Have you heard of Marfan syndrome? Frank Ross, a 10-year-old boy, living in Saint Paul Minnesota, sure has. According to mayohealthhighlights.com, one in 5,000 people are born with Marfan syndrome. Ross is one of those one in 5,000 people.

According to the book “Marfan Does Not Mean Martian,” by Elias Clark, French doctor Antoine Marfan discovered Marfan syndrome, way back in 1896. He noticed a few similar traits with some of his patients; long thin legs and arms, and concave chests, but the cause of the symptoms was never fully explained until 1991.

Years after Dr. Antoine Marfan discovered the symptoms of Marfan syndrome, modern day science defines it as a disorder of connective tissues throughout the body. It is caused by a change in the FBN1 gene that tells the body how to make fibrillin-1, a protein that is an important part of the connective tissue in our bodies. This change creates Marfan syndrome features and causes medical problems.

Despite living with Marfan syndrome, Ross considers himself an average 10–year-old boy. He likes to take archery, participate in Cub Scouts, and read. Ross explained that some of his physical features do make him stand out from other boys his age.

“Well, I’m a lot skinnier than other people and I have a dent in my chest and I’m taller than other people.”

Ross’ mother, Missy Joy said it was the physical symptoms that prompted her to seek medical advice.

“The very first sign we ever saw was when he was just a brand new baby and Frank’s lenses in his eyes are dislocated and this causes something in his iris and the iris is the colored part of your eye, it’s not attached properly, so it trembles. It kind of looks like if you have oil and water and you can see the oil floating on top of the water. That’s the very first thing that we saw, but we didn’t connect it to Marfan syndrome until he was two, when he was diagnosed. Frank’s fingers and toes are very, very, long and they always were, even when he was little, but when they first suggested Marfan syndrome and we started looking up symptoms, that is when we realized that the fingers and the eyes and everything had something to do with it,” said Joy.

Joy did not know to look for those symptoms, because Marfan syndrome does not run in their family.

“It is hereditary, but it seems that in Frank’s case it was a random mutation that happened when he was born. We don’t have a history in our family. I guess, what I want to say is 25% of people who have Marfan syndrome are like Frank, whose parents didn’t have Marfan syndrome.”
How does Ross manage his symptoms? He explains he has scoliosis and has to wear a brace for 18 hours per day. There are also a few other precautions he needs to take daily.

“I have dislocated lenses in my eyes so I wear glasses. My aorta is enlarged so I need to take medicine for it.”

The daily responsibility of keeping Ross healthy is shared with his mother and his care is something she has to think about every single day.

“Making sure that he sees his doctors on a regular basis, he sees his cardiologist once a year. Usually he sees his orthopedic surgeon twice a year, but he’s going to have back surgery so it’s been about four times a year that he has been seeing them. He goes to the eye doctor every year. And making sure to do our best in managing things from our end, he wears a brace to help keep his back straight. He is supposed to wear the brace 18 hours a day, and it was recently changed, it used to be 20 hours a day and we needed to decrease it a bit. We’re doing our best. We’re wearing the brace as much as possible. Also, Frank takes medicine for his heart twice a day and we have to remember to make sure that he takes that,” said Joy.

When Joy was told the diagnosis was Marfan syndrome she knew she needed to educate herself on the disease.

“We wanted to find out as much as we possibly could. We were living in Houghton, Michigan at the time and the doctor that we saw suggested that Frank go to the outreach clinic at the University of Michigan. They were going to be in Marquette so we went over and saw the geneticist from the University of Michigan. Learning as much as we possibly could was very, very important,” said Joy.

Dealing with daily medications and wearing a brace for 18 hours a day isn’t easy. But it has provided life lessons.

“Resiliency. Always get back up on your feet. Things are harder for Frank, and it makes me really proud just to see him try as hard as he can, whether it’s endurance activities or we just go out for a walk. When he tries his best I’m really proud of him,” said Joy.

Ross makes his mother proud and his advice for other youth living with Marfan syndrome is simple.

“Do your best at stuff,” Ross said.

Joy also has a bit of advice for parents who have just found out that their child has Marfan syndrome.
“The best thing I have done as a parent, other than research and making sure he gets to his medical appointments, was connecting with the Marfan Foundation. Here in Minnesota, I’m actually the secretary of our Minnesota chapter of the Marfan Foundation and there are a lot of other parents who are in the group as well as adults, teens and children who have Marfan syndrome. Just having that network of people, so that if something comes up or we need a suggestion of a doctor in a particular area, we can talk to friends from the group, so that we can get their perspective on what they did in that situation. The Marfan Foundation has chapters in all regions of the country, if not every state. There are people available to meet with you and connect with you over the phone and online as well.”

Another thing Joy really recommends if someone has just found out they have Marfan syndrome is to do their best to try and attend the Marfan syndrome national conference.

“It’s a very empowering weekend and that takes place all over the country. It just happens this year it is going to be in Minnesota, at the Mayo Clinic. It’s an entire weekend that has groups that meet and you can learn about specific body areas. They have support groups for parents, and there is a kids’ program. In fact, the first time we met other kids with Marfan syndrome was at a national conference. The first one we took Frank to was when he was three and that was in Boston. The kids get to go and do different activities, they might go to the zoo or when it was in Chicago, they got to go to the Field Museum and the aquarium,” Joy said.

Joy thinks that it is critical that people recognize the symptoms of Marfan, because the earlier you are diagnosed, the better chance that they have at living a healthier life. Joy recommends visiting the Marfan Foundation online.

“They have a wonderful website, it’s marfan.org. They have a section there that is especially for young adults and kids. They have a teen section, but all of your things you could possibly want to know about Marfan syndrome are there. It’s a great foundation that really wants to support research and connecting people with medical professionals and with other people with Marfan syndrome so that you know that you have a support group, people that you can talk to who can relate with what you are going through.”

Although there is no cure for Marfan syndrome, an early diagnosis and proper medical care can help you live a healthier life and like Ross says, help you “Do your best at stuff.”

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