagnosis is based upon echocardiography (both M-mode and two-dimensional), haemodynamic characteristics as determined by cardiac catheterization, cardiac cine angiography and multiple RV endomyocardial biopsies. The complication of CHB caused presyncope and severe deterioration in haemodynamic status. He had had complete left bundle-branch block approximately 1 year previously and it is most likely that this conduction defect progressed to CHB. The insertion of a permanent ventricular inhibited pacemaker gave rise to a marked improvement in haemodynamics as evidenced by M-mode echocardiography, gated blood pool scintigraphy and cardiac catheterization findings before and after temporary RV pacing, as well as after implantation of the permanent pacemaker. Thus, this is the seventh documented case of HCM complicated by CHB, and the third with a permanent RV inhibited pacemaker. Of the remaining 4 patients 2 underwent insertion of a permanent atrial synchronous ventricular pacemaker. It may be argued that the finding of CHB is completely coincidental and due to congenital heart block or ischaemic heart disease. Nevertheless, the onset of CHB in a patient with HCM can cause both significant symptomatic as well as haemodynamic deterioration, and recognition of this complication and management with either a ventricular inhibited or atrial synchronous ventricular pacemaker may prove life-saving. Perhaps the clinician should be particularly concerned at the risk of the development of CHB in a patient known to have HCM who has additional complete left bundle-branch block.

Diastolic murmurs in HCM

Diastolic murmurs in HCM have always been accepted as being unusual, most of these murmurs being attributed to associated conditions such as rheumatic mitral stenosis and atrial septal defect. Our patient had severe pulmonary hypertension and the possibility of pulmonary incompetence must therefore also be considered as being a possible cause of the early diastolic murmur. However, the murmur did not quite behave in such a way to suggest this mechanism. Because of the slight resting gradient across the RV outflow tract, functional tricuspid stenosis could possibly be envisaged, but the mild degree of this obstruction and the early diastolic character of the murmur would mitigate against this possibility.

Right heart failure

The appearance of signs of RV failure in patients with HCM can lead to confusion in the making of the clinical diagnosis. This sequence of events probably contributed to a diagnosis of cor pulmonale being made about a year before the patient's admission to Tygerberg Hospital. Goodwin and Oakley[47] claimed that preterminally, HCM can masquerade clinically as dilated (congestive) cardiomyopathy, and a similar incorrect antemortem diagnosis of dilated cardiomyopathy was made by Koide *et al.*[5] in a patient shown to have histological features of HCM at autopsy. Some cases of HOCM develop LV dilatation and only then ASH.[48] Olson[36] described 5 patients who developed preterminal ventricular dilatation and in whom a clinical diagnosis of dilated cardiomyopathy was disproved on postmortem examination, where features of HCM were demonstrated. Unfortunately, the appearance of significant right-sided cardiac failure in our patient is almost certainly a poor prognostic sign.

Complication of ventricular fibrillation

Ventricular fibrillation as a cause of cardiac arrest has been documented only rarely in HCM.[9,19,26] Ciró and Maron[49] reported a patient with HCM and cardiac arrest secondary to ventricular fibrillation 16 years before death; he had a family history of early sudden deaths due to HCM. Our patient was also successfully resuscitated from frequent episodes of ventricular fibrillation while awaiting cardiac catheterization at Tygerberg Hospital. Whether he will also survive for a substantial number of years seems unlikely considering the degree of RV failure. Previously documented patients with ventricular fibrillation complicating HCM were not in CHB, which could also worsen the prognosis. The only possible therapeutic intervention if our patient's clinical condition deteriorates is mitral valve replacement. Myotomy with or without myectomy would not seem appropriate in his case since no haemodynamic obstruction could be demonstrated.

We wish to thank sincerely Miss H. Weymar of the Cardiology Unit, Tygerberg Hospital, for preparing the manuscript and some of the illustrations. Thanks are also due to Mr Chris Wilberforce, Head of the Department of Photography, for his painstaking preparation of the photographs. Finally, we thank Dr J. P. van der Westhuyzen, Chief Medical Superintendent of Tygerberg Hospital, for permission to publish.

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True hermaphroditism

A case report with observations on its bizarre presentation

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Summary

A true XX hermaphrodite presenting in adulthood with male sex gender identity was found to have separate vaginal and urethral openings in the perineum. A total vaginectomy was performed at the same time as total abdominal hysterectomy and gonadectomy because the vaginal opening would interfere with a urethroplasty to repair the hypospadias and chordee phallus. During this procedure prostatic tissue was excised in the area of the anterior vaginal wall adjacent to the urethra.

Recommendations regarding this rare XX true hermaphroditism are put forward and observations on some bizarre features of the entity are made. The authors plead for the formation of a national register to study this interesting condition.

S Afr Med J 1984; **66:** 855-858.

True hermaphroditism is highly prevalent in the indigenous Black population of South Africa.[1] The condition has been extensively reported, but because of its random presentation at the different levels of the genital tract the guidelines laid down for management cannot always be followed; these often have to be modified according to the individual case.

There is no available literature relating the ratio of ovarian to testicular tissue (O:T ratio) and the accompanying degree of malformation, nor has anyone been able to explain the bizarre way in which one region of the müllerian system can be underdeveloped while other parts are fully developed. An accurately kept national register might throw light on the mechanisms involved.

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Case report

A 26-year-old man was referred to Hillbrow Hospital as a case of intersex. He had applied for work as a migrant labourer; he had previously been employed as a labourer in Transkei where he had been born.

The patient's outward appearance when clothed was basically masculine, but physical examination revealed well-developed female breasts and a slightly rounded contour together with a well-developed musculature, hypospadias, a penis in marked chordee and an absent prepuce (ritual tribal circumcision had been performed in childhood) (Fig. 1). There was a bifid empty scrotum with separate vaginal and urethral openings in the perineum. A well-developed uterus could be felt on rectal examination. Menstrual blood was issuing from the vagina and the patient reported that he had menstruated monthly for the previous 10 years.

The results of the hormone profile are shown in Table I; all values were within the female range. An ECG, a chest radiograph and a skull radiograph were normal. The full blood count, blood urea value, serum electrolyte and creatinine values, thyroid function, the serum cortisol value and liver function were also within normal limits. Karyotyping was done by chromosome analysis of peripheral blood metaphases and revealed a model 46,XX karyotype in 27 cells analysed. No Y chromosome was revealed in a further 30 cells examined with fluorescent techniques. H-Y antigen analysis was not performed, since it is not available in South Africa.

After full psychiatric assessment and a confirmatory laparoscopy, surgical correction in consecutive stages to convert the patient more fully into a male was initiated. The first procedure carried out was a total abdominal hysterectomy and gonadectomy, total vaginectomy, and first-stage repair of the hypospadias by lysis of the chordee to achieve good penile length.

The uterus was of normal size (Fig. 2). The right adnexa consisted of a normal patent fallopian tube and an ovary in which there was a corpus luteum. On the left side there was a gonad on a small pedicle and a normal-sized fallopian tube with the fimbrial end occluded.