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 provide the framework that holds the body together and play an important role in growth and development. Connective tissue is found throughout the body, so patients with Marfan syndrome may have problems with many different parts of the body including bones, joints, eyes, heart, blood vessels, nervous system, skin, and lungs.

Marfan syndrome affects both girls and boys and all ethnic groups. 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?

It is caused by a mutation in the gene FBN1. This gene tells the body how to make a certain protein called fibrillin-1. Fibrillin-1 is a very important part of connective tissue. Three out of every four people with Marfan syndrome inherit the disorder from a parent who also has Marfan syndrome. One out of four people with Marfan syndrome have a spontaneous mutation in the gene — meaning neither parent has Marfan syndrome. The chance of getting Marfan syndrome spontaneously is about 1 of 20,000 births.

HOW IS MARFAN SYNDROME INHERITED?

It is an autosomal dominant medical problem. This type of inheritance means each new child of a parent with Marfan syndrome has a fifty percent chance of getting the disease.

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

The diagnosis of Marfan syndrome is made when certain medical problems are found on exams and testing. Some of the more common problems in people with Marfan syndrome are ectopia lentis (when the lens of the eye is

not in the right spot, discussed later), aortic dissection (a large artery in the body has a weakness in its wall), family history of Marfan syndrome, and genetic testing showing the abnormal FBN1 gene. Other signs of Marfan syndrome include: being very tall, having long arms and legs and having a heart murmur. There is a wide range of variability in how Marfan syndrome affects people. Not every person with Marfan gets every problem that can be seen in Marfan syndrome. Also, some people without the syndrome have one or more medical problems that look like Marfan syndrome- which can be a reason for a referral for a person to be checked out for Marfan syndrome. A person is diagnosed with Marfan syndrome when they have multiple signs of this disease.

IS THERE A GENETIC TEST FOR MARFAN SYNDROME?

YES. There is genetic testing for the mutation in the fibrillin-1 gene, FBN1, that causes Marfan syndrome. The results of the testing are not always straightforward and may need help from a genetic counselor to understand what the results mean.

WHAT ARE THE EYE PROBLEMS SEEN IN MARFAN SYNDROME?

Most people with Marfan syndrome have nearsightedness, or myopia, and an extra curved shape of the eye, or astigmatism. These refractive errors (problems focusing due to a need for glasses) can be very large since the connective tissue problem may affect the cornea (clear covering in the front of the eye), lens (focusing part in the middle of the eye), and how long or short the eye is. Other eye problems in Marfan syndrome include ectopia lentis (see below), corneal thinning, flattened cornea, cataracts (cloudy spot in the lens), glaucoma (high eye pressure), strabismus (eye movement problems), and retinal detachment (where the inner lining of the back wall of the eye falls off).

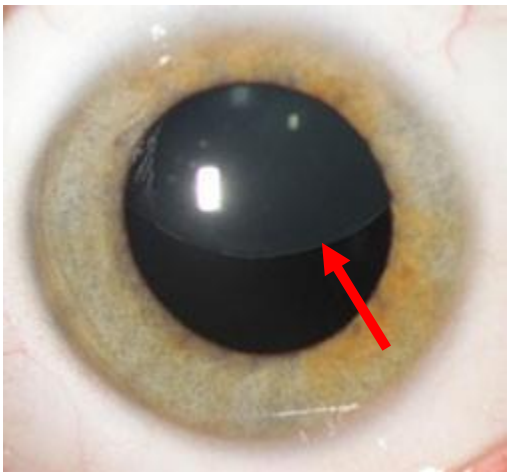

Fig. 1: Subluxated lens (ectopia lentis). The lens is seen in the pupil (dark spot in the center of the colored part of the eye). The lens has a round shape and, in this picture, a slightly gray color. The round edge of the lens is seen in the middle of the pupil because it has shifted up. (see red arrow) Normally, you should not be able to see the edge of the lens in the middle of the pupil.

WHAT IS ECTOPIA LENTIS?

Ectopia lentis is when the lens inside the eye is not in the right spot, it is also called lens subluxation or lens dislocation. 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 the edge. It can get worse over time. This movement of the lens is caused by weakness in the connective tissue that holds the lens in place (zonules). Ectopia lentis happens in roughly 60% of people with Marfan syndrome and is one of the major medical problems 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.

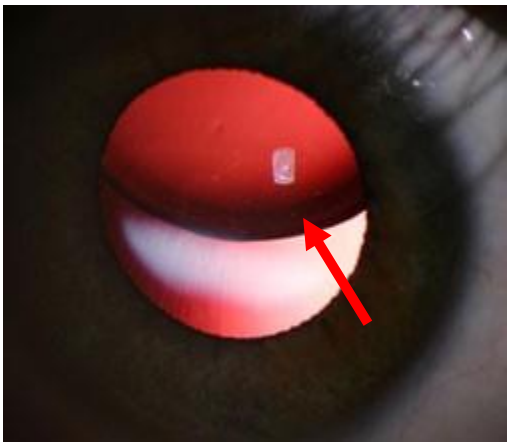

Fig. 2: Same lens as Fig. 1 with retroillumination (a different kind of lighting) The lens is often dislocated (shifted) up. (see red arrow)

HOW DOES MARFAN SYNDROME AFFECT THE HEART AND BLOOD VESSELS?

The most serious medical problems seen with Marfan syndrome involve the connective tissue of the heart and blood vessels (arteries and veins). 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 special sound when a doctor listens to the heart. In about ⅓ of people with MVP, blood leaks backward through the valve (mitral valve regurgitation), producing a heart murmur (an extra sound) that can also be heard through a stethoscope. These problems with the heart can cause breathlessness, an irregular pulse (causing palpitations), or extreme tiredness.

A different problem can occur with the aorta (the major blood vessel/artery coming off of the heart). The aorta can be too large (aneurysm) in with Marfan syndrome. This can cause problems with blood flow (aortic regurgitation) or a tear in the middle layer of the aorta (dissection). Problems with the aorta are the most common cause of death in persons with 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. A person with Marfan syndrome will usually be tall, thin, and loose jointed/flexible. Other signs of the disease include long, thin fingers and toes (arachnodactyly) and flat foot arches (pes planus). Extra curving of the spine (scoliosis and kyphosis) and breastbone shapes that stick out or curve in (pectus carinatum and excavatum) are also common problems. The roof of the mouth (palate) may be highly arched, causing teeth to grow close together, and the face may seem narrow and long. 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.

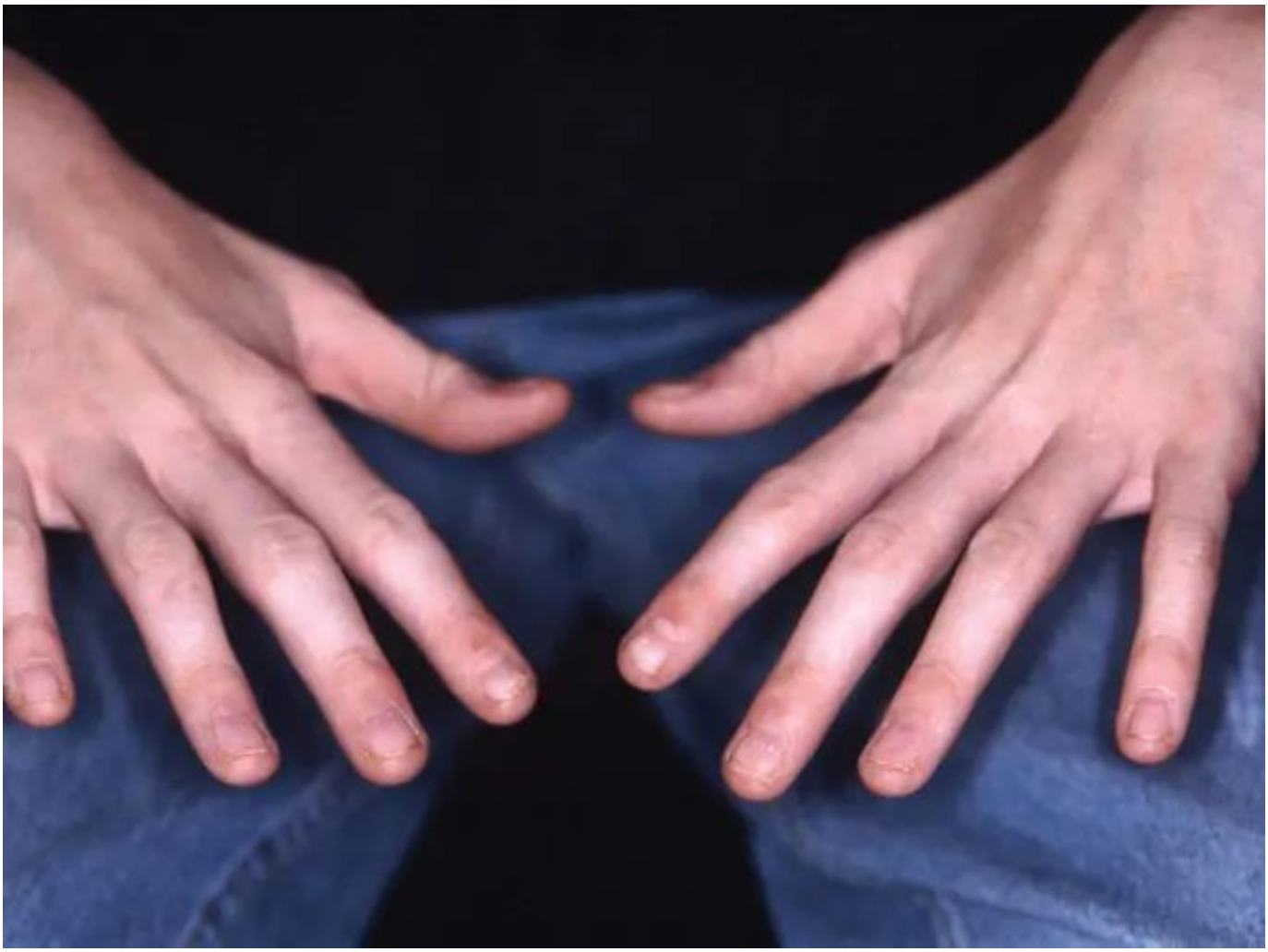

Fig. 3: Person with Marfan syndrome showing long thin fingers (arachnodactyly).

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, with early diagnosis and treatments for its medical problems, we can improve the quality of life and how long someone with Marfan syndrome lives.

WHAT IS THE PROGNOSIS FOR PERSONS WITH MARFAN SYNDROME?

People with Marfan syndrome can develop severe bone/joint, heart, and eye issues, but health care advancements have helped to improve how long people with Marfan syndrome can live. In 1972 the average life span was about 45 years, now on average people with Marfan's live to be about 70 years old.

HOW SHOULD MARFAN SYNDROME PATIENTS BE MONITORED?

- Regular testing (echocardiograms) and check-ups as recommended by a cardiologist to monitor the heart.
- A full eye exam with regular eye care follow up, paying attention to the lens, refractive errors (problems focusing due to a need for glasses), and health of the retina can help protect vision.
- Careful monitoring of the bones and skeleton is recommended.

- Lifestyle adjustments: It is a good idea to avoid of strenuous activities and contact sports to limit injuries to the connective tissues in 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. People with cataracts (cloudy spots 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. Because the connective tissues in people with Marfan syndrome are weak, eye surgery can be more challenging and higher risk. Speak with your 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 specialists if you have questions about those treatments.

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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