The doctor told Evan that he couldn't give him the OK to try out until Evan had a few medical tests. Why? Because he suspected that Evan might have a genetic disorder called Marfan syndrome.

What Is Marfan Syndrome?

Marfan syndrome is named after Antoine Marfan, the French doctor who first described the disorder in 1896. Marfan syndrome affects the body's connective tissue. Connective tissue is found everywhere in the body. Think of it as a sort of "glue" that helps support your organs, blood vessels, bones, joints, and muscles.

In people with Marfan syndrome, this "glue" is weaker than normal. That's because of a defect in the body's production of fibrillin (pronounced: fuh-**brill**-in), a special type of protein that's found in connective tissue. Weakened connective tissue can lead to problems in many parts of the body, especially the heart, eyes, and joints. For example, people with Marfan syndrome may have very loose, flexible joints as a result of these connective tissue problems.

Although Marfan syndrome has no cure (so the person will always have it), the good news is that doctors can treat just about all of its symptoms. Just a few decades ago, most people with the disease didn't live past 40. Now, thanks to new research and treatments, people with Marfan syndrome who are diagnosed early and get good medical care have about the same lifespan as everyone else.

What Causes It?

Marfan syndrome is pretty rare. It only happens to about 1 in every 5,000 people.

Marfan syndrome is a genetic disorder. Genetic disorders are caused by a change in genes that is either inherited (passed on from parent to child) or that happens during very early development in the womb. In the case of Marfan syndrome, a defect in a gene found on chromosome 15 causes problems in the production of fibrillin. The gene is named FBN1.

Although some genetic disorders can affect people of one gender or a particular ethnic heritage more than others, Marfan syndrome can affect both girls and guys from all ethnic backgrounds.

About 75% of the time, the gene for Marfan syndrome runs in families, getting passed down to children from parents who have the disease. A child born to a parent who has Marfan syndrome has a 50% chance of having the disease too.

In the remaining 25% of cases, though, neither parent has the disease. This means the genetic mutation responsible for causing Marfan syndrome occurs in either the egg or sperm cell at the time of conception. No one knows what causes this mutation, but a child born with Marfan syndrome then has a 50% chance of passing it on to his or her children.

What Are the Signs and Symptoms?

People who have Marfan syndrome tend to share certain physical traits:

They are often (but not always) much taller than their peers and have a lean, lanky build with disproportionately long arms and legs. Their fingers and toes are usually long and thin.

Their joints are loose and flexible.

They may have myopia (nearsightedness) or other vision problems (such as lens dislocation).

They may have certain facial characteristics, including a long, thin face; deep-set eyes; a small bottom jaw; a high, arched roof of the mouth; and crowded teeth.

Not everyone with these characteristics has Marfan syndrome, of course. Does your friend have it just because he is tall and thin? Probably not. If you're extremely nearsighted, does it mean you have the disorder? No, again.

People who have Marfan syndrome have very specific symptoms that usually happen together. When these symptoms show up as a group, it alerts doctors to the possibility that a person might have the condition.

Other symptoms of Marfan syndrome can affect the skin and lungs. These symptoms are generally less common and less serious, especially in kids and teens.

Although people with Marfan syndrome often have similar physical features, the disease doesn't affect everyone in the same way. Some people have very mild symptoms, while others have severe ones — even within the same family. This is known as **variable expression**, and it makes it almost impossible to predict how the disease will progress in any affected individual.

How Do Doctors Diagnose It?

Several different types of doctors must be involved in diagnosing Marfan syndrome. They include a **geneticist** (a doctor who specializes in disorders of the genes), a **cardiologist** (heart doctor), an **ophthalmologist** (eye doctor), and an **orthopedist** (bone doctor).

A geneticist will ask whether anyone else in the family had similar symptoms and may even ask if anyone in your family died early of a heart-related death. Then the geneticist will probably do some pretty painless exams, such as taking detailed skeletal measurements, including arm span (hold your arms out to your side — that's your arm span). This test can help because people with Marfan syndrome often have an arm span that's greater than their height. The geneticist may order a blood test to check for a change in the FBN1 gene.

The cardiologist will also do some painless tests. He or she may listen to the heart with a stethoscope to check for a murmur, then may order an X-ray of the chest, an electrocardiogram (or EKG, which measures electrical impulses in the heart), and an echocardiogram (a test that uses sound waves to produce a picture of the heart) to check the size of the aorta and to make sure the valves are functioning well.

The doctor told Evan that he couldn't give him the OK to try out until Evan had a few medical tests. Why? Because he suspected that Evan might have a genetic disorder called Marfan syndrome.

What Is Marfan Syndrome?

Marfan syndrome is named after Antoine Marfan, the French doctor who first described the disorder in 1896. Marfan syndrome affects the body's connective tissue. Connective tissue is found everywhere in the body. Think of it as a sort of "glue" that helps support your organs, blood vessels, bones, joints, and muscles.

In people with Marfan syndrome, this "glue" is weaker than normal. That's because of a defect in the body's production of fibrillin (pronounced: fuh-**brill**-in), a special type of protein that's found in connective tissue. Weakened connective tissue can lead to problems in many parts of the body, especially the heart, eyes, and joints. For example, people with Marfan syndrome may have very loose, flexible joints as a result of these connective tissue problems.

Although Marfan syndrome has no cure (so the person will always have it), the good news is that doctors can treat just about all of its symptoms. Just a few decades ago, most people with the disease didn't live past 40. Now, thanks to new research and treatments, people with Marfan syndrome who are diagnosed early and get good medical care have about the same lifespan as everyone else.

What Causes It?

Marfan syndrome is pretty rare. It only happens to about 1 in every 5,000 people.

Marfan syndrome is a genetic disorder. Genetic disorders are caused by a change in genes that is either inherited (passed on from parent to child) or that happens during very early development in the womb. In the case of Marfan syndrome, a defect in a gene found on chromosome 15 causes problems in the production of fibrillin. The gene is named FBN1.

Although some genetic disorders can affect people of one gender or a particular ethnic heritage more than others, Marfan syndrome can affect both girls and guys from all ethnic backgrounds.

About 75% of the time, the gene for Marfan syndrome runs in families, getting passed down to children from parents who have the disease. A child born to a parent who has Marfan syndrome has a 50% chance of having the disease too.

In the remaining 25% of cases, though, neither parent has the disease. This means the genetic mutation responsible for causing Marfan syndrome occurs in either the egg or sperm cell at the time of conception. No one knows what causes this mutation, but a child born with Marfan syndrome then has a 50% chance of passing it on to his or her children.

What Are the Signs and Symptoms?

People who have Marfan syndrome tend to share certain physical traits:

An ophthalmologist will probably dilate the eye pupil with special drops, then look into it with a slit lamp, a special type of microscope that can help detect lens dislocation and other eye problems.

An orthopedist will check for curvatures of the spine and breastbone, joint problems, and any other bone abnormalities.

If no one else in the family has the disorder, the patient has to show symptoms in three organ systems or physical symptoms and a known disease causing change in the FBN1 gene, before doctors can diagnose Marfan syndrome.

Symptoms of Marfan syndrome may be apparent when a child is very young, while some people don't have noticeable symptoms until their teen years or even adulthood. But some people go through life never knowing they have the disease, and that can be dangerous. Getting diagnosed early means doctors can do a lot to help keep someone healthy.

What Can Happen if Marfan Syndrome Is Not Treated?

It's important to diagnose Marfan syndrome early because there can be serious complications when symptoms are not treated.

The most serious complication of Marfan syndrome involves the heart. Over time, the disorder can cause the aorta — the large artery that carries blood away from the heart to the body — to stretch and dilate (widen). This is called **aortic dilation** (pronounced: ay-**or**-tik dye-**lay**-shun) and if it's not treated, the aorta can eventually begin to tear, allowing blood to leak through. A large, sudden rupture can be fatal.

It's also common for people with Marfan syndrome to have problems with their heart valves. Instead of closing tightly, the valves, which help direct the flow of blood through the heart, can become floppy, causing blood to leak backward through the heart. Leaky heart valves cause the heart to work harder and become enlarged, so they must be carefully monitored.

Marfan syndrome may also cause problems with a person's eyes and skeleton. For example, people with the condition are more likely to get a detached retina (when the light-sensitive tissue at the back of the eye comes loose) or develop scoliosis (curvature of the spine). Again, the good news about these complications is that they can be prevented when Marfan syndrome is diagnosed and treated.

How Is It Monitored and Treated?

Teens with Marfan syndrome must be followed closely by a team of doctors. Because teens' bodies grow and change so quickly, most teens will need echocardiograms at least once a year, plus frequent eye and bone exams. This helps doctors stay on top of any new problems.

Many of the complications of Marfan syndrome can be managed with medications and, if necessary, surgery. Doctors may prescribe special medicines called **beta blockers** and **ACE inhibitors**, which work to lower blood pressure and reduce wear and tear on the blood vessels. This can often delay the progression of aortic dilation. If the aorta does

eventually widen to a potentially dangerous size, or if valve leakage becomes a problem, a doctor may recommend surgery to repair or replace the damaged parts of the heart.

Teens with Marfan syndrome who are nearsighted will probably have to wear glasses or contact lenses. If the lens of the eye becomes severely dislocated or if there are other complications, surgery may be necessary to fix eye problems.

Teens who develop scoliosis may have to wear a special back brace. Sometimes severe cases of scoliosis and chest wall problems may require surgery. Some people may also choose to have surgery for cosmetic reasons.

Anyone with heart problems associated with Marfan syndrome (especially anyone who's had heart surgery) should always take antibiotics before going to the dentist to prevent bacterial endocarditis, an infection of the walls of the heart caused when bacteria enter the bloodstream.

Teen girls with Marfan syndrome also need to be aware that pregnancy puts extra strain on the heart and may increase the risk of damage to the aorta. Teen girls with Marfan syndrome who are pregnant should talk to their doctor immediately.

What's Life Like for Teens With Marfan Syndrome?

Marfan syndrome affects people differently, so life is not the same for all teens who have it. Some have severe cases of Marfan syndrome and many symptoms that require lots of medical care. Others have such a mild form that they simply need to have a checkup once a year.

There are lots of things teens with Marfan syndrome can do to help keep themselves healthy. The most important is to avoid putting extra stress on the heart. That means avoiding any sport where there's a lot of running, physical contact, muscle straining, or the chance of getting hit in the chest — things like basketball, football, baseball, gymnastics, weightlifting, and track.

That may sound like a bummer, but it doesn't mean teens with Marfan syndrome are destined to be couch potatoes. They can and should exercise and be active — they just have to do it carefully.

If you have Marfan syndrome, check with your doctor about what's safe. Safe activities are usually things like non-competitive bike riding, swimming, or dancing — just be sure that it's low impact, can be done at a slower pace, and doesn't involve any quick bursts of exertion. It's also a good idea to avoid exercising in extreme temperatures, such as really cold or hot weather. The key here is to check with your doctor before trying anything.

One thing people who have Marfan syndrome should never do is smoke or use tobacco products.

Although Marfan syndrome does **not** affect intelligence, some students may need extra help in class because of vision or other related problems. And they may need to sit out some activities in P.E. But other than that, teens with Marfan syndrome are just like everyone else . . . only maybe a little taller.

If you have Marfan syndrome, you probably know that it can sometimes be hard when you feel different from your friends. You may get sick of people commenting on your height or teasing you because you stand out. If you feel overwhelmed, talk to someone you trust — a parent, a friend, or a counselor — or look for a support group in your area.

If your friend's been diagnosed with Marfan syndrome and is upset about having to give up a favorite sport, a good way to be supportive is to find new activities to enjoy together. There are far more things teens with Marfan syndrome *can* do than things they can't.

Take Evan, for example. He was pretty bummed to learn basketball wasn't going to be in his future. But then he started taking guitar lessons — and he found that he was actually pretty good.