Marfan Syndrome

Marfan syndrome is an autosomal dominant genetic disorder of connective tissue which occurs from a Fibrillin-1 gene mutation. This effects various organ systems, such as the eye, cardiovascular and musculoskeletal systems. Patients are tall with hypermobile joints, and can have mitral valve prolapse, aortic dissection or aneurysms, as well as subluxation of the lense(s) in the eye.



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Mechanism

Fibrillin-1 Mutation
Fiber-linen (1) Wand Mutant
This syndrome is caused by a mutation in a gene, which encodes fibrillin-1, which is an extracellular matrix component. This leads to defective elastin fibers, which are present throughout the body, but also in the aorta and eye.

Symptoms

Autosomal Dominant
Domino
Marfan syndrome is inherited in an autosomal dominant fashion, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Tall
Tall-guy
Patients with Marfan syndrome are tall, and have above average height.

Arachnodactyly
Spider-fingers
These patients have skeletal abnormalities such as long fingers and toes, known as arachnodactyly. They can also have long arms with thin, weak wrists.

Pectus Excavatum
Pecs Excavator
Sternal defects can occur, called pectus excavatum, where the chest is abnormally indented.

Hypermobile Joints
Flexible Hiker-mobile
As this disorder effects connective tissue, patients can exhibit abnormal joint flexibility. This can also lead to injury more frequently if Marfan syndrome patients are involved in physical activity.

Aortic Aneurysm And Dissection
A-orca with Aneurysm and Dissection
The most severe consequence of this disorder involve the connective tissue of blood vessels. Patients can have aortic aneurysms and dissections due to weakened vessel connective tissue. Aortic dissections should be treated as a surgical emergency because they can be rapidly fatal.

Mitral Valve Prolapse (MVP)
Mitt-troll MVP
Regurgitation can happen and heart murmurs are often present in these patients. This is because degeneration of the heart valves can occur, leading to mitral valve prolapse (MVP).

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Subluxation of Lens (Superior)

Camera-lens Sub

A common ocular feature of this syndrome is subluxation of the lense(s) in a superotemporal direction. Patients can also have vision problems with this disorder, and retinal detachment can occur because of weakened connective tissue. The most common direction of dislocation on exam is superotemporal.

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