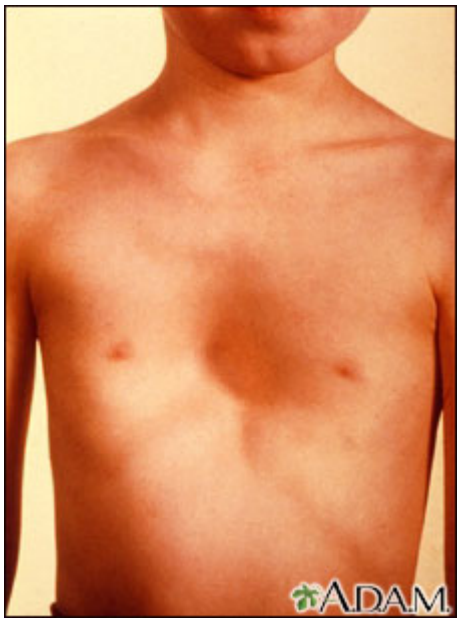
Marfan syndrome

Definition



Marfan syndrome is a disorder of connective tissue. This is the tissue that strengthens the body's structures.

Disorders of connective tissue affect the skeletal system, cardiovascular system, eyes, and skin.

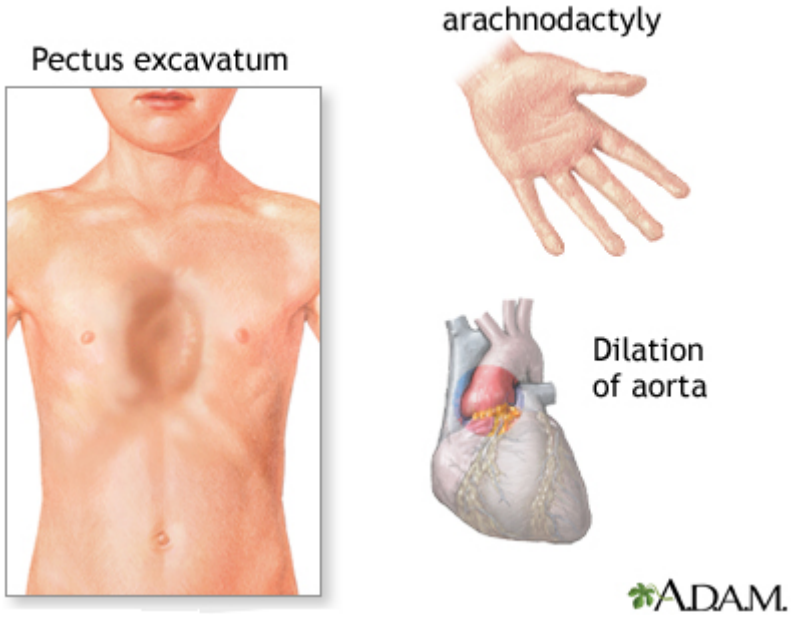
Alternative Names

Aortic aneurysm - Marfan

Causes

Marfan syndrome is caused by defects in a gene called fibrillin-1. Fibrillin-1 plays an important role as the building block for connective tissue in the body.

The gene defect also causes the long bones of the body to grow too much. People with this syndrome have tall height and long arms and legs. How this overgrowth happens is not well understood.



Other areas of the body that are affected include:

- Lung tissue (there may be a pneumothorax, in which air can escape from the lung into the chest cavity and collapse the lung)
- The aorta, the main blood vessel that takes blood from the heart to the body may stretch or become weak (called aortic dilation or aortic aneurysm)
- The eyes, causing cataracts and other problems (such as a dislocation of the lenses)
- The skin
- Tissue covering the spinal cord

In most cases, Marfan syndrome is passed down through families (inherited). However, up to 30% of people have no family history, which is called "sporadic." In sporadic cases, the syndrome is believed to be caused by a new gene change.

Symptoms

People with Marfan syndrome are very often tall with long, thin arms and legs and spider-like fingers (called arachnodactyly). The length of the arms is greater than height when arms are stretched out.

Other symptoms include:

- A chest that sinks in or sticks out, called funnel chest (pectus excavatum) or pigeon breast (pectus carinatum)
- Flat feet
- Highly arched palate and crowded teeth
- Hypotonia
- Joints that are too flexible (but the elbows may be less flexible)
- Learning disability
- Movement of the lens of the eye from its normal position (dislocation)
- Nearsightedness
- Small lower jaw (micrognathia)
- Spine that curves to one side (scoliosis)
- Thin, narrow face

Many people with Marfan syndrome suffer from chronic muscle and joint pain.

Exams and Tests

The health care provider will perform a physical exam. The joints may move around more than normal. There may also be signs of:

- Aneurysm
- Collapsed lung
- Heart valve problems

An eye exam may show:

- Defects of the lens or cornea
- Retinal detachment
- Vision problems

The following tests may be performed:

- Echocardiogram
- Fibrillin-1 mutation testing (in some people)

An echocardiogram or another test should be done every year to look at the base of the aorta and possibly the heart valves.

Treatment

Vision problems should be treated when possible.

Monitor for scoliosis, especially during the teenage years.

Medicine to slow the heart rate and lower blood pressure may help prevent stress on the aorta. To avoid injuring the aorta, people with the condition should avoid participating in contact sports. Some people may need surgery to replace the aortic root and valve.

People with Marfan syndrome who have heart valve conditions may need to take antibiotics before dental procedures to prevent endocarditis (infection of the valves). Pregnant women with Marfan syndrome must be monitored very closely because of the increased stress on the heart and aorta.

Support Groups

National Marfan Foundation -- www.marfan.org

Outlook (Prognosis)

Heart-related complications may shorten the lifespan of people with this disease. However, many people live into their 60s and beyond. Good care and surgery may further extend lifespan.

Possible Complications

Complications may include:

- Aortic regurgitation
- Aortic rupture
- Bacterial endocarditis
- Dissecting aortic aneurysm
- Enlargement of the base of the aorta
- Heart failure
- Mitral valve prolapse
- Scoliosis
- Vision problems

When to Contact a Medical Professional

Couples who have this condition and are planning to have children may want to talk to a genetic counselor before starting a family.

Prevention

Spontaneous new gene mutations leading to Marfan (less than one third of cases) cannot be prevented. If you have Marfan syndrome, see your provider at least once every year.

References

Doyle A, Doyle JJ, Dietz HC. Marfan syndrome. In: Kliegman RM, Stanton BF, St. Geme JW, Schor NF, eds. *Nelson Textbook of Pediatrics*. 20th ed. Philadelphia, PA: Elsevier; 2016:chap 702.

Madan-Khetarpal S, Arnold G. Genetic disorders and dysmorphic conditions. In: Zitelli, BJ, McIntire SC, Norwalk AJ, eds. *Zitelli and Davis' Atlas of Pediatric Diagnosis*. 7th ed. Philadelphia, PA: Elsevier; 2018:chap 1.

Pyeritz RE. Inherited diseases of connective tissue. In: Goldman L, Schafer AI, eds. *Goldman-Cecil Medicine*. 24th ed. Philadelphia, PA: Elsevier Saunders; 2016:chap 260.

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