

**About Health TV with Jeanne Blake**  
**Marfan Syndrome**  
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JEANNE BLAKE: Welcome to *About Health TV*. I'm Jeanne Blake. On this edition of *About Health TV* we'll learn about Marfan Syndrome. Joining us is Dr. Ronald Lacro, a cardiologist and geneticist at Children's Hospital Boston, and his patient Anthony Albano. I have to admit that as a medical reporter for so many years I never learned about this, and it's been really interesting to learn that so many people are living with this. Doctor, for those of us who don't know about Marfan syndrome, please explain what it is.

DR. LACRO: Marfan syndrome is a connective tissue condition or a connective tissue disorder primarily that affects the heart, blood vessels, and skeleton. It was first recognized by a French pediatrician back in 1896, but it's been more recently that we've sort of come to know the disorder more.

JEANNE BLAKE: What's brought it more into the public awareness?

DR. LACRO: Well, part of that has been the fact that some famous people have suffered from the complications of Marfan syndrome, and so public awareness has been increasing over the last couple decades.

JEANNE BLAKE: I just have to back up just a little bit because I would like you to describe ... you say it's a disorder of the connective tissue. What is the connective tissue in our bodies?

DR. LACRO: Right. The connective tissue, basically, is the part that holds all the other parts of the body together. There are a number of connective tissue proteins and one of those proteins is called fibrillin. The fibrillin protein is the one that is abnormal in people who have Marfan syndrome.

JEANNE BLAKE: But how is it changed?

DR. LACRO: The defects of Marfan syndrome are caused by a genetic mutation in the fibrillin gene, and the result is that either the amount or the strength of the fibrillin protein is abnormal in people who have Marfan syndrome. This protein is very abundant in the bones, in the heart and blood vessels, and in the eye, which explains why those particular organs are affected most in this condition.

JEANNE BLAKE: All right. Anthony, it's called a genetic disorder, but no one in your family had it. It just sort of came out of the blue. And you were only two years old.

ANTHONY: Yes. In my case, it's a genetic mutation of the fibrillin that was affected by it, so no one in my family has it; it was never in my hereditary past. Now that I have it, I can pass it on to my kids. It's a 50-50 chance if I have children with someone that doesn't suffer from Marfan's and has a normal genetic background as far as carrying it, so I would give a 50-50 chance to my children and it will go on from there. But in my case, there was no other past history, and I started it as a mutation through the chromosomes and the genetics when my parents had me.

JEANNE BLAKE: Can you please explain how your family was brought to understand that you had this at the young age of two, which is very unusual from what I understand.

ANTHONY: It's unusual but it's definitely a good thing to be diagnosed at such a young age. At birth, my mother felt like something wasn't right, but it wasn't obvious to any medical professional what was going on. So at one time, around the age of two, I had very poor eyesight. So I went to see an ophthalmologist and he diagnosed that I had dislocated lenses, which is a characteristic of Marfan's. So once that was established, they kind of were putting pieces of the puzzle together, to put the rest of the characteristics together and get a diagnosis that it was Marfan syndrome.

JEANNE BLAKE: You were very lucky to be getting good care.

ANTHONY: Yes, definitely.

JEANNE BLAKE: But why do you think your mom thought when you were born that something wasn't right?

ANTHONY: It was almost as ... she says a maternal instinct at that point. So there wasn't anything medically proven or anything like that. It was just a gut feeling that she had, and I guess it came out that it was correct. It might have pursued her to get a little more medical advice to see if there actually was something wrong.

JEANNE BLAKE: Sure, to be a little bit more proactive. As you say, it's important that diagnosis is early, the earlier the better. Can you help us understand why, Doctor?

DR. LACRO: One of the characteristics of Marfan syndrome is that the manifestations that we see in the condition can progress over time, and so the earlier we can make a diagnosis in Marfan syndrome, the more likely it is that we can intervene and prevent serious complications.

JEANNE BLAKE: I'd like to take an opportunity ... the National Marfan Syndrome Foundation has a video that really describes very, very clearly the potentially most damaging and fatal complications. It's called aortic dissection. Why don't we just take a minute and look at this clip of video provided by the foundation and then we'll talk about it.

[VIDEO CLIP]

NARRATOR: In an aortic dissection, blood works its way into the wall of the aorta. The aorta has three layers: an inner layer called the intima, the middle layer called the media, and the outer layer called the adventitia. During an aortic dissection, blood ruptures through the intimal layer and works its way into the medial layer. Once it has worked its way into the media, it can push through the media, causing the two outer layers to separate. It can be extremely rapid, such that almost coincident with the first tear there is a rupture through to the outside of the aorta, with immediate catastrophic complications. The most common place where a person with aortic dissection will present for medical treatment is the emergency room. It is the responsibility of the emergency room physician to recognize the possibility of an aortic dissection, to diagnose an aortic dissection, and to initiate treatment.

[END CLIP]

JEANNE BLAKE: One look at that video, Dr. Lacro, and you really understand why this syndrome is potentially so dangerous. It's fatal if it's not monitored.

DR. LACRO: Exactly. And an aortic dissection is a very serious event.

JEANNE BLAKE: Obviously.

DR. LACRO: And what's predictive of the risk for aortic dissection is the size of your aorta. So the bigger your aorta is, the higher your risk for dissection.

JEANNE BLAKE: So when you talked earlier about it being progressive, does it get bigger as time goes on?

DR. LACRO: It does get bigger as you get older.

JEANNE BLAKE: And so you have to be constantly monitored?

ANTHONY: I was monitored up to a certain point in that they measure the aorta size in centimeters and then they made a judgment call at that point if you should have that surgery or not. And I've had that surgery. I had my aorta replaced at that time when it was at a certain size that it was at a dangerous level, actually before a dangerous level.

JEANNE BLAKE: And how old were you then?

ANTHONY: At the aorta time? About 10, maybe? Yeah, I was around 10 the first time I had that done.

JEANNE BLAKE: It's so interesting, too, because it can be ... for many people it's a silent syndrome. They have no idea that they have it. Can you speak to that, Doctor?

DR. LACRO: There is treatment that's available for people who are at risk for aortic dissection.

JEANNE BLAKE: Indeed, you can have a normal lifespan.

DR. LACRO: Exactly. So the trick is identifying people who have Marfan syndrome so that we can give them preventative care and prevent the serious complications. And that's where the difficulties in diagnosis come into play. Some people with Marfan syndrome tend to be tall and very flexible, and it's those sort of external clues that help us to identify people who may have an enlarged aorta.

JEANNE BLAKE: Let's go through some of those characteristics a little bit more clearly, because there's some pictures that we can show. So if you could just talk a little bit more about it. When you say flexible, what does that mean?

DR. LACRO: People with Marfan syndrome are very flexible or hypermobile. They can, actually Anthony can probably show that their arms are very flexible, and so it's recognizing these relatively minor things about the skeleton that help us to identify people who may have Marfan syndrome.

JEANNE BLAKE: But it seems to me that that's such a ... I don't know, maybe ... not a lot of people can do that, and maybe I would wonder why someone wouldn't wonder why a person was able to do that. What do you hear when people come to you at a later age? That they just thought they were super-flexible?

DR. LACRO: I mean, a lot of people are flexible, gymnasts and athletes are flexible. So it's generally a combination of things. People who are tall and flexible, people who are tall and have eye problems and are flexible. The more of those external features that you have, the higher your suspicion that that person may have Marfan syndrome.

JEANNE BLAKE: I was really shocked to hear that the playwright Jonathan Larson, who wrote the very famous Tony Award-winning play *Rent*, died ... I remember when Jonathan Larson died just before *Rent* opened, but I was really surprised to find out just as I was preparing for this program that he died from aortic dissection. And so he had Marfan syndrome. And so thus it can be a silent killer.

DR. LACRO: Right. The tricky thing about Marfan syndrome is that although we have sort of a classic picture in our mind as to what a person with Marfan syndrome should look like, everyone with the condition looks different. And so people like Jonathan Larson can live normal lives not knowing that they are at risk for the complications associated with Marfan.

JEANNE BLAKE: Anthony, though you found out when you were two years old that you had this syndrome, it certainly has not gotten in the way of your life.

ANTHONY: Not at all, no. It's become my life. I live my life around Marfan syndrome. It's been who I am. And that's the thing about awareness, is getting it at a young age so you can develop your life around it instead of changing your life at a certain point. If you're diagnosed in your 20s, 30s, 40s, your life is changing from that point on. In my case, my life started with it, so I've lived the rest of my life knowing what's going on.

JEANNE BLAKE: Just tell me how you mean that, how it defines who you are and the way that you live your life. How do you think that you live your life any differently than another 23-year-old?

ANTHONY: It would have to be more of all the experience I've gone through as far as surgeries, medical procedures, just living in society with different physical characteristics and just developing things that I'm in. As far as contact sports and things, when you're an adolescent those are a big part of your life. So when I was younger those things were out of the question, so I replaced them with music, which was my alternative as far as contact sports, and I lived in the inner city, so a lot of kids are playing sports, a lot of kids are playing hockey. I became, I call myself a professional spectator. I like to watch those, and then I participate in music.

JEANNE BLAKE: Well, you're an accomplished musician, and I just think it's only fair that we just take a minute to listen to your music, because you brought a tape of it in. Here's Anthony as part of Stock 7, right?

ANTHONY: Yes.

[AUDIOTAPE]

JEANNE BLAKE: Bravo. And where do you play?

ANTHONY: Right now we play around any venue in Boston that we get a show at, so if there's any booking agents, call me and book a show. Well, we play downtown, at any venue downtown in Boston.

JEANNE BLAKE: Good for you. Good for you. And you started playing the guitar when you were how old?

ANTHONY: I was around 12 years old when I started playing the guitar.

JEANNE BLAKE: That's such a critical age, Anthony, developmentally, and I'm wondering what it was like for you as a child. Did you get teased because of your Marfan syndrome at all?

ANTHONY: I was probably prejudged more than teased, and then once people actually got a chance to meet me and see who I was exactly than by what I looked like, it wasn't really teasing. I really never had to put up with that. I was fortunate for it.

JEANNE BLAKE: And how did you feel prejudged though? I'm wondering.

ANTHONY: It just made me have to prove myself.

JEANNE BLAKE: Well, how did you know that? Did you sense that?

ANTHONY: You can sense that by some people's arrogance and by some people's ... a lot of staring, a lot of types of things like that. But I became used to it. It actually bothered people around me more than it actually bothered me.

JEANNE BLAKE: Your mom and dad did a really good job raising you. Your confidence is so high. Can you tell me the kinds of messages they gave you from an early age? Were you warned that you might be treated differently?

ANTHONY: They actually let me live through it as I wanted to. They would let me set the tone. That was what actually gave me the opportunity to be who I am. They didn't try any real regimental things. As far as health issues, yeah, but I could do what I felt I could do and they encouraged me to do what I wanted to do. So it was never really an issue as far as them kind of putting down, you're sick so you can't do this, you can't do that. I got the medical education from Dr. Lacro, who set the record straight of what I could do and what I couldn't do, and then it just came that I developed through the knowledge I had about Marfan syndrome

JEANNE BLAKE: But what kinds of ... did your mom ever warn you that when you go to school somebody might say something to you? Do you remember?

ANTHONY: Not as a warning, no. I experienced it for myself. They wanted me to go as who I was and not be kind of defensive of what people are going to think.

JEANNE BLAKE: That's great.

ANTHONY: Because if they were going to warn me, it would be making me try to prepare and try to judge if people were actually trying to judge me. But they never set that in my head at all.

JEANNE BLAKE: They did a great job.

ANTHONY: Yeah, definitely.

JEANNE BLAKE: Now, I know that we've talked about how you live your life, you know, with your band and all, but I'm wondering what this syndrome means for you medically. I mean, how often do you go to the doctor, and what else do you have to do medically to take care of yourself that another 23-year-old might not have to do?

ANTHONY: Well, we have consistent monitoring of my aorta, since I've had past surgeries on it. It can still increase in size.

JEANNE BLAKE: What do you mean by consistent, Anthony?

ANTHONY: Six months. I go every six months now to see Dr. Lacro. We do an echocardiogram, EKG, x-rays, just to monitor everything that's going on. I've had several past cardiac surgeries, but we just monitor and keep track of ... I'm on different medications, there's about four different prescriptions you get, that regulate my blood because of the artificial valves I've had closed. Just like that, it's maintaining now of what we've already done as far as repairs.

JEANNE BLAKE: Dr. Lacro, what hope is there on the horizon for people living with Marfan syndrome? Anthony, you've mastered this and you sound like you're doing very well. But I imagine that there's research in this field that is promising.

DR. LACRO: Right. The exciting research probably started about 10 years ago when the gene was first identified. So now we know which gene actually causes the condition. And there's exciting work in Baltimore at Johns Hopkins University. And they're looking at how the fibrillin gene functions, particularly in a mouse model. And it turns out that the fibrillin protein is not only important in terms of the strength of our connective tissue, but also it's involved in the regulation of how different parts of the cell communicate with each other. And they've actually been able to control the function of the fibrillin gene in the mouse model. So it gives us some hope for a specific treatment for people with Marfan.

JEANNE BLAKE: And what would that be? What would that treatment be?

DR. LACRO: Well, the specific research that I was referring to is the fact that fibrillin helps control this protein called TGF-beta. It allows the levels of TGF-beta to be too high. And by controlling the TGF-beta levels, they can actually reverse some of the findings in the mouse model. So the theory

is that if we can control TGF-beta in people who have Marfan syndrome, we may be able to slow the growth of the aorta and prevent dissections and allow people to live longer and more successful lives.

JEANNE BLAKE: How many years down the road, Doctor, do you think there will be a treatment available?

DR. LACRO: I would predict that within the next 5 to 10 years there should be effective treatment.

JEANNE BLAKE: Oh, wow. That's a lot sooner than even I would have anticipated. You know, one thing that strikes me, talking about Jonathan Larson and then talking with Anthony, is the variability with people who have it. Of course that contributes to making it so difficult to treat, but why do you think that's so?

DR. LACRO: It's a part of the syndrome that the syndrome not only affects many different parts of the body, but the expression in each person is different, which makes it harder sometimes to identify people, and so I think that the clues that we need to look for are things like tall stature and the usual problems and flexibility, curvature of the spine. These will be things that can help us move toward Marfan.

JEANNE BLAKE: There's exciting developments. We do have viewers from other cities around the country, but here in New England people will have a special clinic to go to, and I know that you're not going to toot your own horn, but you've started a clinic that's very important. I'd like you to talk about it just for a minute.

DR. LACRO: I think since the care of people with Marfan syndrome is so specialized, and experience with Marfan syndrome is really helpful in caring for people who have it, and I think Anthony can give some evidence of that. So we at Children's have been providing care for kids with Marfan syndrome for several years now, and this year we're going to expand our services to start a greater New England Marfan syndrome program which will serve not only children with Marfan syndrome, but young adults and older adults.

JEANNE BLAKE: That's great. Anthony, any final words?

ANTHONY: This clinic is a big thing. Even in my state of life right now, I've always been seen at the Children's Hospital as a child. Now that I'm at 23, going into adulthood, it's a good transition to have more care towards people my age.

JEANNE BLAKE: So even as a 23-year-old, you'll still be able to go to Children's Hospital in Boston?

ANTHONY: Yes, which is easier. I have a lot of specialists at Children's as far as orthopedics, ophthalmologists, cardiology, and it's a multi-system disorder, which this Marfan clinic kind of put in one place so you're not chasing your doctors around.

JEANNE BLAKE: Enough of that, right? You've got to get out there and play your guitar. I want to thank you both for coming in. This has been so interesting to learn more about this, and I wish you continued good health, and I know all our viewers will be wanting to wish you all the best. And Doctor, good luck with your clinic and many people will be well served by that. Thanks so much. And we'd like to thank you for joining us on *About Health* TV. And I'll see you next time. I'm Jeanne Blake.

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