Marfan Syndrome Clinic

Mayo Clinic
Division of Cardiovascular Diseases
Division of Cardiovascular Surgery
Marfan Syndrome

Marfan syndrome is a heritable disorder of connective tissue. One of the most common single-gene mutations, it affects one person in 10,000. Marfan syndrome is caused by a mutation in the gene encoding fibrillin-1 on chromosome 15, and is inherited in an autosomal dominant pattern in 75 percent of cases; the rest arise from a spontaneous mutation. Fibrillin-1 is a structural protein needed for orderly elastin deposition in tissues. Elastin-rich tissues are not as strong in Marfan patients, which leads to the clinical manifestations of the disorder.

Features of Marfan Syndrome

Marfan syndrome typically affects the skeletal, ocular, and cardiovascular systems and may also affect additional organ systems. Diagnosis is based on the family and medical history, as well as recognition of typical manifestations in important target organs.

Skeletal features — including tall stature, thoracic cage deformities, and scoliosis — often raise the initial suspicion of Marfan syndrome. In contrast to those who are simply tall, Marfan patients usually have disproportionately long extremities, with long fingers and long, narrow feet. Thoracic cage deformities result from rib overgrowth, which can either push the sternum in (pectus excavatum) or out (pectus carinatum). Joint laxity and ligamentous laxity contribute to the predisposition to scoliosis and to flat feet (pes planus). The palate tends to be high and narrow, causing the teeth to be crowded.

The characteristic ocular findings in Marfan syndrome include severe myopia and dislocation of the ocular lens (ectopia lentis). Ectopia lentis occurs in approximately 65 percent of patients, often within the first five years of life. Adults with Marfan syndrome also have an increased risk of retinal detachment, glaucoma and cataracts.

Cardiovascular involvement is the most common cause of morbidity and mortality. Mitral valve prolapse is common in the general population, but in the Marfan syndrome it usually is severe and frequently leads to disruption of the valve apparatus and clinically important mitral valve regurgitation. The aortic valve and aortic root are commonly affected. Progressive root enlargement is typical and eventually results in aortic valve regurgitation and the propensity to aortic dissection if untreated. Average survival in the Marfan syndrome in the era before modern cardiovascular surgery was only 32 years, with most patients dying of either aortic dissection or heart failure caused by aortic or mitral valve regurgitation.

Marfan syndrome also may affect the lungs with spontaneous pneumothorax, apical blebs detected on chest radiography, or restrictive lung disease (from scoliosis). There is a propensity toward obstructive sleep apnea. Skin problems may include stretch marks unrelated to weight gain or pregnancy and recurrent or incisional hernias.

Diagnosis

The diagnosis of Marfan syndrome is made after a comprehensive clinical evaluation that includes a family and medical history, a complete physical examination and a genetic evaluation. Additional testing often required to secure a diagnosis includes an echocardiogram, an eye examination by an ophthalmologist, and a detailed skeletal examination. Because the fibrillin-1 gene is large and over 200 different mutations have been described so far, there is no readily available laboratory test for making the diagnosis.
Surgery

Advances in cardiovascular medicine and surgery have resulted in marked improvement in survival for Marfan patients, whose life expectancy is now close to normal.

- Prophylactic aortic root replacement is usually recommended when the dimension reaches 50 or 55 mm. Valve-sparing procedures for the aortic valve also have been developed in recent years.
- Mitral valve prolapse leading to severe mitral valve regurgitation is the most common indication for cardiac surgery in children with Marfan syndrome. In most cases, intraoperative repair of the native mitral valve is preferred over replacement with a prosthetic valve.

Treatment of pectus deformities has been revolutionized since the late 1990s. A minimally invasive surgical correction for pectus excavatum creates a more normal chest wall and enables patients to recover much more quickly, while a new bracing regimen for pectus carinatum during the teenage years can help those patients avoid surgery entirely.

Case Management

Marfan syndrome management consists of regular follow-up with specialists who are familiar with the complications of the disorder. Orthopedic care is often needed for scoliosis and problems with the hips and feet, particularly early in life. Annual eye examinations focus on the status of the lens and retina. Cardiac follow-up includes annual echocardiography and other imaging studies of the heart and aorta. Beta blockade has been shown to slow the rate of aortic root enlargement.

Marfan Syndrome In Brief

- Marfan syndrome occurs as a spontaneous mutation in 25 percent of cases, so a family history may not be present.
- Although skeletal and ocular involvement cause characteristic external features, the cardiovascular involvement is the major cause of morbidity and mortality.
- Beta-blocker therapy slows the rate of aortic enlargement, decreases the risk of aortic dissection and may delay the need for aortic operation.
- Elective ascending aortic surgery has markedly improved the life expectancy of Marfan patients.
- The Marfan Syndrome Clinic at Mayo Clinic in Rochester, Minn., offers multidisciplinary longitudinal care for patients and their families with suspected or known Marfan syndrome or other heritable disorders of connective tissue.

Marfan Syndrome Clinic

Mayo Clinic has treated more than 600 patients with Marfan Syndrome since 1986. In August 2002, Mayo Clinic established a new multidisciplinary clinic to serve patients and their families with Marfan syndrome and other heritable connective tissue disorders. The Marfan Syndrome Clinic offers an integrated medical, genetic and surgical team of specialized Mayo Clinic physicians to provide diagnosis and state-of-the-art treatment for patients with suspected or confirmed Marfan syndrome. The Marfan Syndrome Clinic provides patients with education regarding exercise, endocarditis prophylaxis, pregnancy, genetic implications and the lifelong management of their condition. Genetic testing is available and may be recommended under certain circumstances, but is not routinely performed.

To request a phone consultation or a referral appointment, please call: 507-284-3328

Marfan Syndrome Clinic Web site: http://mayoclinic.org/marfan-rst