Marfan Syndrome

WHAT IS MARFAN SYNDROME?

Marfan syndrome is an inherited (runs in families) medical problem that affects the strength of connective tissues in the body. Connective tissues hold the body together and play an important role in its growth and development. Since connective tissue is found throughout the body, Marfan syndrome can affect many areas, such as the bones, joints, eyes, heart, blood vessels, nervous system, skin, and lungs.

Marfan syndrome affects both girls and boys of all races. About 1 in 5,000 people are born with the disorder, and an estimated 200,000 people in the United States are currently diagnosed with it.

WHAT CAUSES MARFAN SYNDROME?

Marfan syndrome is caused by a change (mutation) in the gene FBN1. This gene tells the body how to make a certain protein called fibrillin-1 which is very important for connective tissues.

Most people (3 out of 4) with Marfan syndrome inherit it from a parent who also has the condition. About 15–25% of people with Marfan syndrome do not have a family

history. Instead, the change in the gene happens on its own. The chance of getting Marfan syndrome spontaneously is about 1 of 20,000 births.

HOW IS MARFAN SYNDROME INHERITED?

Marfan syndrome is inherited in an **autosomal dominant** manner. This means if a parent has Marfan syndrome, each child has a 50% chance of having it too.

WHAT DOES MARFAN SYNDROME LOOK LIKE, AND HOW IS IT DIAGNOSED?

The diagnosis of Marfan syndrome is made when certain findings are seen together on exams and testing. Common findings include a lens in the eye that is out of its normal spot (ectopia lentis), problems and weakness in the wall of a big artery in the body called the aorta (aortic dissection), being very tall with long arms and legs and having a heart murmur (extra heart sound). Marfan syndrome affects people differently. Not every person with Marfan syndrome has all the same findings, and some people without Marfan syndrome may look like they do. Not every person with Marfan syndrome gets every problem that can be seen in Marfan syndrome.

IS THERE A GENETIC TEST FOR MARFAN SYNDROME?

Yes. There is a genetic test for the FBN1 gene. Sometimes the results can be confusing, so families may need a genetic counselor to explain them.

WHAT ARE THE EYE PROBLEMS SEEN IN MARFAN SYNDROME?

Eye problems occur in the majority (80%) of patients with Marfan syndrome. Common problems include:

- Nearsightedness (myopia)
- Irregular shape to the eye (astigmatism)
- Lens out of place (ectopia lentis)
- Thin or flat cornea
- Cloudy lens (cataract)
- High eye pressure (glaucoma)
- Eye movement problems (strabismus)
- Loosening of the light-sensitive layer in the back of the eye (retinal detachment)

Retinal detachment is more common in teens and young adults with Marfan syndrome. It is a medical emergency and usually needs surgery right away to prevent loss of vision.

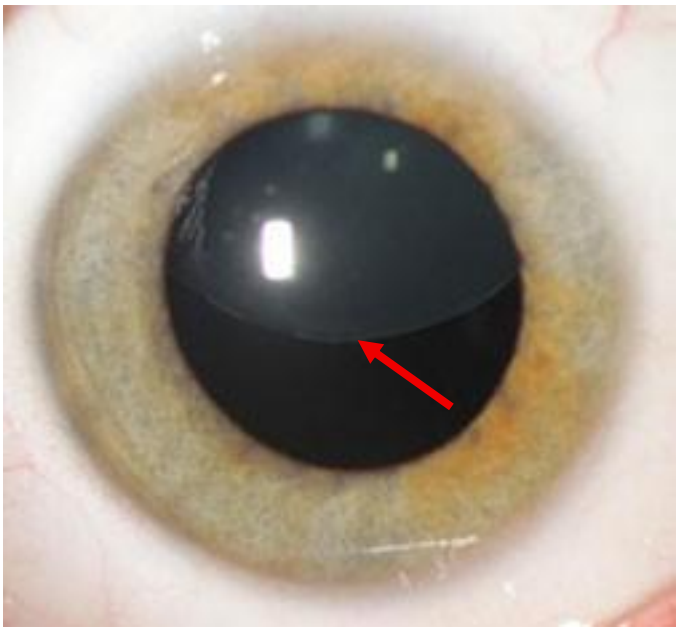

Fig. 1: Subluxated lens (ectopia lentis). The lens is seen to be moved out of place in the pupil (dark spot in the center of the colored part of the eye). The lens has a round shape and, in this picture, a slightly gray color. The round edge of the lens is seen in the middle

of the pupil because it has shifted up. (see red arrow) Under normal circumstances, the edge of the lens should not be visible in the middle of the pupil.

WHAT IS ECTOPIA LENTIS?

Ectopia lentis is when the lens inside the eye is not in the right place, it is also called lens subluxation or lens dislocation. This happens because the fibers (zonules) that hold the lens in place are weak. The lens moves away from its normal location in the eye so that the person is not looking through the center of their lens, but through or beyond the edge. The dislocation can get worse over time. About 6 out of 10 people with Marfan syndrome have this problem and is one of the major medical findings that often need to be present to diagnose this condition [See figures 1 and 2]. If a person does not have dislocated lenses, though, it does not mean that they do not have Marfan syndrome.

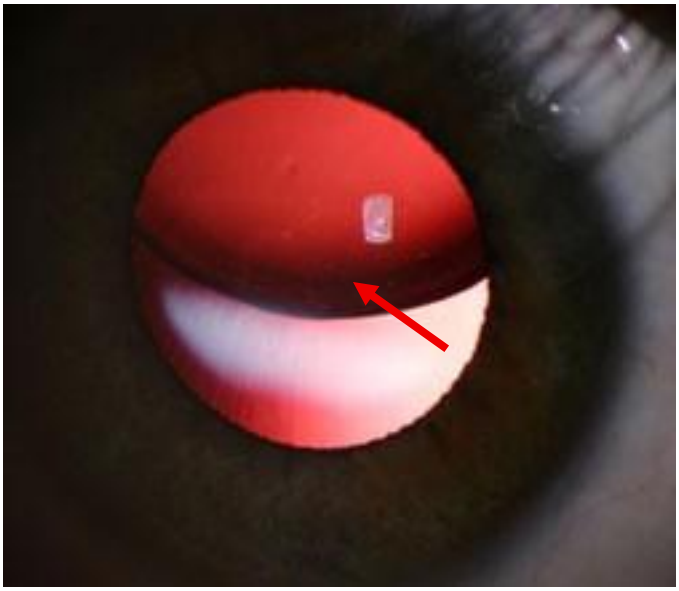

Fig. 2: Same lens as Fig. 1 with retroillumination (a special kind of exam light) The lens is often dislocated (shifted) upwards (see red arrow).

HOW DOES MARFAN SYNDROME AFFECT THE HEART AND BLOOD VESSELS?

The most serious problems with Marfan syndrome affect the heart and blood vessels in the body. There can be problems with one of the valves in the heart called the mitral valve. The valve can be too large and can move in the wrong direction. This is called mitral valve prolapse (MVP) and can make a heart murmur (an extra heart sound) when a doctor listens to the heart. In about one third of people with MVP, blood leaks backward through the valve (mitral valve regurgitation). This leaky valve can lead to shortness of breath, a fast and irregular pulse, or feeling very tired.

For people with Marfan syndrome, problem can also occur with the aorta (the major blood vessel/artery coming off of the heart). The aorta can be too large (aneurysm). This can cause problems with blood flow (aortic regurgitation) or a tear in the middle layer of the aorta (dissection). This is the most common life-threatening problem in Marfan syndrome. It is very important that people with Marfan syndrome have regular cardiac (heart) exams.

HOW DOES MARFAN SYNDROME AFFECT THE BONES?

Many patients are checked for Marfan syndrome because of how it affects the bones. People with Marfan syndrome often look tall and thin, with long and thin arms, legs, fingers (arachnodactyly), toes as well as loose joints. Other signs of the disease include:

- Flat foot arches (pes planus).
- Curved spine (scoliosis and kyphosis)
- Breastbone that sticks out or curve in (pectus carinatum and excavatum).
- High, narrow roof of the mouth with crowded teeth

- Narrow and long face.

Persons with Marfan syndrome may also have arms longer than what would be expected for their height. It is believed that Abraham Lincoln had Marfan syndrome.

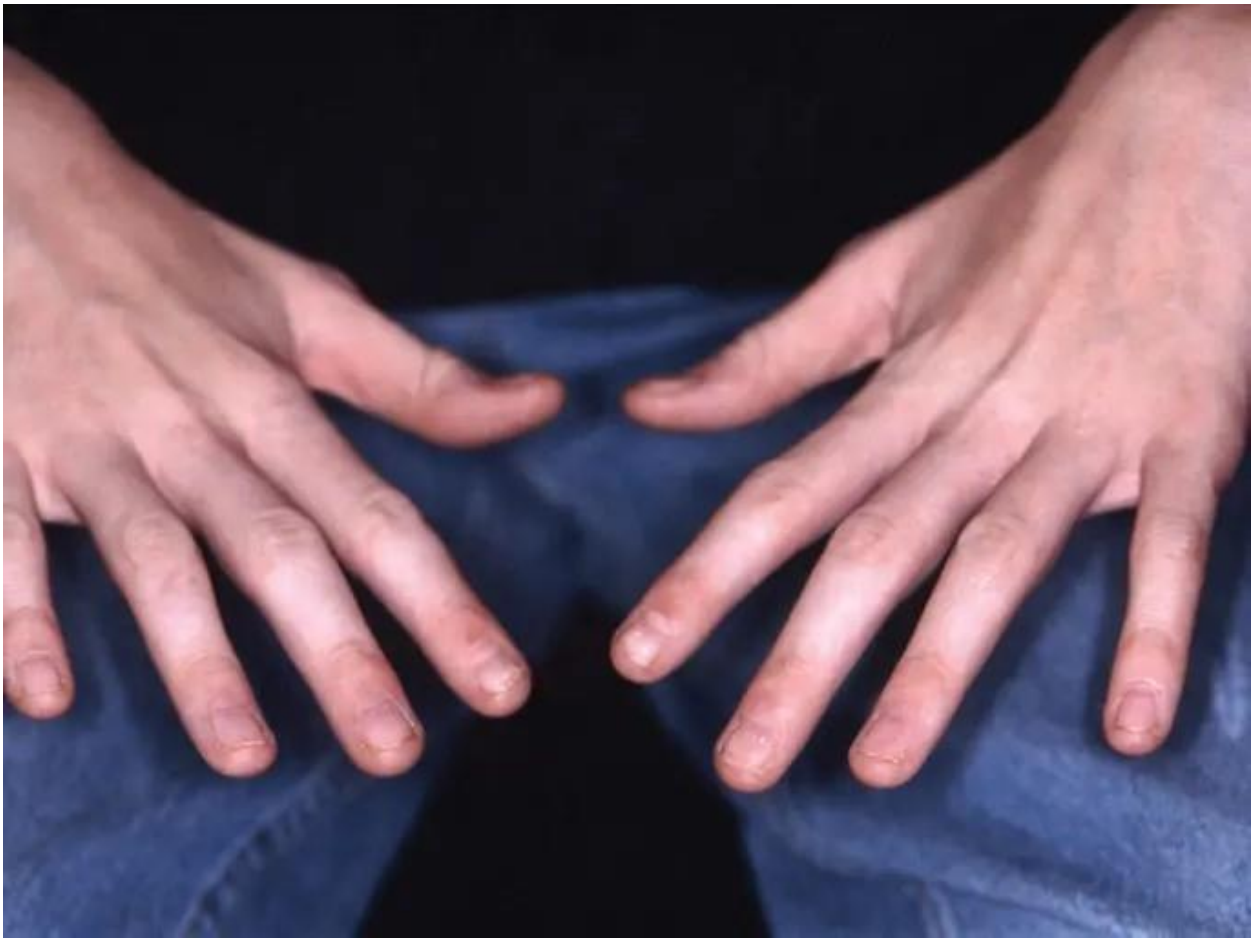

Figure. 3: Long, thin fingers in Marfan syndrome (arachnodactyly).

HOW DOES MARFAN SYNDROME AFFECT THE SKIN?

Marfan syndrome may cause striae, or bands of thin wrinkled skin. At first, the bands in the skin can look red, but they can turn purple and white over time. They tend to show

up in areas where joints are doing a lot of work — such as the shoulders, hips, and lower back.

HOW DOES MARFAN SYNDROME AFFECT THE LUNGS?

People with Marfan syndrome can have poor flexibility in the smaller air sacs of the lungs, but it generally does not cause a serious problem. One out of twenty people with Marfan syndrome can get feeling of breathlessness or chest pain from a spontaneous collapse of the lung (pneumothorax). Some people with Marfan syndrome also have problems with abnormal breathing while they sleep (obstructive sleep apnea).

DOES MARFAN SYNDROME HAVE A CURE?

No. There is no cure for Marfan syndrome at this time. However, early diagnosis and treatment for its medical problems in Marfan syndrome can significantly improve quality of life and life expectancy.

WHAT IS THE PROGNOSIS FOR PERSONS WITH MARFAN SYNDROME?

Health care advancements have helped people with Marfan syndrome to be able to live longer. In 1972 the average life span was about 45 years, but now on average people with this condition live to be about 70 years old.

HOW SHOULD MARFAN SYNDROME PATIENTS BE MONITORED?

- Regular heart tests (echocardiograms) and check-ups as recommended by a cardiologist.

- Regular eye exams to check vision, lens, refractive errors (problems focusing due to a need for glasses), and the retina.
- Monitoring of the bones and skeleton.
- Avoiding heavy exercise and contact sports will protect the body

WHAT TREATMENTS ARE THERE FOR MARFAN SYNDROME?

There are different treatments for the different problems that people with Marfan syndrome can have. In terms of eye problems, people with dislocated lenses and refractive errors (problems focusing due to a need for glasses) may be treated with glasses or contact lenses to help with vision. If the vision does not get better with these treatments, surgery may be helpful. Children with cataracts (cloudiness in the lens of the eye) may also need glasses, contacts or surgery to help their vision. Glaucoma (high eye pressure) may need treatment with medications or surgery. If there is a retinal detachment, surgery may be needed to fix this problem. As the connective tissues in people with Marfan syndrome are weak, eye surgery can be more challenging and may have a higher risk. Speak with your eye doctor (ophthalmologist) if you have more questions about eye surgery in Marfan syndrome.

Different problems with the heart, lungs and bones have different treatments as well. Be sure to speak with your primary care doctor and specialists if you have questions about those treatments.

SUMMARY FOR PATIENTS AND CAREGIVERS

Marfan syndrome is a condition that affects connective tissue, which supports many parts of the body. It can cause problems with the eyes, heart, blood vessels, bones, lungs, and skin. The most serious risks are heart and aorta problems, which need regular check-ups. Eye problems, such as lens dislocation or retinal detachment, are also common. Although there is no cure, many treatments can improve quality of life. With proper care, people with Marfan syndrome now live much longer than in the past.

WHERE CAN I FIND MORE INFORMATION ABOUT MARFAN SYNDROME?

For more information please visit:

- https://marfan.org (the Marfan Foundation website)
- https://eyewiki.org/Marfan_Syndrome

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