Marfan Does *Not* Mean Martian

By Elias Clark Turner

The Marfan Foundation, founded in 1981, is a non-profit voluntary health organization that creates a brighter future for everyone affected by Marfan syndrome and related disorders.

- We pursue the most innovative research and make sure that it receives proper funding.
- We create an informed public and educated patient community to increase early diagnosis and ensure life-saving treatment.
- We provide relentless support to families, caregivers, and healthcare providers.

We will not rest until we've achieved victory—a world in which everyone with Marfan syndrome or a related disorder receives a proper diagnosis, gets the necessary treatment, and lives a long and full life.

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By Elias Clark Turner

Illustrations by Alexandra Dubow

This book is dedicated to Dr. Hal Dietz with gratitude
for all he does each day to improve the lives
of people with Marfan Syndrome.

The day that Marvin moved into the neighborhood, his family arrived in a silver automobile with doors that opened like wings spreading out and upwards. First Marvin's dad got out of the car, stretched, and I noticed right off the bat what a tall guy he was. His arms looked longer than normal and I wondered how he had ever fit into his car. He was thin as a pencil and his glasses were thick with big black rims. He didn't stand up straight which kind of made him look like a question mark.

Then Marvin's mom got out. She looked absolutely normal—about as tall as most of the grown-ups I knew. She looked around and smiled, saw me watching, and she waved.

Next Marvin stepped out. The first thing I noticed was his chest. It stuck out strangely in a point from the middle of his shirt. Either that or he was carrying something pointy in his undershirt. He also had these really long skinny arms. But in most other ways he resembled his father, except that he looked to be about my age.

Looking at him standing next to his dad, I wondered if everyone in this family had always looked that way.... And then I saw him and his dad give each other a "thumbs up" about the new house. I had never seen thumbs like those before.

Before heading into his new house, the boy looked around. But unlike his mom, when he saw me, instead of waving, he looked away and quickly walked up the path. I hoped he was just shy. But he sure seemed strange. I seriously started wondering whether a family of Martians had just moved in across the street.

That was on Sunday.

On Monday, the first day of school for the new year, my mom drove me to Thomas Edison Elementary where I was to become a member of Miss Barbasol's fourth grade class. Her last year's class had really loved her. I was excited she would be my teacher.

As I closed the car door, I noticed the same silver car I had seen the day before sitting in the parking lot. It looked like that the strange kid I had seen yesterday was going to be attending my school.

My mom walked me to my classroom where Miss Barbasol was standing outside greeting her new pupils and talking to their parents. I noticed the new kid's parents ahead of us in line. They seemed to be talking with the teacher for a very long time. When we got to the door of the classroom, I said hello to Miss Barbasol and hugged my mom good-bye. I went inside and found that there were name cards on all of the desks. I went around until I located my correct place at a table. My table partner was going to be someone named Marvin. All I could think to myself was "Marvin the Martian... like the cartoon." Then it occurred to me who Marvin might be.

Our table was in the front of the classroom just a couple feet from the whiteboard. I liked sitting close to the front because I could always see what the teacher was doing. The first bell rang and sure enough, the kid I saw yesterday walked in. He came over and sat down at my table. He kind of slumped over and I could clearly see the low end of his chest poking out under his shirt. It looked like it was an uncomfortable way to sit. I figured he might be sitting that way so no one would notice but it was kind of obvious. Now I saw he was wearing glasses.

"I have to sit close to the board," the boy kind of mumbled. It helps me not get headaches."

"Wow," I thought. This guy has it hard.

I felt a little worried but I pushed past it and introduced myself to my table partner.

"I'm Joe Smith," I said.

He quietly replied, "My name is Marvin McGilligan. My family just moved into town."

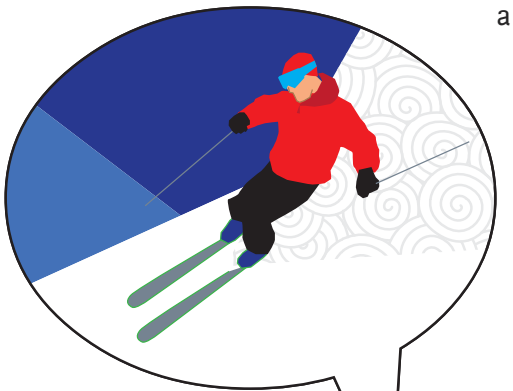
"I saw you arrive yesterday. I live across the street from you. Your dad has a really cool car. Looks like a spaceship. What kind of car is it?" I asked.

Marvin sat up a little straighter and said "Yeah, I think it's cool, too. It's called a Delorean. I like how the doors lift up like wings. My dad works for the company that built it. They give him one to drive around because he is in advertising."

"That's awesome!" I blurted. "My dad's a contractor. He specializes in kitchens and bathrooms." Then I realized how funny that sounded. Marvin just smiled.

Just then, the second bell rang with a loud buzz and Miss Barbasol entered the classroom and shut the door. She came up front and introduced herself, then announced we would go around the classroom and each of us would give our names and say something we did over the summer.

My friend Jimmy talked about how he went to a YMCA day camp and learned how to swim. Jeanine Focaccio told about taking horseback riding lessons and that she had even had tried jumping her horse once. That was cool. I could tell some of the girls were jealous.



Sam had gone with his family to Chile to go skiing which everybody thought was funny because it's so hot in the summer. But he was skiing in a country called Chile so it kind of made sense.

I talked about how my friends had taken me down to the skate park over the summer and taught me how to Ollie on a skateboard.

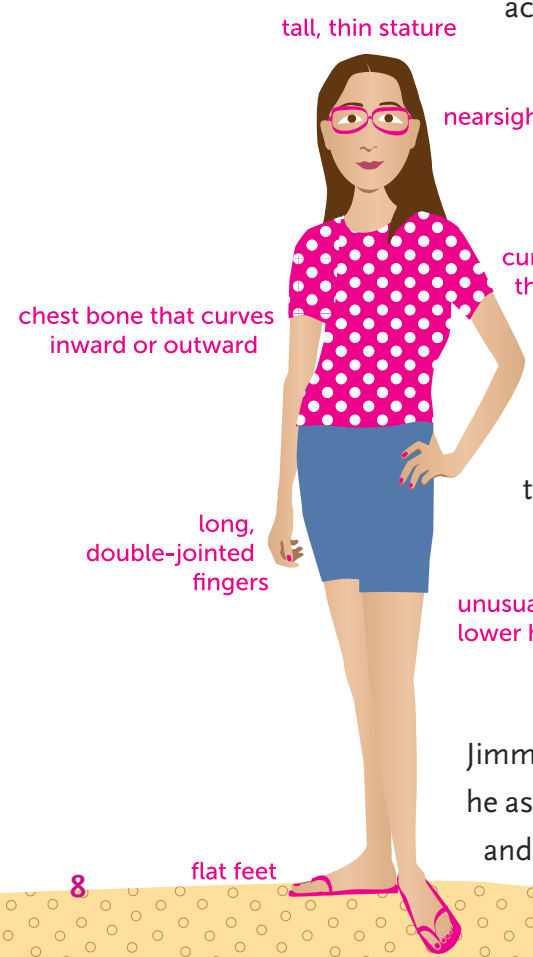
Marvin listened to all of these adventures with a sad look on his face. When it got to his turn to share, he said he had taken a computer game class—which I thought was cool— but it did not get nearly as many wows as skiing in chilly Chile. I wondered if he hadn't had any big adventures because of a problem with his chest.

Eric talked, then Suzy, then eventually everybody in the room had given their report and it was time to go out to the field to have P.E. Miss Barbasol announced with enthusiasm that today we were going to play Dodge Ball. Marvin looked worried. Somehow I got the feeling that Dodge Ball was not his favorite game. We all got up, pushed in our chairs, and made our way to the door.

I was almost out the door when I noticed that Marvin had hung back and was talking quietly with Miss Barbasol. She put a sympathetic hand on his shoulder and told him in a hushed voice that it was okay for him to go to the library while the rest of the class went outside. He seemed relieved but still sad. He took a hall pass from our teacher and walked slowly off toward the library. Maybe he was used to this.

When we got out to the field, Miss Barbasol was met by our school nurse, Mrs. Atkins. She asked us each to grab a bucket chair from her stack and to sit down quietly facing her. "Boys and girls," she said, "you have a new student in your class this year. His name is Marvin and he is unable to participate in our P.E. activities this year because he has a medical condition that makes it easier for him to get hurt.

It is called Marfan Syndrome, and basically it means that Marvin's body is not as strong as yours and mine. Marfan syndrome is a disorder of the connective tissue. Connective tissue holds all parts of the body together and helps control how the body grows. Because connective tissue is found throughout the body, Marfan features can occur in many different body systems, including the heart, blood vessels, bones, joints, and eyes. Sometimes, the lungs and skin are also affected.



"People who have Marfan syndrome have to follow special physical activity guidelines. They are not supposed to play competitive sports such as basketball and football because it puts too much stress on the aorta, the main blood vessel that carries blood from the heart. Sports and other physical activity can also cause eye problems for people who have Marfan syndrome.

"In every other way, Marvin is just like us. He looks different than we do. But you must try to look past this and get to know Marvin for who he is.

"Now, none of you are to tease him or leave him out of fun things because of it. His problem is inherited. His father has this problem, too. Marvin did not ask to be born with a genetic disorder and he wants to be treated like all of you. So please go out of your way to include Marvin in your activities and make him feel welcome."

Jimmy raised his hand. "Can't he just go to the doctor and get medicine for it?" he asked. Miss Barbasol answered that Marfan syndrome is a genetic disorder and there is no cure yet.

“Marvin can take medicine to protect his aorta which is connected to his heart, and he can take medicine to stop pain when he gets hurt, but there is no medicine that can take his problem away.”

Everyone was quiet. Jeanine Focaccio sniffled. She whispered to Nancy Shoemaker, “That’s really sad. I guess he can’t ride horses.”

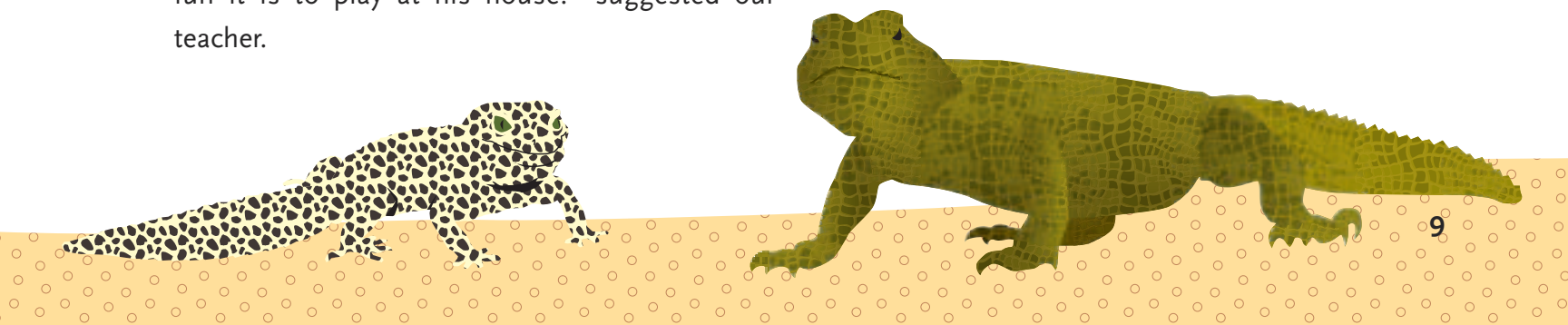
“Or ride a bike,” added Nancy.

“What CAN he do,” I asked out loud?

“Marvin likes doing other activities like building Legos, playing computer and video games, reading, and I hear he has some unusual pets,“ Miss Barbasol explained.

“It sounds like playing at Marvin’s house would be really fun,” said Sam. “I only have one good Lego kit. I bet he has more. And my mom won’t let me have any pets at all.”

“Why don’t you invite him to play at your house sometime, Sam, and maybe you will find out just how fun it is to play at his house!” suggested our teacher.



After the discussion ended, Mrs. Atkins complimented us on our listening and our questions and she collected our buckets. Then Miss Barbasol told us that we still had time to play a short game of Dodge Ball, so we split into teams and moved over to the blacktop. I had a lot of fun, but I kept thinking about Marvin and felt sorry for him. I wished he could be having fun, too. I decided I would invite him over to our house after school.

When P.E. ended, we had Math, then Library. After Library, it was time for lunch. My mom had packed me an extra-special lunch that day- a submarine ham and cheese sandwich and a bag of fresh pretzels. I even had two juice packs. I sat down by Marvin, hoping to invite him over after school. Some kids had bought the school lunch. It looked pretty bad.

But Marvin and I had homemade lunch in common He was slowly eating a PB&J on whole wheat bread. Someone at his house was a health nut. He had carrot sticks. Poor Marvin. Carrot sticks. Now I really felt sorry for him and I offered him some of my pretzels and my extra juice. His eyes lit up and he gladly accepted them.

“Marvin, we just got this new trampoline in our backyard. It’s not that big but it’s lots of fun. You wanna come over and try it out after school?” I asked.

“Well, I would but I am not allowed to play on trampolines because I might twist my ankles,” he explained, but can we do something else?”

“Sure,” I said. Then I had to stop and think. What COULD Marvin do at my house? “We have this big tin of Lincoln Logs. Maybe we could build something with them.”

“Sure, that sounds like fun. I love building things. But I have to check with my mom.”

After school at pick up, I saw Marvin’s silver car pull up. He went over but before getting in, leaned in and asked his mom if he could come over to play at my house that day. Then he came back over to me looking disappointed and said, “I can’t come over. I have to go get an echocardiogram. But can I come over tomorrow instead?”

“Sure,” I answered. “What’s an echocardiogram?”

So Marvin took a deep breath and explained. He said it was a test where the doctor took pictures of his heart to see if the tubes leading up to his heart and away from his heart were the right size.” I was confused.

“How do they do that without cutting open your chest!” I blurted. “Is that why you have that big point there?” It was a slip of the tongue.

Marvin laughed and said, “No, Silly. That’s just the way the bone in my chest grows. And the doctor’s assistant takes the pictures with a special machine called ultrasound that doesn’t look like a camera at all.”

I suddenly remembered when my mom got an ultrasound when she was pregnant with my little brother. I got to watch. Now I understood. "So they cover you in slimy goop and stick wires on you to take the pictures?" I asked.

"Yep. Then they slide this thing that looks like a microphone around in the goop and they click it to take the pictures on a computer screen. I can watch that or watch a movie while I am being slimed."

"That sounds like it tickles."

"It sure does."

“Marvin,” called his mom. “C’mon, Honey. We don’t want to be late.”

“I gotta go, Joe. See you tomorrow,” he said happily.

“Bye, Marvin. Have a nice sliming,” I called after him.

On Tuesday, Marvin whistled as he came into class. He sat down at our table and said, “Mom gave me permission to come over and play today. Is it okay with your mom?”

“Sure is,” I said. “She can pick us up after school. Maybe we can stop for frozen yogurt on the way home.”

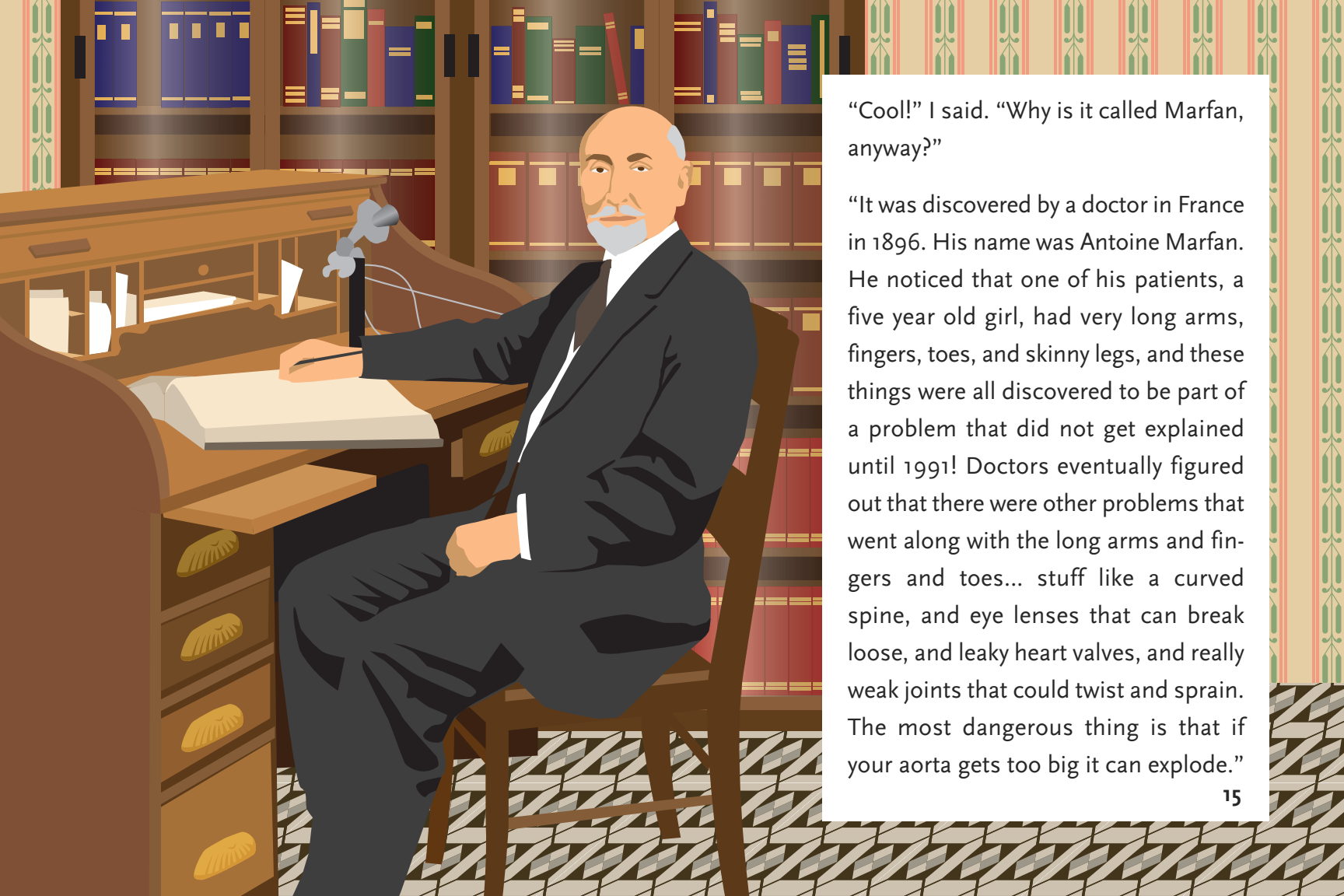
“Sounds delicious,” he said. “I want to get mine with three toppings. My mom only lets me have granola.”

“Okay, then let’s load up on sugar. So how’d your echocardiogram go?” I asked.

“Went fine, but the doctor put me on a new medication. I have to take a pill everyday. She said it might help me get stronger so I could do more someday. But it’s still experimental. I am becoming a science project.”

“Maybe we can enter you in the school science fair this year,” I joked.

“Not a bad idea,” said Marvin. “They are going to study me at Johns Hopkins University so we might as well.”



"Cool!" I said. "Why is it called Marfan, anyway?"

"It was discovered by a doctor in France in 1896. His name was Antoine Marfan. He noticed that one of his patients, a five year old girl, had very long arms, fingers, toes, and skinny legs, and these things were all discovered to be part of a problem that did not get explained until 1991! Doctors eventually figured out that there were other problems that went along with the long arms and fingers and toes... stuff like a curved spine, and eye lenses that can break loose, and leaky heart valves, and really weak joints that could twist and sprain. The most dangerous thing is that if your aorta gets too big it can explode."

“It sounds horrible. Does this mean you a walking pipe bomb, Marvin?”

“Nope. My aorta is not that bad. And I have that new medicine. Plus they keep a pretty good watch over me. There is even surgery people with Marfan get to prevent their aortas from exploding. My grandma already had that operation and she is fine now. And besides, no two people in a family have Marfan in all the same places. We are each one of us pretty unique. My dad has a curved spine but I don’t! “

“Phew!” I said, relieved that at least Marvin did not have that problem. “So your grandma even has it? How did you get Marfan, exactly?”

“It’s inherited. It’s in our genes in my family. It started on my dad’s side of the family and his mom’s mom had it first. No one knows how the first person gets it though.

“What do you mean it’s in your genes?” I asked.

“Well it’s in our chromosomes and those are in our genes,” he tried to explain. I must have looked pretty mixed up because he continued, “in 1991, a doctor was able to trace the problem to a chromosome. Do you know what a chromosome is?” I didn’t .

“Well it’s a collection of our DNA. We have 46 chromosomes in each of the cells in our bodies.

Each chromosome has many genes. And one of the genes in the 15th chromosome of people with my disorder has a bad mutation. My dad has this mutation, too. He passed it along to me. My mom doesn’t

have it at all. But just because my dad has it, there was a 50 percent chance I might get it too, and well. that's the way the cookie crumbled."

"What a tough break for you, " I said. But on the bright side, it kinda sounds like an X-Men movie." Maybe you have super powers you have never discovered!"

"Well, I have tried to move things with my mind but it hasn't worked yet. But I think I might be pretty good at music. I can figure things out on piano by ear. My doctor said a lot of people with Marfan turn out to be musicians and artists. Some of them even get famous like this guy from Broadway named Jonathon Larson who made a musical called "Rent."

"We're still young," I said. "Maybe you'll find your superpower by the end of sixth grade."

"Yeah, if I practice piano, maybe," I shrugged.

"Does your brother have Marfan Syndrome too," I asked Marvin?

"Nope," he replied. "He got a better cookie. He doesn't have to go to the doctor and take pills or anything. And he is even learning to ride a bike which makes me pretty annoyed. I mean he is only five and he gets to do everything— even learn to play soccer. I can't do that because a ball might hit me in the head and mess up my eye lenses. It's amazing where connective tissue is. But my dad says he is going to teach me to sail and to play golf next summer because those are two sports people with Marfan CAN do!"

"Okay, now I am jealous," I answered. "My dad doesn't know how to sail and he is usually too tired to do sports on the week-end. I think it's because he is building stuff all week and he has to get up really early every day. Can I come sailing with you sometime?"

Marvin laughed. "I'll tell my dad and we can invite you. I think we're going to get a boat next summer. So... what did you bring for lunch today? My mom gave me liver sausage with mayonnaise." Marvin grimaced.

"Don't worry," I told him. I asked my mom for a second bag of Fritos and for some insane reason, she agreed. I think maybe my dad replaced the downstairs toilet handle before breakfast and that made her really happy.

"Is that what you meant when you said your dad specializes in bathrooms?" joked Marvin. We both smiled. It was going to be a fun year.

Glossary of Terms

Aorta: The large artery that carries blood away from the heart to other parts of the body.

Cell: The basic subunit of any living organism; the simplest unit that can exist as an independent living system.

Chromosome: A structure found in the cell nucleus that contains the genes; chromosomes are composed of DNA and are found as pairs in each cell. Each parent contributes one chromosome to each pair, so every child gets half of his/her chromosomes from the mother and half from the father.

Connective Tissue: It is the glue and scaffolding of the body that includes the substances between cells (extracellular matrix) consisting of collagen and elastic fibers.

DNA, *Deoxyribonucleic Acid*: The substance of heredity; a large molecule that carries the genetic information necessary for all cellular functions, including the building of proteins.

Echocardiogram: A painless test that uses sound waves to take pictures of the heart and blood vessels close to the heart.

Genes: Tiny parts of each cell passed from parents to their children that tell the body how to grow and function.

Genetic Disorder: A pathological condition caused by an absent or defective gene or by a chromosomal aberration. Also called hereditary disease, inherited disorder.

Heart Valves: A part of the heart that opens and closes with each heartbeat. Our hearts have four valves that work together to help keep blood flowing through the heart and body.

Inherited: When a person receives a feature from a parent through the genes. The feature can be how a person looks or, as with Marfan syndrome, can be a genetic disorder.

Joints: Where two bones are joined together to allow the bones to move. For example, elbows, knees, ankles, and shoulders are all joints in our body.

Marfan Syndrome: Marfan syndrome is a genetic disorder that weakens multiple body systems, including the heart, blood vessels, bones and joints, lungs and eyes. The life-threatening part of Marfan syndrome is the weakening of the aorta, the large blood vessel from the heart. Early diagnosis and treatment are essential for maximizing life expectancy.

Migraine Headaches: The most common type of vascular headache involving abnormal sensitivity of arteries in the brain to various triggers resulting in rapid changes in the artery size due to spasm (constriction). Other arteries in the brain and scalp then open (dilate), and throbbing pain is perceived in the head.

Mutation: A permanent structural change in DNA that can cause a condition.

Spine: The column of bone known as the vertebral column, which surrounds and protects the spinal cord. The spine can be categorized according to level of the body: i.e., cervical spine (neck), thoracic spine (upper and middle back), and lumbar spine (lower back).

Ultrasound: High-frequency sound waves. Ultrasound waves can be bounced off of tissues using special devices. The echoes are then converted into a picture called a sonogram. Ultrasound imaging, referred to as ultrasonography, allows physicians and patients to get an inside view of soft tissues and body cavities, without using invasive techniques. Ultrasound is often used to examine a fetus during pregnancy.

What is Marfan syndrome?

Marfan syndrome is a disorder of connective tissue. Connective tissue holds all parts of the body together and helps control how the body grows. Because connective tissue is found throughout the body, Marfan syndrome features can occur in many different parts of the body. Most often the condition affects the heart, blood vessels, bones, joints, and eyes. Sometimes, the lungs and skin are also affected. Marfan syndrome does not affect intelligence.

What causes Marfan syndrome?

Marfan syndrome is caused by a change (mutation) in the FBN1 gene that tells the body how to make fibrillin-1, a protein that is an important part of connective tissue. This change creates Marfan syndrome features and causes medical problems.

What are the features of Marfan syndrome?

Marfan syndrome features occur in many different parts of the body. It is rare that a person has every feature. Some Marfan syndrome features are easy to see. Other features, such as heart problems, require special tests to find them. The common features include:

Bones and joints

- Disproportionately long arms and legs
- Tall and thin body type
- Curvature of the spine (scoliosis or kyphosis)
- Chest sinks in (pectus excavatum) or sticks out/pigeon breast (pectus carinatum)
- Long, thin fingers
- Flexible joints
- Flat feet
- High arched palate
- Teeth that are too crowded

Eyes

- Severe nearsightedness (myopia)
- Dislocated lens of the eye
- Detached retina
- Early glaucoma or cataracts

Heart and blood vessels

- Enlarged or bulging aorta, the main blood vessel that carries blood away from the heart (aortic dilation or aneurysm)
- Tear in the inner wall of the aorta that causes blood to flow between the layers of the aorta wall (aortic dissection)
- "Floppy" mitral valve (mitral valve prolapse)

Other body systems

- Stretch marks on the skin, not explained by pregnancy or weight gain/loss
- Sudden collapse of the lung (spontaneous pneumothorax)
- Widening or ballooning of the dural sac surrounding the spinal cord (dural ectasia)

If you have Marfan syndrome, it is something you are born with, although you may not notice any features until later in life. Marfan syndrome features can appear at any age—including in infants, teens, and older adults—and they can get worse as people age.

How is Marfan syndrome diagnosed?

A Marfan syndrome diagnosis can often be made after exams of several parts of the body by doctors experienced with connective tissue disorders. The evaluation includes:

- A detailed medical and family history, including information about any family member who may have the disorder or who had an early, unexplained, heart-related death
- A complete physical examination

You should also have tests to identify Marfan features that are not visible during the physical exam, including:

- Echocardiogram. This test looks at your heart, its valves, and the aorta (blood vessel that carries blood from the heart to the rest of the body).
- An eye examination, including a "slit lamp" evaluation to see if the lenses in your eyes are out of place. It is important that the doctor fully dilates the pupils before doing this test.

Genetic testing can provide helpful information in some cases.

- For individuals with a family history of Marfan syndrome, genetic testing can help confirm or rule out the diagnosis of Marfan syndrome in family members who may be at risk.
- Some of the features of Marfan syndrome can be found in disorders related to Marfan syndrome; therefore, genetic testing may be helpful when a diagnosis cannot be determined through an exam by doctors.

It is possible for you to have one or more features of Marfan syndrome, but not enough for you to have a Marfan syndrome diagnosis. You may need additional exams by other doctors and additional genetic testing to see if you have a disorder that is related to Marfan syndrome.

What should you do if you suspect Marfan syndrome?

Find a doctor who knows about Marfan syndrome. The first choice of doctor to look for is a medical geneticist (a doctor who specializes in genetic conditions such as Marfan syndrome).

A second choice is a cardiologist (heart doctor). Make sure the cardiologist has treated people who have Marfan syndrome.

To find a doctor:

- Ask your primary doctor for a referral
- Call the doctor referral service at your local hospital
- Call your insurance provider
- Call The Marfan Foundation at 800-862-7326, ext. 126

Many people with Marfan features (whether they have a diagnosis or not) need medical treatment and follow-up care. Make sure to talk with your doctor about the care that is right for you.

Elias Turner is a student at San Dieguito High School Academy in Encinitas, California. This book was written as a project for his biology class during freshman year. He was diagnosed with Marfan syndrome in preschool. His mother, uncle, and grandmother also have Marfan syndrome. He is grateful to The Marfan Foundation for their advocacy on behalf of all young people living with Marfan syndrome seeking ways to create positive, meaningful and healthy lives.