South Florida Hospital News and Healthcare Report
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# Marfan syndrome, an underdiagnosed life-threatening disease

**What is Marfan syndrome?**

Marfan syndrome was first described in the 1890s by the French professor of pediatrics Antoine Bernard-Jean Marfan when he presented the case of a five-year-old girl to the Société Médicale des Hôpitaux de Paris. He reported her disproportionately long limbs, long slender fingers and other skeletal abnormalities.

We now know Marfan syndrome is an autosomal dominant inherited

connective tissue disorder with variable penetrance. Only one copy of the defective gene has to be inherited in an autosomal chromosome to acquire the disease. The clinical manifestations vary considerably from person to person making it difficult to diagnose.

The syndrome results in connective-tissue weakening due to mutations in the gene that tells the body how to produce fibrillin-1, Which helps give connective tissue its elasticity and strength. This gene mutation is also responsible for excessive growth factor beta signaling that could explain the long-bone overgrowth and long fingers seen in Marfan's syndrome.

The tissues affected the most, have high concentrations of this protein. The incidence is approximately one out of 5,000 births, men and women are equally affected. While 75 percent of cases are inherited, 25 percent of cases result from spontaneous mutations; this means that the affected individuals are the first ones in the family to have the disease. A significant number of individuals with Marfan syndrome are diagnosed later in life, during adulthood.

**Signs of Marfan syndrome:**

Features of affected individuals are variable, some individuals have connective tissue changes that can be seen at birth and some are only found later in life during adulthood.
Marfan syndrome mainly involves any organ/system with connective-tissue, however, the cardiovascular, skeletal and ocular systems are the most commonly affected.
The cardiovascular features are the more clinically significant and these include weakening of the wall of the aorta. This results on considerable dilation of this artery and its root also called an aortic aneurysm.

A significantly dilated aorta or rapidly dilating aorta is a high risk for aortic dissection and rupture which is a life-threatening condition that needs immediate surgery. Other cardiovascular features commonly seen with the syndrome are mitral valve prolapse that could result in heart failure

symptoms due to severe regurgitation and severe aortic valve insufficiency due to aortic root dilation. Affected individuals can also develop spontaneous pneumothorax which is the collapse of a lung, restrictive lung disease and emphysema changes. The skeletal and ocular features include ligament laxity, long bone overgrowth giving tall stature, slender long fingers and dislocated eye lenses due to the weakening of the connective tissue holding this structure in place.

**The survival of untreated patients with Marfan syndrome is reduced by about one third with death occurring at an average age of 30-40 years.Diagnosis:**
A detailed family history, medical history and physical examination should be conducted. A complete multidisciplinary evaluation includes cardiology, ophthalmology, and genetics examinations to confirm the diagnosis. If Marfan syndrome is diagnosed, routine monitoring for aortic growth with echocardiography or computed tomography should be completed. For individuals with family history of Marfan syndrome genetic testing can confirm or rule out the disease. Genetic testing is also helpful in the diagnosis of other connective-tissue diseases that could result in features similar to Marfan's syndrome. Some of this related disorders are Loeys-Dietz syndrome, Ehlers-Danlos syndrome, bicuspid aortic valve, and Familial Thoracic Aortic Aneurysm and Dissection.

**Treatment:**
Early diagnosis, improved medical management and lifestyle modifications in conjunction with cardiac specialists have improved life expectancy to near normal. Medical management with beta-blockers and/or angiotensin receptor blockers is recommended to delay or prevent aortic aneurysm and dissection.
Surgery is reserved for cardiovascular complications of Marfan syndrome including prophylactic aortic root replacement for ascending aortic aneurysm with or without significant aortic valve insufficiency, emergent ascending aorta and root replacement for aortic dissection or rupture, aortic arch replacement for arch aneurysm, descending thoracic aortic replacement for acute or chronic dissection, and mitral valve repair or replacement for symptomatic severe regurgitation

secondary to mitral valve prolapse.

**Surveillance:**
Once diagnosed with Marfan syndrome the patient has to be under strict surveillance and follow-up. Routine echocardiogram and aortic imaging should be done every year by a cardiac specialist. If aortic surgery has been performed, then imaging surveillance should take place at shorter intervals.
Early diagnosis, medical and surgical treatment, and lifestyle modifications are very important to prevent life-threatening complications of Marfan syndrome. Improvement in recognition and management of this condition results in nearly normal life expectancy. Accurate genetic diagnosis is essential not only for individuals with Marfan syndrome but also for those with related connective tissue disorders. Knowing the signs and features of Marfan syndrome can save lives.

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