Marfan Surgery

Dr. Paul Friedman: Hello, my name is Paul Friedman. I'm chair of the Department of Cardiovascular Medicine, and I'm delighted to have with me my colleague, Dr. Alberto Pochettino who is a professor of surgery and an expert in aorta surgery. And today we're gonna talk about Marfan syndrome and specifically its impact on aorta. Alberto, Dr. Pochettino thank you for joining me.

Dr. Alberto Pochettino: It's a pleasure.

Dr. Paul Friedman: Why don't we start with the just basics. What is Marfan Syndrome and why do you as a surgeon get involved?

Dr. Alberto Pochettino: So, Marfan Syndrome, we note now it's a genetic abnormality that affects the specific protein called fibrillin. The description of Marfan dates way before we knew about the specific genetics. And it has to do with a, a phenotypic appearance, phenotypic meaning what people look like. And it had to do with hyper extensibility of, of digits and joints. It had to do with a high incidence of skeletal abnormality like scoliosis, like pectus. It had to do with eye abnormality. One of the classic one is dead lens dislocation, although other, like higher risk of retinal detachment compared to the general population. And of course, the last one that I left is aortic abnormality. That's where, where I get involved. So what of this as a common denominator have to do with abnormality of collagen? Obviously collagen has to do with, with what holds the lens. For example, in the eye, collagen has to do with our joints and how tendons work. And so that's where the skeletal muscle abnormality come in. And of course, collagen is a very important factor in how the aorta is put together. So when there, there is this genetic abnormality, all of the structures can become abnormal. And the issue is, while many of the other abnormality are more of a quality of life issues and things that need to be addressed, aortic abnormality can be life-threatening. If the aorta sort of dissects and ruptures, it's, it's a much more life-threatening than some of the other features of Marfan.

Dr. Paul Friedman: So first, which patients with Marfan syndrome should be evaluated for aortic disease and how is that evaluation done?

Dr. Alberto Pochettino: So in general, anybody who is identified as having Marfan should have evaluation of their aorta. Now, often the, the way that the individual looks like or some of the other skeletal abnormality come up first, it is not unusual for somebody in their teens to come in to be presented with scoliosis. And, and the pattern may be such that one may suspect that there is Marfan. And today it can be checked very quickly with genetic testing. And of course, once the genetic testing is performed and it points to Marfan, I think any individual that has that genetic abnormality should have at least a screening test. And screening comes down to echocardiogram.

While all vessels can be affected in Marfan syndrome, the classic presentation and what is the majority of the issue has to do with the aortic root and the ascending aorta. And those can be imaged very well with an echocardiogram. If they're abnormal, then you can go to a CT scan or an MRI, which should be a, you know, a, a much more accurate and and precise ways to, to assess. But the starting point, I think it's, it's an echocardiogram.

Dr. Paul Friedman: Do all marfan patients get aortic abnormalities? How often do you recommend we screen?

Dr. Alberto Pochettino: So that's a good point because sometime patient will come in and, you know, they're afraid. It, it is not true that all marfan develop aortic abnormality. So, so it is, it is a, a false to say that you're gonna develop in your lifetime no matter what. It is common and it can be life threatening and therefore it, anybody who has that genetic abnormal body should be screened. And if their aorta is normal, then, then obviously the screening can be relaxed as, as much as one, you know, clinically feels appropriate. But it shouldn't be completely ignored. Just be, if you have screened it once, you shouldn't just say you're done. That's it for the rest of your life, you should keep an eye on it. Whether it's once a year, every other year, every four years, that has to be depending on the specific situation of the individual. I think that kind of screening should be continued. But many patients do not develop aortic disease. And if they don't, that's obviously that's great for them. But, but you don't wanna miss it if it develops later in life. And I've seen patient in their late fifties, early sixties who first developed an aortic root dilatation and lo and behold, they have had marfan all along, but phenotypically it didn't present until later in life.

Dr. Paul Friedman: Now, obviously, since I'm speaking with a surgeon, I want to ask you about the surgeries that are available to prevent bad things from happening to patients with these high risk. When, when do you operate on the aorta in patients with marfan disease and what preventive operations are available?

Dr. Alberto Pochettino: So that brings back to dilated root and an abnormal aortic wall. What is the risk of of having that? So the primary risk comes down to aortic dissection. So aortic dissection are generally divided in two types, type A and type B. Type A is by far the more common and it, the risk of type A dissection in a Marfan population is much higher than the general population and it does correlate to a great degree to the size of the order. So what we do in screening is to look at the aortic root and look at this in aorta. And when it's above a certain size, then that would generate a referral for preventive surgery. Now, what is preventive surgery? Type A dissection is a tear in the intima of the ascending aorta, usually right above the root, you know, in the area of the center tubular junction as we call it. So preventive surgery means removing that aorta at risk of dissection, essentially the root and the ascending aorta, that's what preventive surgery comes down to. So about 90% of all dissection in anybody but especially Marfan, are type A dissection. So by removing the aorta from the valve area, from the root to the beginning of the arch, you have removed

essentially 90% of the risk of dissection. So what kind of surgery that would be? That would be a root replacement and an ascending replacement. The issue then comes what to do with the valve. You know, obviously the root is the part of the order that contains the valve and the takeoff of the coronary arteries. So traditionally until the late eighties, the only root replacement was one that required valve replacement with it. And you know, early on, you know, again, in the, in the mid to late eighties, people started to look at Marfan patient that came in for elective surgery and, and often noted that the valve looked pretty good, so why should we throw it away? So various surgeon developed methods to retain the aortic valve while replacing the aortic root around it. And generally speaking, those operation are called valve sparing root replacement. So when the valve functions and when you're looking at a surgery, it appears normal. The goal of the preventive surgery is to keep the valve in place and replace the aortic wall from root to the, to distal ascending aorta. So that's preventive operation.

Dr. Paul Friedman: What's the patient experience with that? How long are they typically in the hospital? Are there, you know, various strategies and surgical approaches for managing an enlarged ascending aorta in a patient with marfans?

Dr. Alberto Pochettino: So the results of that operation root replacement and ascending replacement have become quite good. Meaning the mortality are in the range of 1% in most who are otherwise healthy immunity. They have no coronary disease, they have had no strokes. Typically, they tend to be relatively young, although we have done that operation in even in patient in their seventies. And the results are not that different even in, in, in that patient population. So typically the patient has a hospital stay that is less than a week, you know, five to seven days or the usual range. It is a big operation, but, but with the modern technique, you know, you can do it within, you know, four to six hours is the time that it takes to do the operation. And, and typically the, the ICU stay is less than 24 hours, typically overnight. And the next day most patients are moved to a regular floor where they can recover, get out of bed and move around. And once various surgical issues like tubes and wires and all that are out, they can leave. Now if, if the valve has to be replaced, it doesn't really add that much more to either the surgery or the recovery. So the, the, the morbidity and mortality profile of whether you spare the valve or not, it's really not that different. Of course if you replace the valve then you have to deal with the type of valve you put in. And if it's a mechanical valve, that means, you know, anticoagulation, which often delays going home by a day or two because you have to achieve a reasonable INR, you know, warfarin being the primary anticoagulation for mechanical valves.

Dr. Paul Friedman: Now if you're either a primary care doctor or a cardiologist and you're seeing a patient in follow-up after surgery, specific follow up tips, tricks, observations, things to look for in the patient who's now had the ascending aorta replaced.

Dr. Alberto Pochettino: I mean, typically if the valve is, was spared, you certainly wanna make sure that, you know, cardiac function has remained normal and you wanna follow that valve. I mean, it's a native valve and you wanna make sure that it remains in good shape. So I would say, I mean, early on you wanna, you wanna make sure that some of the technical issue that occurs after any major operation don't, you know, are not in there. For example, pericardial effusion, obviously any wound abnormality and, and, and some of the lung related sort of elects and pleural effusion, all of those things are early on. But once you're out of the early phase, you're just looking for cardiac function overall and specifically aortic valve function. One of the things that people have noted is that it turns out the leaflet themself contain collagen. So while we have taken out the aorta, that was a risk of, of, of dissection we have left the valve and there is debate even among surgeon, you know, how long would the valve sparing operation last. Given that the collagen within those leaflets is still abnormal. I mean most patients and most of the data suggests that do very well for long periods of time. You know, the valve sparing operation has been around for about 20 years. So within that 20 year observation, I would say 80 to 90% of patients will remain with a good functioning valve. But there are exceptions. So, and some of those valves that were perfect when you, when you spare it 10 years later, may develop marfan related abnormality in the leaflet and tears and so forth. And that needs to be kind of addressed if, if it's severe enough that it requires additional intervention.

Dr. Paul Friedman: Now we've really been focusing on type A dissections, ascending aorta, dissections 'cause they're the most common. But obviously as, as you've just pointed out, collagen is abnormal everywhere. What about the descending aorta? Are there risks of type B dissections?

Dr. Alberto Pochettino: Definitely one of the frustrating things sometime in patient with that a preventive operation is if they develop a type B dissection five to 10 years later. And, and obviously, well we had this big operation, you know, I thought I was fine and, and now and now I'm not. And so unfortunately again, it's, it's not as common, but it can happen. Now the the unique thing about type B dissection is that most of the time in the general population are related to hypertension. So your classic 70, 70-year-old hypertensive patient developed type B dissection. The usual treatment is medical therapy, meaning blood pressure control and then follow up. And in general, the rule of thumb is that in a non marfan patient, about one third of the patient that developed type B dissection will go on to develop what's called a dissecting aneurysm. So an aneurysm in the substrate of dissection. Now in Marfan unfortunately that percentage is much, much higher. In fact depends, it's mostly age related. But if the, if, if a marfan patient is young enough, almost certainly will develop a dissecting thora abdominal aneurysm, which is a big deal because those are very extensive dissecting aneurysm that go from the arch down to the iliac often. And, and if they're large enough then they require pretty com complicated and extensive operation of replacement. So, so it it, it does, even though it's rare, it does affect the patient and often will require in their lifetime, typically within months, often, especially if they're quite young. Major operation. Now in a, in a standard non marfan patient stent grafting is used extensively to treat type B dissection. Unfortunately, stent grafting has been shown not to be a good idea in Marfan patient because the principle of a stent graft is you landed into a normal aorta as a landing zone to exclude the, the aneurysm. But there is no normal aorta in a Marfan patient. So if you landed it in any part of the

marfan aorta, that marfan aorta under radial force will degenerate and then develop an, an aneurysm at the landing zone. So there are issues, you know, and, and often an open operation is the only option in, in that Marfan population.

Dr. Paul Friedman: Do you see any changes in the future for the surgery for patients with Marfan disease and aortopathy?

Dr. Alberto Pochettino: Well, what has happened over the years is that our ability to address aorta early, you know, preventively has gotten much better. I mean I've been doing this for 25 plus years and, and certainly the mortality that we saw when I started during my training was much, much higher than it is today. So we can afford to intervene earlier because our results are better now because not everybody develops aortic disease. You know, the questions has been raised, you know, should we do something in every Marfan to prevent developer than aneurysm? And the problem is we just don't know which one will develop, although it's common. So there are other more experimental sort of issues that have been looked at, like wrapping the aorta early on before any dilatation has occurred. And, and that at this point is, is a little bit a stretch given that while common it's not a universal issue, that, that Marfan will develop aortic original disease. Now stent grafts, you know, a lot of companies are trying to come up with stent grafts that would be friendly to, to marfan it, it really hasn't happened. Typically what happens if a Marfan patient is at some replacement of the aorta with some background in place. So some of the patient have operated doing a preventive operation, they have Dacron in place so that you can use Dacron in your landing zone and then you can span the aortic, the, you know, the abnormal aorta in the Marfan. So there are, there are things that can be done, but there hasn't been a, a sort of se seismic change in, in our approach to, to, to, to allow different technology to be used. I'm sure people will continue to look at it.

Dr. Paul Friedman: Well, fascinating area and really a difficult, challenging, complex disease we've learned so much and continue to learn more. Dr. Pochettino, thank you for joining me.

Dr. Alberto Pochettino: You're very welcome. It's a pleasure.