

About Health TV with Jeanne Blake
Scleroderma
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JEANNE BLAKE: Welcome. Dr. Massarotti, let's start with you. What is scleroderma?

DR. MASSAROTTI: Scleroderma is a rheumatic disease. It's considered to be an autoimmune disease, primarily affecting women, and usually young women. It's characterized by thickening of the skin, among other features. The skin, however, isn't the only organ involved, though. Other organs can be involved – the esophagus, heart, lungs, and kidneys. It's a progressive disease in general.

JEANNE BLAKE: It's serious, and can be life-threatening.

DR. MASSAROTTI: That's right. Not everyone with scleroderma will have those features. At the very beginning of the disease it may be difficult to sort out whether one has scleroderma. Most patients with scleroderma will have Reynaud's phenomenon, where their fingers and toes become discolored when exposed to the cold. That an extremely common phenomenon. But the progression to scleroderma from Reynaud's is very rare. In Liz's case, she began with features of scleroderma, but then, I will let her tell you, started out with something in the hands, and over the years her skin has become much more thickened, much thicker.

JEANNE BLAKE: Do we know its cause?

DR. MASSAROTTI: Like most rheumatoid disease, the cause of scleroderma is not known. There is research being done every day to identify what might cause it. We know there are disorders in the environment that mimic scleroderma, and from those mimickers we might be able get an idea of what causes it. But there's a trigger. It may be a contaminant of some sort that could be in the environment or elsewhere that triggers this in this process.

JEANNE BLAKE: In people who are vulnerable to that.

DR. MASSAROTTI: In people who might be genetically susceptible. In some diseases there is a specific genetics susceptibility that has been identified. For example, rheumatoid arthritis. In scleroderma, the specific genetics susceptibility is not known and has not been found.

JEANNE BLAKE: Okay, last question before we get into the specifics.

DR. MASSAROTTI: Sure.

JEANNE BLAKE: Rheumatic. Help us understand. When you say it's a rheumatic ... it's in the rheumatic family, what does that mean?

DR. MASSAROTTI: A rheumatic disease is a disease that directly or indirectly affects the joints. So examples of rheumatic diseases might be rheumatoid arthritis, lupus is another rheumatic disease. One might say they have rheumatological pain.

JEANNE BLAKE: But this is so much more than just the joints. It's the skin and it's all the organs, so I'm confused by that.

DR. MASSAROTTI: So rheumatic doesn't mean that it's just confined to the joints. It means that it evolved in the joints in some way.

JEANNE BLAKE: I gotcha. Great. Now, Liz, you learned in 1996 that you had scleroderma. What were your early symptoms? You were very young. How old were you at the time?

LIZ: I was 33.

JEANNE BLAKE: That's early for someone who is diagnosed with this. It runs from 35 to 55, right?

LIZ: I didn't have a long list of symptoms. Mine came on pretty rapidly. I was working downtown as a waitress at the Copley Plaza and had such difficulty with simple things, opening wine, champagne, picking up silverware, doing simple things at home. I had a lot of other things, such as some fatigue, some body aches, and it got pretty pronounced, and I thought maybe my age, maybe I have to go to a therapist. I had my own ideas of what might be going on. I had five children at home, I thought I was tired.

JEANNE BLAKE: It's been described as sort of like the pain ...

LIZ: That one I haven't heard, Jeannie, but I've said it's like being a size 4 in a size 2 skin. I feel like I'm shrink-wrapped. I can't describe it any other way. I'm tight all around. I have tons of collagen and

that just makes my skin as hard as anything. So I went to my primary care doctor and had some blood work done. It came back abnormal, and when that happened, that was the first step, and they explained that it pointed to something, possibly an autoimmune disorder, something in that direction. My doctor is a fine physician and didn't know it was scleroderma, and he suggested I see a dermatologist for skin biopsies. I went back two days later and there was a dermatologist there and he looked at my hands and said to me, "I'm sorry, you don't need a skin biopsy. This is scleroderma." I looked at him.

JEANNE BLAKE: That's amazing, because it can be – Dr. Massarotti, it can be difficult to sometimes diagnose, and I think that Liz is lucky in a way to learn early on.

DR. MASSAROTTI: That's true. Although I agree that you're lucky, because knowing helps you to define your life better, and uncertainty can make people feel very anxious. It's largely a disease that you treat symptomatically, you treat the symptoms.

JEANNE BLAKE: So the unlucky part is that not a lot can be done?

DR. MASSAROTTI: Right, regardless of when one diagnoses it, except for some features of the disease, which are very treatable. The hypertension and high blood pressure that can be seen with it can be a sign of kidney disease, and that's very treatable and very important to treat. So that's important to recognize early on.

JEANNE BLAKE: When you told people that you had this disease, you started over the last few years to learn about more in your community, and that's the reference to the disease. Tell us a little about that.

LIZ: What do you mean? I was diagnosed. I got the diagnosis of scleroderma immediately before a rheumatologist, because that's who oversees my care and that's who oversees everything and takes care of me. But breaking the news to family and friends is really how I found out about the others. Grapevine, word-of-mouth gossip, whatever you want to call it, South Boston is a tight community. Just with the diagnosis and going in and resigning from work, for instance. One of the other women I work with said, "My cousin has scleroderma," and I asked her if she was sure, and she said she was sure. She's had it since, I think she said 1989. I kind of filed it away and didn't know what to make of this, and then within days my cousin told me her best friend's mother had scleroderma. This is the second person within just a very short period of time. Both are South Boston residents with the same disease.

I kept an unofficial list and slowly three more were brought to my attention. I found out the last woman lived four doors away from me. I just couldn't believe it, and that's what prompted me to talk with Dr. Massarotti and she, of course, because of patient confidentiality, couldn't share a lot of information, couldn't share any information with me. These are her patients. But she encouraged me right away to go for it, and I contacted a few different agencies and finally got the right one. It took some time, but I got Suzanne Condon at the Department of Public Health, put in writing what my fears were, whether I thought there was a high rate in my neighborhood, and would they be willing to come take a look.

JEANNE BLAKE: Okay, hold on. I just want the doctor to put this into some perspective. This is statistically high for a neighborhood like South Boston?

DR. MASSAROTTI: True. The number of cases that have been preliminarily identified as having scleroderma, or having features of scleroderma, are somewhere around 26. I believe the South Boston population is about 35,000 to 40,000. That's much higher, about seven times higher than ...

JEANNE BLAKE: Seven times higher than it would be in the general population.

DR. MASSAROTTI: Exactly. So it does raise your eyebrows.

JEANNE BLAKE: So the Department of Public Health did respond to your letter and they are currently investigating the potential of a cluster of scleroderma in South Boston?

LIZ: They contacted all the major medical facilities in Boston, for the medical world we're so blessed. Dr. Massarotti called right back from all the different hospitals, Boston officials, community health officials, South Boston Community Health Center and Health Initiatives, and some of the other people who happened to be sick. And they meet with us every so often to let us know where they are. But we worked together to get where we are now.

JEANNE BLAKE: Dr. Massarotti, how does one begin – I'm familiar with clusters of breast cancer, I just investigated on it, and it's really, really difficult to determine an environmental cause. Take us inside this kind of investigation to the degree that you can and tell us what the process is and what the hope might be that a cause would be isolated that's environmental.

DR. MASSAROTTI: Sure. I think this work is patient-driven and I think that's one of the most remarkable things. Liz really recognized something, and I think many other diseases that have been

identified in clusters and then triggered, the patient recognizes somebody who is very motivated to find out why. And because scleroderma is something whose cause is unknown, the motivation to try to find the cause is sometimes even greater. There's always the question Why me? or Why did this happen? The first point is that there were a large number of cases within a geographically confined area, i.e. 26 patients in about a 35,000 or 40,000 population, which is more than one would expect based on what the incidence of scleroderma would be. So the next step is to find out if those 26 patients actually have scleroderma, because they're seen at different institutions, and all of us have had experience saying that my doctor said I had this particular disease and they go to another doctor and this doctor said it's something different. So the first step is to confirm that the diagnosis is scleroderma in the 26 patients, and that's coming along. The confirmation requires being seen by physicians at Boston Medical Center, Dr. Korn's group and our group, Dr. Kalish and myself. So that's one part. The second part is then, in addition to the examination, asking a very detailed history of each patient about their history, their family history, their occupational history, their exposure history, finding out as many details as possible about all aspects of their lives.

JEANNE BLAKE: And then do you look for other links within it? I mean, 26, is that a small group to consider for a cluster? You'd rather – and this sounds a little bit odd to say – but you'd rather be working with 100, you'd be more likely to see other factors emerge that might make the cause ...

DR. MASSAROTTI: Right. The larger the number, the more meaningful the percentages in those numbers will be.

JEANNE BLAKE: Right.

DR. MASSAROTTI: And the fact that you have 26, you'll be able to learn some things, hopefully, that will be helpful to the larger community. But you may not be able to learn as much as one would think. So that's one piece, speaking to the patients, doing the questionnaire, and examining all of these particular areas, and then evaluating what is called a control population. And in a control population, it's finding out if other people who live in South Boston had similar exposures why they didn't get scleroderma. So the control population is very, very important.

JEANNE BLAKE: This is not an area that I've done a lot of work on. Do they do soil tests and water tests in homes of all the people who are living with scleroderma?

DR. MASSAROTTI: At this point, no. At this point, it's really at the investigative phase.

JEANNE BLAKE: But eventually, in looking at environmental factors, that all will be tested, I would think.

DR. MASSAROTTI: It's not clear what's going to be tested right now. I think you sort of have to see what the landscape is, so to speak, no pun intended. I mean, I think the natural inclination for any one of us living in a particular area when we hear something happen is that it's something we've been exposed to, the water, the electricity, the oil, the soil, but I think careful investigation and thoughtful investigations are important. You need to kind of go step by step.

JEANNE BLAKE: Well, that's why I'm not an epidemiologist. I just don't have the patience for it. But I think the people of South Boston do need to recognize that it could be a coincidence. They do occur and there can be clusters that are driven by coincidence, right?

DR. MASSAROTTI: True. If I'm not mistaken, I think many of the clusters that have been identified have turned out to be coincidences.

JEANNE BLAKE: All right. That was actually very interesting education. Thank you, Doctor. Liz, you mentioned earlier that you have five children, ages 18 to 10.

LIZ: Eighteen, to the youngest, who is ten.

JEANNE BLAKE: How are you with talking to them about what you're living with?

LIZ: I have six years under my belt with this disease, but in the beginning it was a very frightening time for me. They were very emotional, and I felt a mini-meltdown because I couldn't share any of the fear I had. There was a tremendous amount of fear I had, and I felt my husband and my kids didn't need that. I grew up with a mom who had Hodgkin's disease, and I remember being a young kid watching my mother being very sick and in and out of the hospital, which I haven't been, dealing with the Hodgkin's disease and how frightened I was as a child. And here I was many years later going through the same thing, almost, as a parent, and my children having to deal with this. I'm trying to spare them with as little information as possible, trying not to frighten anyone. I'm tending to focus on Dr. Massarotti to bounce everything off of.

JEANNE BLAKE: I want to go back to this, but your children knew something was up.

LIZ: Oh, sure they did. They saw me.

JEANNE BLAKE: I wonder what the message was that they picked up that there was something going on, but they didn't know what it was.

LIZ: They knew. First of all, having to leave my job, a working-class family – my husband's a fireman – and with me working as a waitress and having to leave that job. Then they saw the changes in me. There were big changes, and I'm talking about even not so much in appearance, but having to do for myself. It's like living with hockey gloves on. So we're talking six years, and Paulie was almost 4 years old – my youngest I'm talking about – when I was diagnosed. And I'm asking young children to help me button my shirt.

JEANNE BLAKE: Let's talk about your oldest child. Do you remember anything that one of the children said, Mom, what's going on?

LIZ: I remember them asking me was I going to be okay. I remember one of them asking if I was going to die, and I was just trying to reassure them that they would be okay. No matter what happens, they were going to be okay. They had to learn to deal with what has happened to me, and I think I purposely kept information from them. Dr. Massarotti even met with them, and she assured them that right now there is no vital organ involvement, in her own way talking to children, and that there's probably a good chance there won't be, but they have seen the changes.

JEANNE BLAKE: And they do understand that there's a potential that something bad could happen to their mom.

LIZ: Yes, but it's almost like ... not that I'm in denial about it, Jeanne, because I'm not, but it's never spoken about. I don't know if that's a good thing or a bad thing.

JEANNE BLAKE: What do you think, Dr. Massarotti? Take care of people who are living with varying degrees of this disease, what's your advice on how to talk within the family when a person has any kind of a serious, incapacitating disease?

DR. MASSAROTTI: Well, I think to be open and honest and compassionate and to be respectful of the patient and where they are in their lives and with their families and kind of put yourself in their shoes. And to remember what the patient needs to know for themselves at that time. You're trained in medical school and in training to help people with these particular issues. One never lies, they're being very honest and helpful and you have to be there as much as you can for them.

JEANNE BLAKE: When there are children involved, it's a conflict, I think, for every parent to know how much to tell their child. And if a parent has a potentially life-threatening disease, and Liz's is potentially life-threatening, the chances are, as you described earlier, that she will live a long life, maybe with some restrictions, very likely with more restrictions as her skin gets tighter. If she were to not survive the disease, do children carry into their lives this notion that they weren't told the truth?

DR. MASSAROTTI: I'm sure that's happened. I'd like to think that that's not happened with any patient that I've treated. I think the same rules apply to explaining things to a child as they do to an adult. I think children tend to look at things as black and white when they're very, very young. As they get older, they tend to have more cognitive understanding and are more emotionally developed to handle the nuances of a life where things aren't so black and white. But I think not being honest and not answering the questions in a way they can understand at that time will do the child a disservice. And sometimes professional persons, child psychiatrists or child psychologists, may be needed to kind of help the child understand the home situation and illness in a parent, especially if there are signs that the child might not be functioning well at school or at home or regressing in some other way.

JEANNE BLAKE: Liz, how do you think ... I always believe that when a family is handed something to deal with like your family is dealing with, that there is a silver lining and that there are benefits to the family if it's handled well. Do you find in a way that you're closer or that your communication is more open or that you as a family unit deal with things in a different way that's positive, that's been a positive outcome?

LIZ: I think there have been some positive aspects to having scleroderma. I can't believe I'm saying that, but I do. It doesn't change the family dynamic. In my family, if anything, it's helped them to understand that they can depend on one another more, and I don't know if it's a good thing, but they're growing up faster and they're beginning to take more responsibility. Sometimes there will be days that they're not taking care of me, but there will be days that I'm not feeling well and it's going to be up to them. My husband is there, but these kids are dealing with different issues than the children of my friend, than the children of my neighbor, so the good part is that we're going to deal with it together and

we're going to get through it together. Whatever will come will be explained; Dr. Massarotti will keep us informed. I think they understand, too, that this is a neighborhood issue at this point, that there are other people sick besides Mom and we need to find out ... they need to focus on that, too. Sometimes that takes a lot of the fear, even for myself, knowing you can focus on someone else, that there's someone else going through this also. I don't know if that answers your question, but it helps us deal with it.

JEANNE BLAKE: It sure does. And on days when you say that you're not feeling well, what are some of the things that you're feeling, being tired?

LIZ: Very rarely am I tired. I don't have a lot of days of pure fatigue. I have dreadful heartburn, but my doctor prescribed something for it, everything from eating to digestion, everything to do with food. And I love food, food is a sign of love in many families. You love to prepare it and serve it and show off and eat. I've lost all joy for that. That's a biggie. Everything from a July 4 barbecue to a night out with your husband in the North End or the children. That's gone. There are things that I've had to almost say goodbye to. They're a part of my past. It's not a part of my life right now. There are different things. There's pain. There's everyday pain, and Dr. Massarotti might be able to describe what it is. I think it's because my skin is hard and it's pulling on my joints and my fingers. It burns sometimes, it's been tight sometimes, it hurts. The joints right here feel like sometimes they're going pop out. I don't know how medical that is, but this is a painful disease.

JEANNE BLAKE: Well, it looks painful. Your hands look as though they hurt, and your description, articulate description of having size 2 skin in a size 4 body really does describe it. Dr. Massarotti, where on the national level is the medical community in terms of research?

DR. MASSAROTTI: Research is examining a number of different aspects of the cause of scleroderma and what we call the clinical manifestations. For instance, kidney disease was a major cause of death up until maybe 15 years ago and now lung disease is a major cause of death in the most severe cases of scleroderma. So the clinical aspects have been well-defined. Research is now focused on trying to define products that might allow the skin to be looser. Liz was involved in one of the clinical trials at Boston Medical Center for a particular type of therapy to try to loosen the skin. So there are clinical trials of specific medication to try to see if one can loosen the process. On the flip side, in terms of the cause end of things, at a very molecular level certain compounds have been found to be in excess in patients with scleroderma, and so understanding why those compounds ... collagen becomes excessive... is where some of the research is at.

JEANNE BLAKE: Well, we're out of time, but I want to thank both of you for coming in and talking about this. Liz, we just wish you all the very best, and I think on behalf of everybody in South Boston and people living with this disease, good for you for pushing and getting the state in there to look at it, and hopefully others will benefit from that.

LIZ: Thank you.

JEANNE BLAKE: And a final note. I talked with your daughter, I don't know which one it was, but she's polite, so you're doing a good job!

LIZ: Thank you.

JEANNE BLAKE: That's one of the reasons I wanted to ask about what was going on in your family, because you can tell so much about the way a child conducts him or herself on the phone. She got an A+. She is a good kid, and she obviously told you I called. That's another good thing that doesn't happen all the time. All right, thanks to both of you and our very, very best wishes to you. We want to thank you for joining us on this edition of *About Health TV*. I'm Jeanne Blake and I'll see you next time.

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