Thoracic cage deformities in the early diagnosis of the Marfan syndrome and cardiovascular disease

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The Marfan syndrome is frequently complicated by cardiovascular abnormalities. Of these, aortic dissection and aortic valve regurgitation are the most life-threatening. The most noticeable abnormalities of the Marfan syndrome—the skeletal abnormalities—may be subtle and limited. Presented here are five reports of cases of the Marfan syndrome. All patients had potentially lethal cardiovascular complications. Either the syndrome had not been previously diagnosed or the patient had not been adequately monitored despite the presence of thoracic cage deformities present from youth. The purpose of this report is to heighten recognition of the association of thoracic cage deformities with the Marfan syndrome to permit early diagnosis of the associated cardiovascular complications. Surgical management of these complications can favorably alter the natural history of the Marfan syndrome.

The Marfan syndrome is a hereditary disorder of the connective tissue¹ first described in

1896.² The genetic pattern is one of autosomal dominant trait with incomplete penetrance, making the phenotypic presentation variable. The prevalence of this disorder is estimated to be 4 to 6 persons per 100,000,³ with no racial or ethnic predisposition. Diagnostic criteria for the Marfan syndrome are broad and each criterion has degrees of abnormality; therefore, if only the more severe cases are included in this estimate, the estimate may be too low.

The diagnosis of the Marfan syndrome is based on the presence of two or more characteristic familial, ocular, cardiovascular, or skeletal features. The skeletal abnormalities of dolichostenomelia (increased limb length), increased joint laxity, increased floor-to-pelvis measurement, and arachnodactyly are easily recognized signs of the Marfan syndrome. Pectus excavatum, pectus carinatum, and thoracic scoliosis may also be present. These manifestations may occur early in life and can be clues to early diagnosis. The case reports of five patients with the Marfan syndrome are presented here. All had thoracic cage abnormalities present from youth, and all eluded diagnosis or careful monitoring until cardiovascular complications arose. The cases are presented to heighten recognition of the association of thoracic abnormalities with the Marfan syndrome.

Report of cases

Case 1

A 36-year-old woman saw her primary physician and complained of shortness of breath and cough-

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Figure 1. *Left: Chest prominence (arrow). Right: Lateral chest x-ray film shows protrusion of left rib cage (arrow) (case 1).*

ing. She was known to have had a cardiac murmur for 11 years. When antibiotic therapy failed to relieve her symptoms, she was referred to the Cleveland Clinic. Examination revealed pistol-shot pulses, a vibrating systolic murmur, and a diastolic murmur of aortic valve regurgitation. Also noted was a left parasternal chest prominence (Fig 1). A chest x-ray film and an echocardiogram revealed a markedly dilated aortic root. The patient underwent successful repair of the ascending aorta with an aortic-valved conduit with reimplantation of the coronary arteries.

Case 2

A 39-year-old man with a diagnosis of the Marfan syndrome had had a cardiac murmur since birth. The patient had not received regular medical care. At age 29, acute aortic valve regurgitation developed and required aortic valve replacement. Ten years later, he presented at our clinic with symptoms of congestive heart failure. He was tall and thin with a markedly straightened dorsal spine signifying loss of the normal dorsal kyphosis (Fig 2). A murmur consistent with aortic valve regurgitation was noted. A chest x-ray film revealed marked thoracic lordosis, cardiomegaly, and a normal-appearing aorta. An echocardiogram showed aortic root dilation and aortic valve regurgitation. The

patient successfully underwent surgical placement of an aortic-valved conduit with reimplantation of the left coronary artery. Reimplantation of the right coronary artery was not surgically possible.

Case 3

A 26-year-old woman had a strong family history of the Marfan syndrome. Nevertheless, the condition was not so diagnosed until she had an acute aortic dissection. She was found to have a systolic ejection murmur, multiple systolic clicks, joint laxity, arachnodactyly, and a prominent pectus carinatum (Fig 3). Echocardiograms revealed a dilated aortic root and an aortic dissection distal to the left subclavian artery. The patient underwent successful surgical repair of the aneurysm and dissection.

Case 4

A 32-year-old man had the Marfan syndrome diagnosed at age 12. The diagnosis was based on skeletal and ocular abnormalities. There was no family history of the disorder. The patient was clinically well until age 32 when he was seen at the Cleveland Clinic with aortic valve endocarditis and aortic valve regurgitation. An echocardiogram revealed an aortic root measuring 5.8 cm in diameter. The patient had a murmur characteristic of

Figure 2. *Left: Loss of normal dorsal kyphosis. Right: Lateral chest x-ray film shows marked dorsal lordosis and relative lumbar kyphosis (case 2).*

Figure 3. *Left: Prominent pectus carinatum. Right: Posteroanterior chest x-ray film shows widening of the ascending and descending aorta consistent with ascending aortic aneurysm and descending aortic dissection (case 3).*

aortic valve regurgitation, pectus excavatum (Fig 4), ectopia lentis, and a high arched palate. There were no arachnodactyly or dolichostenomelia. He underwent successful surgical repair with aortic-valved conduit with reimplantation of the coronary arteries.

Case 5

A 37-year-old woman with intermittent, sharp chest pain was seen at the Cleveland Clinic. She had been told as a child that she had a cardiac murmur but there had been no further investigation.

The patient had had three first-trimester spontaneous abortions and chondromalacia patellae. She denied chest trauma but was concerned about the pain because her father had had surgery for a thoracic aortic aneurysm. The findings of the cardiac examination were normal; however, skeletal examination revealed dolichostenomelia, arachnodactyly, torus palatinus, and pectus excavatum. She had seen numerous physicians but had never had her condition diagnosed as the Marfan syndrome. Despite a chest x-ray film that revealed a normal-appearing heart and aorta, an echocardiogram

Figure 4. Lateral chest x-ray film shows sternal depression (arrows) consistent with pectus excavatum (case 4).

Figure 5. Two-dimensional echocardiogram. Anterior and posterior walls of the aortic root (arrows) dilated to 4.1 cm at level of sinuses of Valsalva (case 5).

Figure 6. Transesophageal echocardiogram. Note true and false lumens of this aortic dissection. Blood flows from true to false lumen.

showed a dilated ascending aorta (Fig 5) at the level of the sinuses of Valsalva. The patient now undergoes a yearly echocardiogram to assess the aortic root size.

Discussion

Thoracic cage abnormalities can be an early clue to the diagnosis of the Marfan syndrome. Classic skeletal signs include dolichostenomelia, arachnodactyly, and increased floor-to-pelvis measurement. Pectus carinatum, including unilateral pectus carinatum, pectus excavatum, loss of the normal dorsal kyphotic curve, and scoliosis, can also indicate this syndrome. A high index of suspicion of the Marfan syndrome in patients with skeletal abnormalities can lead to early detection of its cardiovascular complications. These complications are responsible for more than 95% of the discernible causes of death in patients with the

Marfan syndrome and are the primary reason that these patients' life expectancy is halved.⁴

Cardiovascular abnormalities in patients with the Marfan syndrome are many,^{3,5-9} but the most life-threatening—and the cause of death in 29% to 43% of these patients^{10,11}—is dissection of the ascending aorta. Dilation of the aorta or annulo-aortic ectasia¹² progresses gradually over years, but dissection with aortic valve regurgitation seldom occurs with an

aorta smaller than 6 cm in diameter. Medical and surgical intervention can favorably alter the natural history of this syndrome.¹³

Physical examination is often not sufficient to detect cardiovascular abnormalities. A chest x-ray film is helpful in assessing heart size and aortic diameter, but an echocardiogram is more precise.⁶ The addition of color flow Doppler and transesophageal echocardiography (Fig 6) to conventional echocardiography further improves the specificity for the diagnosis of aortic dissection.¹⁴

Good history taking and attention to subtle abnormalities of the chest wall can aid in the prospective diagnosis of the Marfan syndrome and its cardiovascular complications, providing the opportunity for elective surgical repair. Such repair can be performed successfully in nine out of ten cases,¹⁵ clearly a favorable alteration in the natural history of the syndrome.^{4,13}

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