MARFAN OR MARFANOID: A CASE STUDY

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SUMMARY

A patient with marfanoid habitus was admitted to the General Hospital, Kuala Lumpur in October 1985 for surgical closure of an atrial septal defect. He was suspected to have Marfan Syndrome but there was no involvement of the aorta nor the eye. The clinical features were intermediate between that of the Marfan Syndrome and the Ehler's Danlos Syndrome. It is suggested that this could be a separate distinct entity within the heritable disorders of connective tissue known as the Marfanoid Hypermobility Syndrome.

INTRODUCTION

The objective of this study is to emphasize the difficulties encountered in making a diagnosis of the Marfan Syndrome and to review some of the diagnostic aids in individuals suspected of having this syndrome where only equivocal features are present.

CASE HISTORY

The patient, a 12-year-old Malay male, was found to have a heart lesion on routine school medical examination. He is the sixth in a family of seven children; the other children were in good health. There is no past history or family history of significance.

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He was 157 cm tall (5ft 2in) and weighed 29.2 kg. His upper-segment to lower-segment ratio was 0.85. The arm-span to height ratio was 1.06 (Fig. 1). He had arachnodactyly (Fig. 2): the Steinberg's Thumb Sign and the Walker-Murdoch Wrist Sign were both positive. He also had marked laxity of the joints of his wrists and fingers. His feet were long and narrow with normal arches. He had a marked pectus carinatum but there was no kyphoscoliosis. The skin was of normal consistency but it was hyperextensible. There was no striae. The subcutaneous fat was sparse and the muscles poorly developed. He also had a high arched palate.

The B.P. was 90/60 mm Hg. The pulses were normal. A mid-systolic click at the apex and an ejection systolic murmur at the left sternal edge were heard. There was also fixed split of the second heart sound.

An ophthalmic examination showed mild myopia. There was no dislocation of the lens or retinal detachment. The rest of the physical examination was normal.

The chest radiograph showed anterior chest wall deformity together with features of an intra-cardiac shunt. The ECG showed sinus rhythm, normal axis and urine examinations were normal.

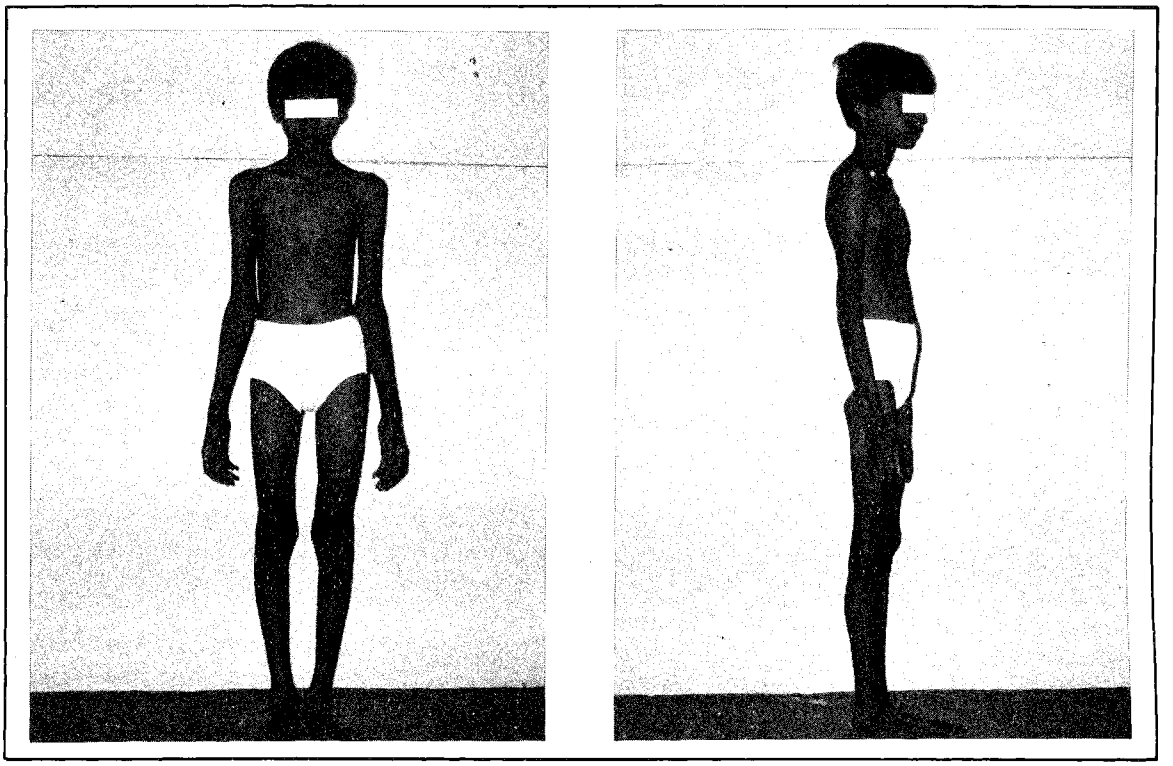

Fig. 1 General body configuration showing marfanoid habitus and pectus carinatum.

On echocardiography, the mitral valve prolapse was well demonstrated, the right ventricle was dilated, paradoxical septal motion was present and the aorta appeared normal. Cardiac catheterization on 27 May 1985 confirmed the presence of an ostium secundum atrial septal defect with a large shunt but with normal pressures. Patch closure of the atrial septal defect was done on 7 November 1985 in General Hospital, Kuala Lumpur. At the time of operation, the aorta appeared small, as expected for an intracardiac left to right shunt.

DISCUSSION

The Marfan Syndrome has an autosomal dominant mode of inheritance with incomplete penetrance resulting in variable clinical expression. When the classical triad of abnormalities of the eye, aorta and skeleton are present together with a positive family history, the diagnosis is obvious.[1] But when the features are equivocal, the diagnosis may be difficult, if not impossible, to make. The gracile habitus of many patients with atrial septal defect and the high incidence of chest deformity in these patients may further confuse the clinician.

This patient has many features of the Marfan Syndrome (e.g. pectus deformity, arachnodactyly, dolichostenomelia). However, there was no ocular involvement and the cardio-vascular manifestations were non-specific (mitral valve prolapse is frequently associated with atrial septal defects). The absence of involvement of the aorta was evident, both from the pre-operative studies as well as intra-operative assessment. The joint laxity seen here also far exceeds that found in the Marfan Syndrome and it approaches that of the Ehler's Danlos Syndrome Type 1. But the other typical features of the latter were absent. Joint laxity, per se, is of little diagnostic specificity.

The measurement of the upper-segment to lower-segment ratio is generally used in the evaluation of the skeletal changes of the Marfan Syndrome and is usually at least 2 SD below the mean

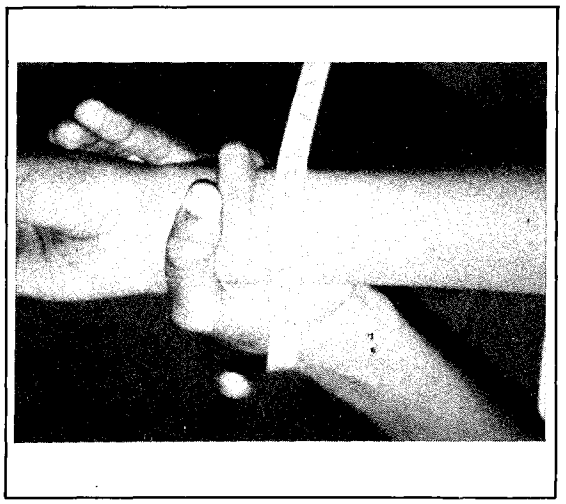

Fig. 2(a) The Steinberg's Thumb Sign. The patient makes a fist over the clenched thumb: The sign is positive when the thumb extends significantly beyond the ulnar margin of the hand (as shown above).

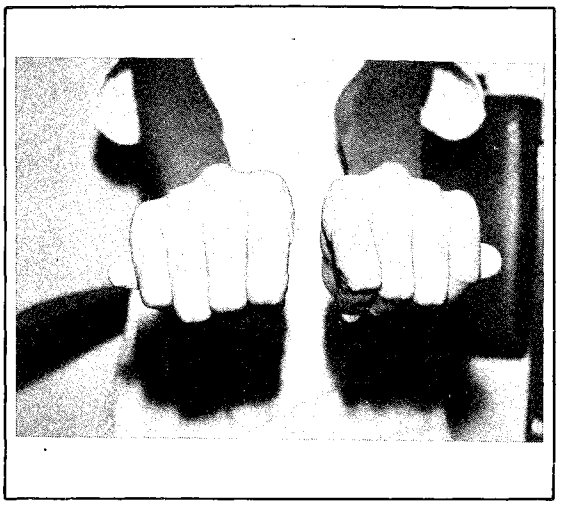

Fig. 2(b) The Walker-Murdoch Wrist Sign. The patient grasps the wrist and attempts to encircle it with the thumb and fifth finger as shown. The sign is positive when there is overlapping by a considerable extent, usually 1 to 2 cm.

for age, sex, race in patients with the Marfan Syndrome. This patient had a low ratio, but it fell within 2 SD of the mean. A similar result was obtained with the measurement of the arm-span to height ratio. The thumb sign and the wrist sign were both positive but this may reflect the longitudinal laxity of the hand rather than arachnodactyly. The metacarpal index was initially proposed as a more specific criterion for these changes and the upper limit of normal of 8.8 for men and 9.4 for women accepted. This patient had a metacarpal index of 8.2

Therefore, it was felt that more emphasis should be placed on the presence of 'hard' features (e.g. lens dislocation, aortic dilatation, severe kyphoscoliosis, petus deformities) than on soft features (e.g. myopia, mitral valve prolapse joint laxity, arachnodactyly) in the diagnosis of the Marfan Syndrome.[2]

It was also suggested that this patient may have the Marfanoid Hypermobility Syndrome,[3] a separate distinct entity within the heritable disorder of connective tissue that combines features of both the Marfan Syndrome and the Ehler's Danlos Syndrome.

ACKNOWLEDGEMENTS

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REFERENCES

[1] McKusick V A. *Heritable disorders of connective tissue* (4th ed.). St. Louis: C V Mosby, 1972.

[2] Pyeritz R E, McKusick V A. The Marfan Syndrome: diagnosis and management. *New Eng J Med* 1979; 14 : 772–777.

[3] Walker B A, Beighton P H, Murdoch J L. The Marfanoid Hypermobility Syndrome. *Ann Intern Med* 1969; 71 : 349–352.