Marfan's Syndrome Presenting as a Type 3 Aortic Dissection*

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Case Report

A 29-year-old previously healthy white man without an established diagnosis of Marfan's syndrome presented to the emergency room after the acute onset of intense left flank pain followed by syncope. The patient was found to be hypotensive, diaphoretic, and mildly lethargic. The patient's past medical history was also reviewed after surgery. Physical examination revealed a tall (6'5"), thin man without obvious skeletal or pectus deformities. No ocular abnormalities were noted. A high arched palate was noted. Pedal pulses were diminished bilaterally. Cardiac examination disclosed an early peaking systolic ejection murmur, but no diastolic murmurs, gallops, or clicks were appreciated.

An intravenous pyelogram was performed initially and demonstrated marked lateral displacement of the left kidney by a large retroperitoneal mass. Because of protracted hypotension, a fall in hemoglobin from 11.2 to 10.0 and a low central venous pressure, an emergency aortogram was performed with the following findings: (1) large dissecting thoracoabdominal aortic hematoma extending from T5-T9 to the iliac arteries. It measured 12.2 cm in maximum internal diameter. Retroperitoneal extravasation was also noted. (2) Anuloaortic ectasia, extending 7.5 cm cephalad from the aortic valve, pear shaped with maximum aortic root diameter of 9 cm (Fig. 1). No aortic regurgitation was noted angiographically.

The patient was taken to surgery, where an extensive chronic descending aortic dissection was found. The false channel was obliterated, and a 30 mm woven Dacron graft was inserted. His postoperative course was complicated by mild hypoxia, bibasilar atelectasis, and renal insufficiency, but he improved steadily and was discharged two weeks after surgery.

Echocardiogram performed after surgery disclosed a dilated aortic root but no diastolic mitral valve fluttering to suggest aortic regurgitation. This was consistent with the postoperative clinical examination which demonstrated no murmur of aortic insufficiency. Left ventricular size was normal. Radiographs of the hand revealed a markedly abnormal metacarpal length to width ratio.

Significant past problems included operations for bilateral inguinal hernias as well as a spontaneous pneumothorax. He had been told about a mitral murmur earlier in life, but denied other medical problems. Recently he had been very active in long distance running. There was no family history of Marfan's syndrome.

Questions

(1) What is the natural history of patients with Marfan's syndrome who present with extensive anuloaortic ectasia but no aortic regurgitation?

(2) Is the risk of proximal dissection high enough to warrant prophylactic repair before clinically significant aortic insufficiency occurs?

(3) If surgery is not warranted now, what indication should be used to determine appropriate timing of surgery (ie, aortic regurgitation by clinical examination, symptomatic aortic regurgitation, signs of congestive heart failure)?

(4) How common is a type 3 aortic dissection in Marfan's syndrome? How does this affect overall survival?

Comments

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Marfan's syndrome is a genetic disorder of the connective tissue which has four clinical manifestations. These include ocular, skeletal and cardiovascular man-

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Figure I. Anuloaortic ectasia, with localized expansion just distal to the aortic outflow tract (arrows).
manifestations, as well as a family history of the disorder. The transmission rate of the syndrome to children of a Marfan's patient is about 50 percent.

Half of the patients with Marfan's syndrome die at an average age of 32 years; 95 percent of these deaths are caused by cardiovascular complications. These cardiovascular complications consist of aneurysms involving different parts of the aorta and dissection of the aorta, which occur more commonly in the Marfan's patient than the normal population. Dissection and laceration may lead to rupture into the pericardium with a fatal outcome; however, death from heart failure is less common.

Left-sided heart valvular lesions may present in the form of aortic and/or mitral insufficiency with bacterial endocarditis, leading to heart failure. The treatment of choice is surgery for patients who develop any one of these cardiovascular complications. These operations can be done electively, with a survival rate approaching 100 percent. Recently it has been shown that life can be prolonged by preventing complications of cardiovascular manifestations. The actuarial survival curve constructed from our series of cases indicates the probable survival of 62 percent at 15 years. Three out of 11 late deaths were caused by aortic rupture occurring while the patients were under observation and had no visible changes in their clinical status. Theoretically, seven out of the 11 deaths (63 percent) could have been prevented by a more aggressive application of presently available surgical treatment.

Aortic insufficiency usually develops in patients with an anuloectatic aorta, or patients who have undergone aortic valve replacement by graft and resuspension of aortic valve for acute dissection. All patients who have had a fusiform aneurysm of the ascending aorta treated by separate graft replacement, excision, and end-to-end anastomosis or separate valve replacement, or separate graft replacement and valvuloplasty, develop large sinus aneurysms three to six years after operation and recurrence of aortic insufficiency. In contrast, aortic insufficiency does not recur in patients treated with a composite valve graft replacement. These successful results can be explained by the fact that the aortic anulus and the aortic valve are diffusely involved in these cases and total replacement is essential to relieve the condition and prevent recurrence.

Traditionally, the timing of the treatment of the aneurysm of the ascending aorta in patients with Marfan's syndrome was based upon the presence of hemodynamic changes produced by valvular insufficiency, sudden mural changes such as laceration or dissection of the ascending aorta occurring in the aneurysm, or enlargement of the ascending aorta to 5 cm in diameter. It can now be shown, however, that because of the unpredictability of sudden death, the prophylactic operation should be undertaken as soon as the lesion becomes established. As previously discussed, three of the 11 late deaths in our series of patients occurred suddenly from ascending aortic aneurysm rupture due to either mural laceration or superimposed dissection while waiting for these hemodynamic and mural changes to occur.

The principle of treatment of the patients with aortic dissection or more distant degenerative fusiform aneurysm, such as descending thoracic aorta, thoracoabdominal or infrarenal aortic aneurysm, is identical to that previously described for similar lesions in patients without Marfan's syndrome. Patients with type 3 aortic dissection should be followed because one third of those patients develop aneurysmal dilatation of false lumen of the dissection, and one third of those developing aneurysmal dilatation die of aortic aneurysm rupture.

Fifty percent of the patients in the series required one or more operations within one to ten years of the first operation because of development of a new lesion or because prior treatment was based upon old techniques. The preferred surgical treatment for patients who present combined lesions of the ascending and descending or thoracoabdominal aorta is a staged operation. The interval between operations is six weeks. Patients who have a mitral valve prolapse might need mitral valve reconstruction or mitral valve replacement.

Our primary conclusion from our series is that the aneurysm of the ascending aorta should be treated when the diagnosis is made because, with this form of disease, the patient may die suddenly either from spontaneous rupture or dissection. In these cases, composite valve graft should be employed to prevent recurrent aneurysm. We also conclude that the aortic valve should be replaced in all patients who have acute dissection of the previously normal ascending aorta along with aortic insufficiency due to the recurrence, which occurred in all patients in our series who had valvular resuspension procedures. Finally, close and frequent follow-up is important in these cases because of the frequency with which they developed new cardiovascular problems. Management of such complications as mitral valve disease or other aneurysms is done according to the principles established for the non-Marfan patient.

**Comments**

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This young man, fortunate as he is to have survived to this point, is at considerable risk for further dissection and sudden death. He could benefit from the recent, gratifying improvement in the ability to diagnose and manage aortic complications of the Marfan

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syndrome. My comments on this case and recommendations for further management reflect experience with many hundreds of persons with Marfan syndrome, including over 100 patients who have required aortic repair.

In the first place, there are sufficient criteria in the skeletal, pulmonary, and cardiovascular systems to establish the diagnosis of Marfan syndrome, even in the absence of a positive family history. He likely represents a sporadic case due to germinal mutation in a patient.

Second, descending aortic dissection in the Marfan syndrome is far less common than that of the ascending aorta; the thoracoabdominal aorta is most often involved by a DeBakey type I dissection. Nonetheless, aneurysmal enlargement, with or without dissection, involving primarily the descending thoracic or the abdominal aorta occurs in Marfan syndrome patients more frequently, and at a younger age, than in the general population. An interesting and unusual feature is the extent of the aortic dilatation (75 mm) in the absence of aortic regurgitation. Most patients will have developed some degree of valvular incompetence by the time the root dilates to 55 mm.

Third, we have learned that management of ascending aortic aneurysms in Marfan syndrome and in idiopathic anuloaortic ectasia should be based, in the first instance, on the aortic root diameter. The presence of hemodynamically significant aortic regurgitation obviously will argue for surgical correction; the most important point, however, is that dilatation of 60 mm and greater should be a strong indication for surgical correction, even in the absence of aortic regurgitation.

Fourth, the surgical procedure of choice involves repair with a composite prosthetic valve—tubular graft, with end-to-side anastomosis of the coronary arteries. The aneurysmal aorta is then wrapped around the graft to provide hemostasis.

In conclusion, this patient should have elective repair of the ascending aorta as soon as possible because of the risk of dissection, with the concomitant likelihood of involvement of the coronary arteries or cardiac tamponade. Until surgery, he should discontinue vigorous exertion and isometric exercise. Negative inotropic doses of propranolol or atenolol should be instituted with the intent of continuing therapy following surgery. Postoperatively, he should have periodic evaluations of the entire aorta; computed tomography provides a convenient, noninvasive mechanism for this.

REFERENCES

Editorial Note: The patient was noted several days after surgery to have a blowing diastolic murmur consistent with aortic regurgitation. Nuclear angiography revealed an ejection fraction of 36 percent and dilated left ventricle. He was readmitted to the hospital four months later for elective repair of the anuloaortic ectasia. A composite graft was inserted and the patient had an uneventful postoperative course. He was doing well six months after surgery.