Familial occurrence of mitral valve prolapse: is this related to the straight back syndrome?

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SUMMARY Familial prevalence of mitral valve prolapse in a Chinese population was determined in 22 propoiti of whom 10 had straight back (group A), three had abnormally high metacarpal index (group B), and nine had neither (group C). Of 71 (32 male and 39 female subjects) first degree relatives screened, mitral valve prolapse was found in 19 (seven male and 12 female subjects) (26.8%). The familial prevalence among groups A, B, and C was 20%, 30%, and 38.5%, respectively. Our study indicates that the familial occurrence of mitral valve prolapse does not depend on its association with the straight back syndrome.

The familial occurrence of idiopathic mitral valve prolapse has been well described.¹⁻¹⁰ It has been suggested that the condition is transmitted in an autosomal dominant manner with incomplete penetrance, the expressivity among female subjects appearing to be more complete.⁹ In another recent study on patients with straight back syndrome, mitral valve prolapse was found in 67%.¹¹ The straight back syndrome was considered an autosomal dominant condition with the antigenic determinants possibly located on chromosome 6. It is thus possible that the familial occurrence of mitral valve prolapse is the result of its association with the straight back syndrome. This possibility, not previously studied, is the subject of the present paper.

Subjects and methods

A family study was carried out in 22 patients with idiopathic mitral valve prolapse. None of the 22 suffered from the Marfan's syndrome. The diagnosis of mitral valve prolapse was based on one or more of the following criteria.

(1) The presence of non-ejection systolic click(s) (with or without a late systolic murmur) in the absence of other causes such as a small ventricular septal defect with aneurysm of the interventricular septum,^{12,13} a left sided pneumothorax,¹⁴ a calcified left atrial myxoma,¹⁵ hypertrophic obstructive cardiomyopathy,¹⁶ a previous mitral valvotomy,¹⁷ and pneumoperitoneum.¹⁸

(2) The presence of mid-systolic buckling^{19,20} or pansystolic bowing greater than 3 mm²¹ in the M-mode echocardiogram.

First degree relatives of the 22 patients, who were willing to participate, were studied. Each had a complete history and physical examination and M-mode echocardiography. Auscultation of the heart was performed in various positions (supine, left lateral decubitus, sitting, and standing) and with the Valsalva manoeuvre. M-mode echocardiograms were recorded on Kodak linagraph direct print paper using an E for M VR12 recording system and a 2.25 MHz transducer. In the echocardiographic examination, the transducer was positioned in a vertical manner so as to avoid false positive pansystolic bowing caused by excessive inferior angulation.^{9,22-24}

Radiological examinations were carried out in the 22 propoiti. Posteroanterior and lateral chest radiographs were taken with the patient erect and in full inspiration. A posteroanterior radiograph of the left hand was also taken with the forearm in pronation and fingers extended. From the radiographs, the following information was obtained.

(1) The ratio of the anteroposterior diameter to the transverse diameter of the thorax. The anteroposterior diameter of the thorax was measured, on the lateral radiograph, from the posterior border of the sternum perpendicular to the anterior surface of the eighth thoracic vertebral body. The transverse diameter of the thorax was measured at the level of the diaphragms on the frontal radiograph. A straight back was diagnosed when the ratio was more than 2 standard deviations (SD) below the normal mean value

(mean $\pm$ SD being 0.437 ± 0.042 for normal Chinese men and 0.440 ± 0.041 for normal Chinese women).²⁵

(2) The metacarpal index of the left hand as described by Walker and Ashcroft.²⁶ A metacarpal index more than 2 SD above the normal mean value was considered abnormal (mean $\pm$ SD being 7.47 ± 0.48 for normal Chinese men and 7.89 ± 0.48 for normal Chinese women).²⁵

(3) Various thoracic skeletal abnormalities were noted.

Results

Of the 22 propoiti, there were nine male and 13 female subjects. The mean age was 26.5 years (range 14 to 48 years). The diagnosis was based on the presence of non-ejection systolic click(s) in three, of echocardiographic features of prolapsed mitral valve in 14, and the presence of both in five. On auscultation, four had isolated click(s), nine had a systolic murmur at the apex, four had click(s) and a systolic murmur, two had an ejection systolic murmur at the aortic or pulmonary area, one had a precordial systolic honk, and two had no abnormal auscultation signs. A total of 71 first degree relatives (32 male and 39 female subjects) were screened. Mitral valve prolapse was found in 19 (seven male and 12 female), giving a familial prevalence of 26.8%. This was significantly higher ($p < 0.001$, χ^2) compared with the 7.9% prevalence in the normal Chinese population.²⁷

Of the 22 propoiti, 10 had a straight back, three had an abnormally high metacarpal index, and nine had neither of the two. None had both skeletal abnormalities (Table 1). The familial prevalence of mitral valve prolapse in these three groups of propoiti were 20%, 30%, and 38.5%, respectively (Table 2).

Other thoracic skeletal abnormalities found in the 22 propoiti included dorsal scoliosis in two, dorsal kyphoscoliosis in two, pectus excavatum in one, bilateral clinodactyly in one, and absence of 12th ribs in four.

Discussion

Various skeletal abnormalities including pectus excavatum, pectus carinatum, scoliosis, rib deformities, and straight back are common in patients with mitral valve prolapse.²⁸⁻²⁹ Our study²⁵ on 115 Chinese with mitral valve prolapse showed that 36 (31.3%) had a straight back. Udoshi *et al.*³⁰ found mitral valve prolapse in 40% of patients with thoracic skeletal abnormalities while Davies *et al.*¹¹ found mitral valve prolapse in 67% of patients with the straight back syndrome.

The presence of a straight back can be defined by several methods. Thoracic kyphosis can be measured as an angle subtended by a segment of the thoracic spine³¹ but there may be a measurement error of 5 degrees. Davies *et al.*¹¹ proposed measuring the distance from the middle of the anterior border of the

Table 1 Abnormal metacarpal index and straight back in 22 propoiti with idiopathic mitral valve prolapse

		No. of propoiti with ratio between anteroposterior diameter and transverse diameter of thorax		
		Above normal mean value	<2 SD below normal mean value	>2 SD below normal mean value
No. of propoiti with metacarpal index	Below the normal mean value	0	4	2
	Less than 2 SD above normal mean value	2	3	8
	More than 2 SD above normal mean value	2	1	0

SD, standard deviation.

Table 2 Relation between straight back syndrome and familial occurrence of mitral valve prolapse

	Propoiti with straight back	Propoiti with abnormal metacarpal index	Propoiti with neither straight back nor abnormal metacarpal index	Total
No. of families screened	10	3	9	22
No. of first degree relatives screened	35 (16 M, 19 F)	10 (6 M, 4 F)	26 (10 M, 16 F)	71 (32 M, 39 F)
No. of first degree relatives detected to have mitral valve prolapse	7 (20%) (2 M, 5 F)	3 (30%) (1 M, 2 F)	9 (38.5%) (4 M, 5 F)	19 (26.8%) (7 M, 12 F)

eighth thoracic vertebral body to a line connecting the fourth and 12th thoracic vertebrae and recommended a value less than 1.2 cm as indicative of a straight back. Both methods suffer from having a relatively large standard deviation so that the diagnostic sensitivity is reduced. Nevertheless, loss of thoracic kyphosis is associated with decrease in the anteroposterior diameter of the thorax. Hence, in the study by Twigg *et al.*³² as well as in our previous²⁵ and present studies the diagnosis of a straight back was made using the ratio between the anteroposterior diameter and the transverse diameter of the thorax.

Our present study supports the belief that mitral valve prolapse is transmitted in an autosomal dominant manner with incomplete penetrance (26.8% among first degree relatives), the expressivity among female subjects (30.8%) appearing more complete than among male subjects (21.9%).⁹ More important, our study shows that the familial occurrence of mitral valve prolapse does not depend on its association with the straight back syndrome. Though the straight back syndrome was described as an isolated entity,^{11,32} the various associated skeletal abnormalities in patients with mitral valve prolapse, including a straight back in 16.7%,²⁸ 23.4%,²⁹ and 31.3%,²⁵ and our present finding of familial occurrence of mitral valve prolapse being independent of the presence of a straight back suggest that this is not so. It is likely that both a straight back and mitral valve prolapse are features of a more generalised disorder, which is transmitted in an autosomal dominant manner with incomplete penetrance. These two features may occur together in the same individual or they may be dissociated. There is still no evidence that mitral valve prolapse is a *forme fruste* of the Marfan syndrome.

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