Marfan Syndrome

What is Marfan syndrome?

Marfan syndrome is a genetic condition that affects the body's connective tissue. Connective tissue holds all parts of the body together and helps control how the body grows.

Connective tissue is all over the body. Because of this, Marfan syndrome can affect many different parts of the body.

Marfan syndrome can affect:

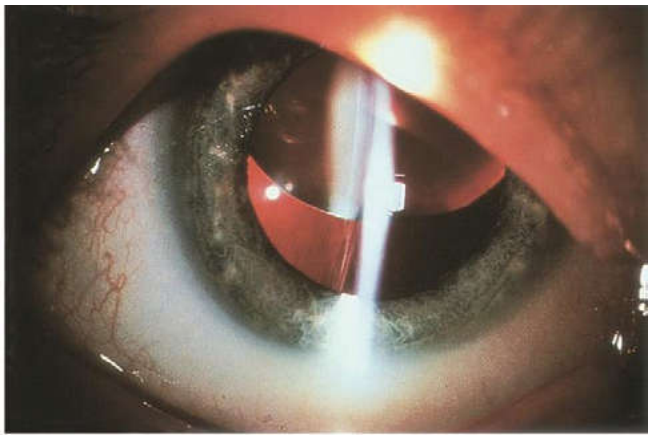
- the heart
- blood vessels
- bones
- joints, and
- eyes

People are born with this condition, but the symptoms of Marfan syndrome may not appear for a while.

What are the symptoms of Marfan syndrome?

Symptoms of Marfan syndrome can involve many parts of the body.

People with Marfan syndrome usually first notice its effect on their skeletal development. Someone with Marfan syndrome is usually very tall, thin, and loose-jointed. When they stretch out their arms from their sides, their arm length from one side to the other is greater than their height.



Ectopia lentis—when the eye's natural lens becomes dislocated—is often the first sign of Marfan syndrome.

Other common Marfan syndrome symptoms include:

- long, slender fingers and toes
- curvature of the spine
- protruding or indented breastbone
- bands of thin, wrinkled skin around the hips, shoulders or lower back
- cardiovascular problems (heart murmurs, enlarged or bulging aorta)

Eye symptoms of Marfan syndrome

Eye problems are common in people with Marfan syndrome. Most people with Marfan syndrome have myopia (nearsightedness) and astigmatism. More than half of people with Marfan syndrome have a condition called ectopia lentis. With ectopia lentis the eye's lens becomes dislocated. Because the lens helps focus light rays on the retina, visual

acuity worsens. Ectopia lentis is a key symptom of Marfan syndrome and is often the first sign of the disorder.

Other Marfan syndrome symptoms involving the eye include:

- thinning of the cornea
- flattened curvature of the cornea
- early onset of cataracts (clouding of the eye's normally clear lens)
- Glaucoma (high pressure inside the eye that can lead to vision loss)
- Strabismus (when the eyes are not aligned properly and point in different directions)
- retinal detachment (when the light sensitive tissue peels off from the back of the eye)

Who is at risk for marfan syndrome?

The cause of Marfan syndrome is a mutation on a gene that tells the body how to make fibrillin. Fibrillin is a critical part of connective tissue.

Marfan syndrome is usually an inherited genetic disorder. Three out of four people with Marfan syndrome inherited it from a parent. That means you are at greatest risk if you have a parent with Marfan syndrome. A parent with Marfan syndrome has a 50-50 chance of passing the defective gene along to his or her children.

About one-quarter (1 out of 4) of Marfan syndrome cases are not inherited. This means these people have a spontaneous new gene defect. The disorder affects both sexes equally and can occur in any ethnic group.

How is Marfan syndrome diagnosed?

A dislocated lens in the eye is often the first sign of Marfan syndrome. Because of this, an ophthalmologist can play an important role in diagnosing the disorder.

A slit-lamp eye exam will determine if you have dislocated lenses. Using this special microscope, your ophthalmologist can examine your eye in detail. This makes it easier to spot abnormalities.

Your ophthalmologist will refer you to other doctors if he or she suspects Marfan syndrome. They will conduct further tests to determine if you have the syndrome. This includes heart tests. Doctors diagnose it using a list of features found in Marfan syndrome.

Marfan syndrome treatment

Marfan syndrome can cause several different eye disorders. You should see an ophthalmologist regularly. Your ophthalmologist can treat many of the eye problems associated with Marfan syndrome.

Dislocated lenses. He or she can correct vision problems caused by dislocated lenses. Special glasses and eyedrops that make your pupil larger can improve your vision. Sometimes dislocated lenses may need to be removed if they are interfering with vision.

Cataracts. Having Marfan syndrome also puts you at higher risk for several eye diseases. You may develop cataracts at a younger age. If cataracts do develop, cataract surgery can improve your vision.

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Glaucoma. You are also at higher risk for developing glaucoma. Several glaucoma treatment options are available.

Lazy eye. Children diagnosed with Marfan syndrome can also develop amblyopia (lazy eye). It is important that treatment starts early for sight to be recovered in the weaker eye.

Retinal detachment. Marfan syndrome puts you at higher risk for having a retinal detachment. Be aware of the symptoms of retinal detachment. Seek immediate help from your ophthalmologist if you have any of the signs of a detached retina.

Signs of retinal detachment include a sudden onset of:

- floaters (small specks, dots, circles, lines or cobwebs in the field of vision)
- flashes (flashing lights or lightning streaks in the field of vision)
- dark shadows in your peripheral (side) vision

Summary

Marfan syndrome is a genetic condition that affects the body's connective tissue. Marfan syndrome can affect many different parts of the body.

Eye problems are common in people with Marfan syndrome. Most people with Marfan syndrome have myopia (nearsightedness) and astigmatism. More than half of people with Marfan syndrome have a condition called ectopia lentis. With ectopia lentis the eye's lens becomes dislocated.

Marfan syndrome is usually an inherited genetic disorder. Three out of four people with Marfan syndrome inherited it from a parent.

A dislocated lens in the eye is often the first sign of Marfan syndrome. Because of this, an ophthalmologist can play an important role in diagnosing the disorder.

Your ophthalmologist can treat many of the eye problems associated with Marfan syndrome.

If you have any questions about your vision, speak with your ophthalmologist. He or she is committed to protecting your sight.

Get more information about Marfan syndrome from EyeSmart—provided by the American Academy of Ophthalmology—at **aao.org/marfan-syndrome-link**.

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