HYPERTROPHIC CARDIOMYOPATHY IN NOONAN'S SYNDROME — A CASE REPORT

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SYNOPSIS

A 10 year follow-up of a case of hypertrophic cardiomyopathy in Noonan's Syndrome is reported. The management is general of the cardiovascular complications are discussed.

INTRODUCTION

The clinical and cardiovascular manifestations in Noonan's Syndrome have been extensively reviewed (1,2). The most common cardiac abnormality is valvar pulmonic stenosis.

With the advent of echocardiography, involvement of the left ventricle in particular asymmetrical septal hypertrophy have been found to be not uncommon (3).

The present case is of particular interest as it is a case of hypertrophic cardiomyopathy with documented significant left ventricular outflow tract gradient which has been followed up for a decade.

CASE REPORT

C.Y.P. was referred to the Tan Tock Seng Hospital in 1976 for evaluation of asymptomatic heart murmur. He was then 12 years of age.

He was a full term baby. he apparently was noted to be a delicate child since infancy and on entering school and was found to be mentally dull. IQ testing showed a level of 66. He also developed autistic behaviour and dropped out of school in 1976.

He had right orchidopexy and herniotomy for undescended testes in 1971.

He is the youngest in a family of 8 children. There was no family history of any cardiovascular disease.

Clinical assessment in 1976 showed a child with short stature and dysmorphic facies. He had antimongoloid slant of the eyes and prominent epicanthic folds. He had low set ears, low hairline and webbing of the neck (Fig. 1). He had cubitus valgus.

Cardiac examination revealed a normal sized heart with a left ventricular impulse, and an ejection systolic murmur of grade 3/6 intensity at the left sternal edge.

Baseline investigations were carried out and in 1977 his family members consented to a cardiac catheterisation. The pressures are shown in the Table, the significant findings being the presence of substantial outflow tract gradients in both ventricles especially the left. He was started on beta-blockers in the hope that this will decrease the pressure gradients (4).

He remained well through the years and in Class I (New York Heart Association Functional Classification).

However, serial clinical assessment showed gradual cardiomegaly which was obvious in the ECG (Fig. 2) and CXR (Fig. 3). Echocardiographic features aside from documenting hypertrophic cardiomyopathy (Fig. 4 & 5) also provide distinctive featurs of left ventricular outflow tract obstruction (Fig. 6).

Repeat cardiac catheterisation was carried out in September 1986 which revealed a higher left ventricular outflow tract gradient (Table 1, Fig. 7). The RVOT gradient was normal. We suspected that the increased thickness resulting in stiffness of the mid and basal interventricular septum might have abolished the Bernheim effect.

He was subsequently put on Amiodarone as he was documented to have asymptomatic ventricular ectopics.

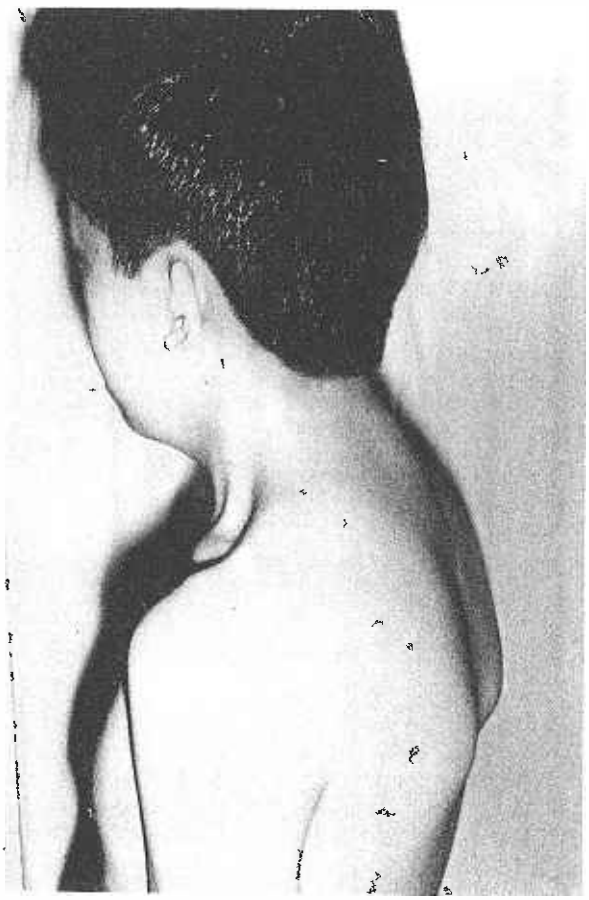

Fig. 1: The low set ears, low hairline and webbing of the neck are characteristic of Noonan's syndrome.

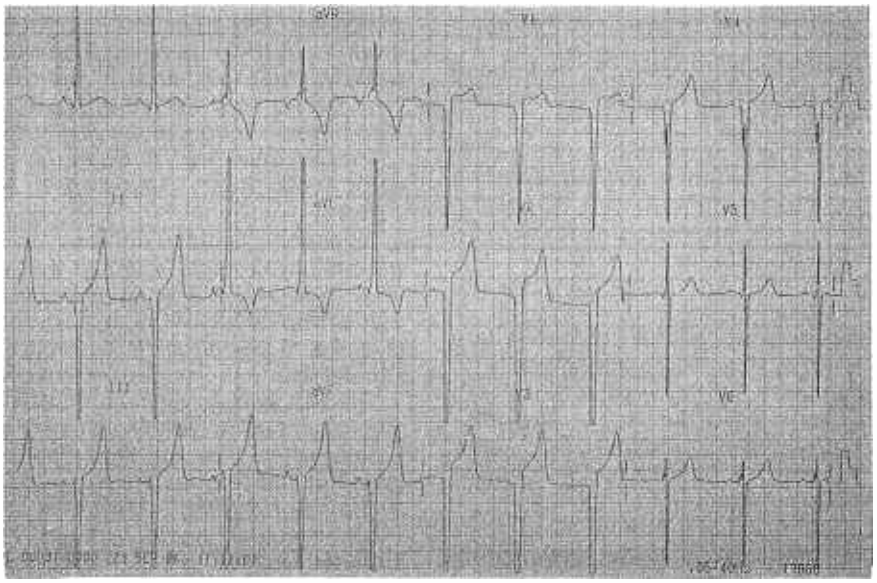

Fig. 2: The ECG shows typical QRS voltage change of left ventricular hypertrophy.

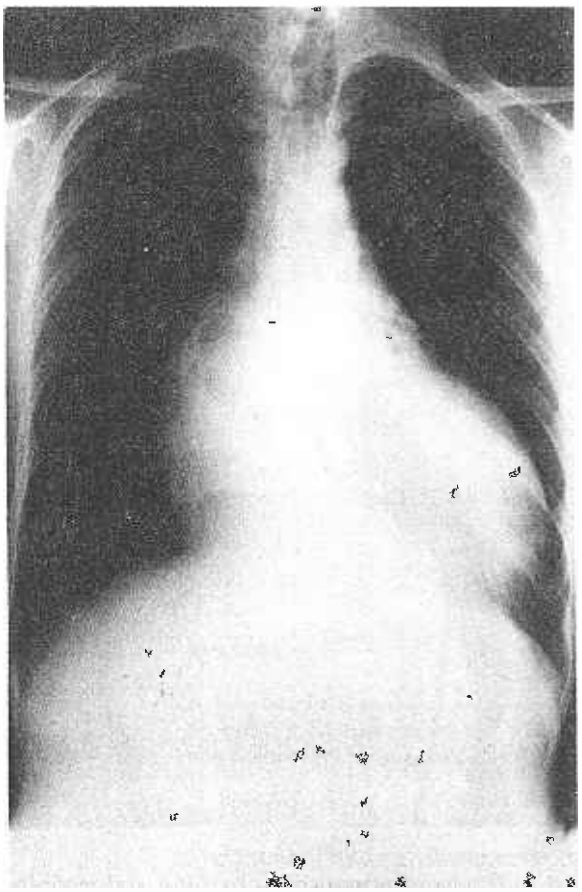

Fig. 3: The CXR shows an increased cardiac silhouette. Since LV cavity dilatation is unusual, this suggests massive, generalised ventricular wall thickening and atrial dilatation.

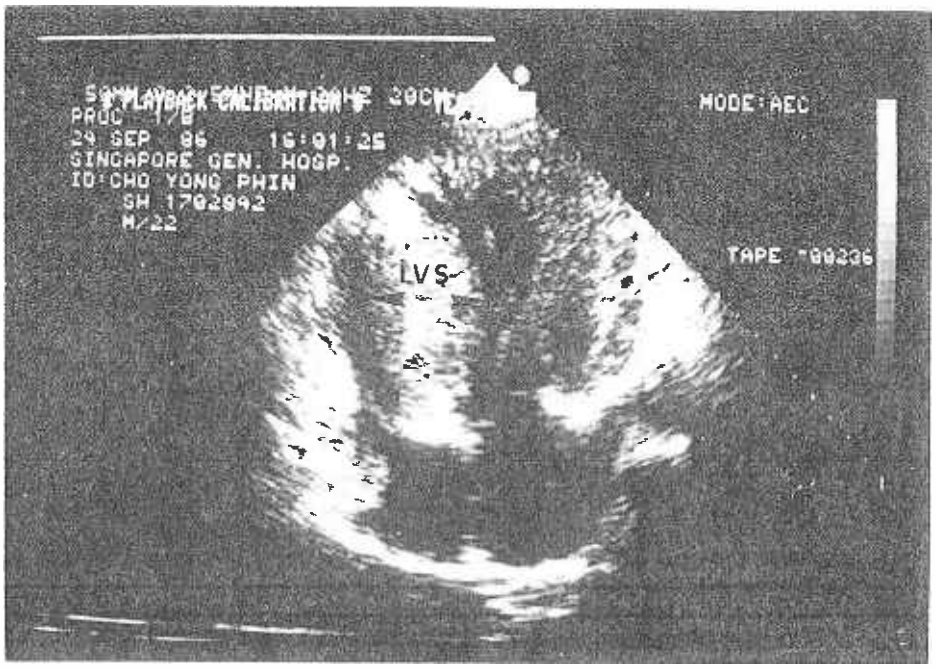

Fig. 4: Left parasternal long axis view (2D Echo) shows the thickened interventricular septum (ivs). The ratio of the septum to posterior left ventricular wall is 1.7.

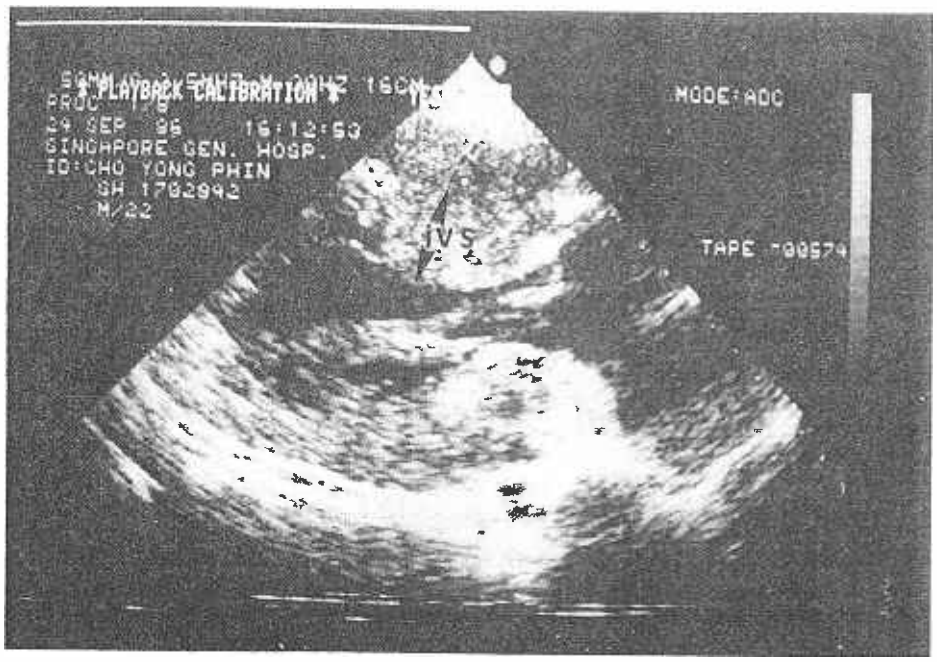

Fig. 5: Apical 4-chamber view (2D Echo) again showing the thickened ivs (arrowed) as well as the dilated left atrial cavity.

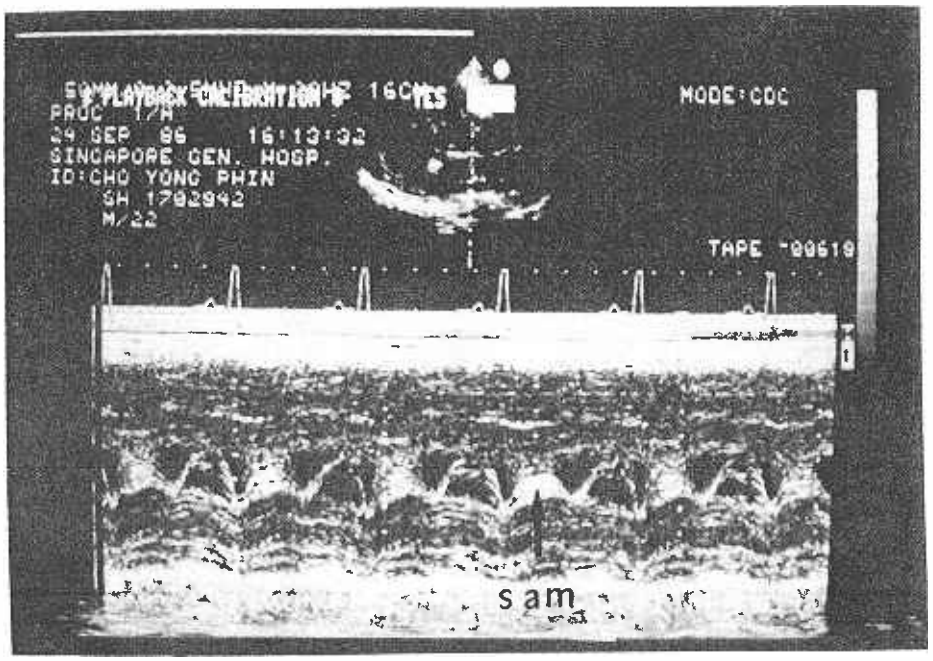

Fig. 6: M-mode Echo at the level of the mitral valve showing the systolic anterior motion of the anterior mitral leaflet (SAM). This suggests outflow tract obstruction.

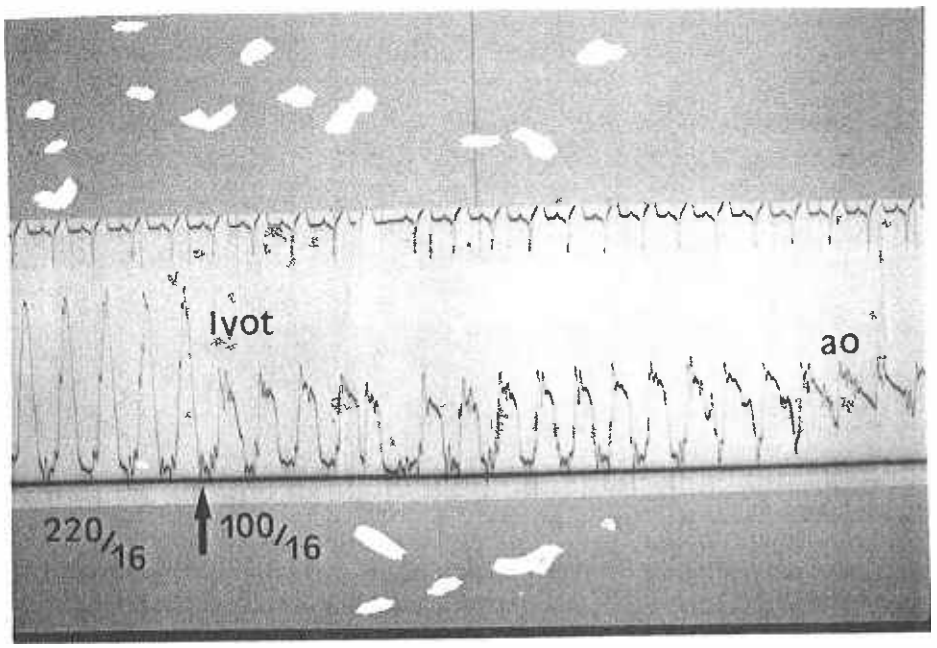

Fig. 7: Cardiac catherization reveals a significant pressure gradient of 120 mm Hg on catheter withdrawal from left ventricular outflow tract (lvot) to the aorta (ao). Arrow denotes site of outflow tract opbstruction.

TABLE 1
CARDIAC CATHETERISATION DATA

	1977	1986
RA	7	4
RV	83/0-2	24/4
	30/8	
PA	30/10	25/10
LV	140/38	220/16
	58/22	100/16
AO	60/50 58	90/50 62
Gradient	80 mm	120 mm

DISCUSSION

We report a 10-year followup of a case of Noonan's Syndrome associated with hypertrophic cardiomyopathy.

Although Nora et al reported an incidence of 32% of his case-studies of Ullrich-Noon syndrome having asymmetrical septal hypertrophy on echocardiography; none of his cases showed systolic anterior motion of the mitral valve and presumably any significant LV outflow tract gradient.

There are few reports in the literature of hypertrophic cardiomyopathy with LVOT obstruction associated with Noonan's syndrome. From a review of all documented cases with both these entities, our patient appear to be the case with the highest pressure gradient recorded. Beta-blockade may ameliorate the obstruction (5,6) although other cases showed further rise in gradient with rapid deterioration and death (7).

Our patient did not show a satisfactory response to Propranolol yet remained asymptomatic for a decade. With the data from his recent cardiac catheterisation showing a basal left ventricular outflow systolic pressure gradient of 120 mm we are considering surgical means to relieve the obstruction. The Mayo Clinic has the largest study on surgical management of hypertrophic cardiomyopathy (8). In this group of 40 patients the mean basal pressure gradient was 79 mmHg.

The proponents of partial septal myomectomy suggest a favourable effect of surgery on survival in this subset of cases. This must be weighed against the contention that the LVOT gradient is abolished because of reduction in ventricular performance from myocardial damage and hence is deleterious (9). The operative mortality of 5-10% has also to be considered.

Another consideration in our patient is his age of presentation which albeit asymptomatic carried with it a higher risk of sudden death (10). Todate no pharmacological agent have been proven conclusively to prevent this although Amiodarone shows promises in the suppression of ventricular tachyarrhythmias in such cases (11). We hope it will be effective in our patient.

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