Scleroderma Q&A: Your questions about the autoimmune disease answered

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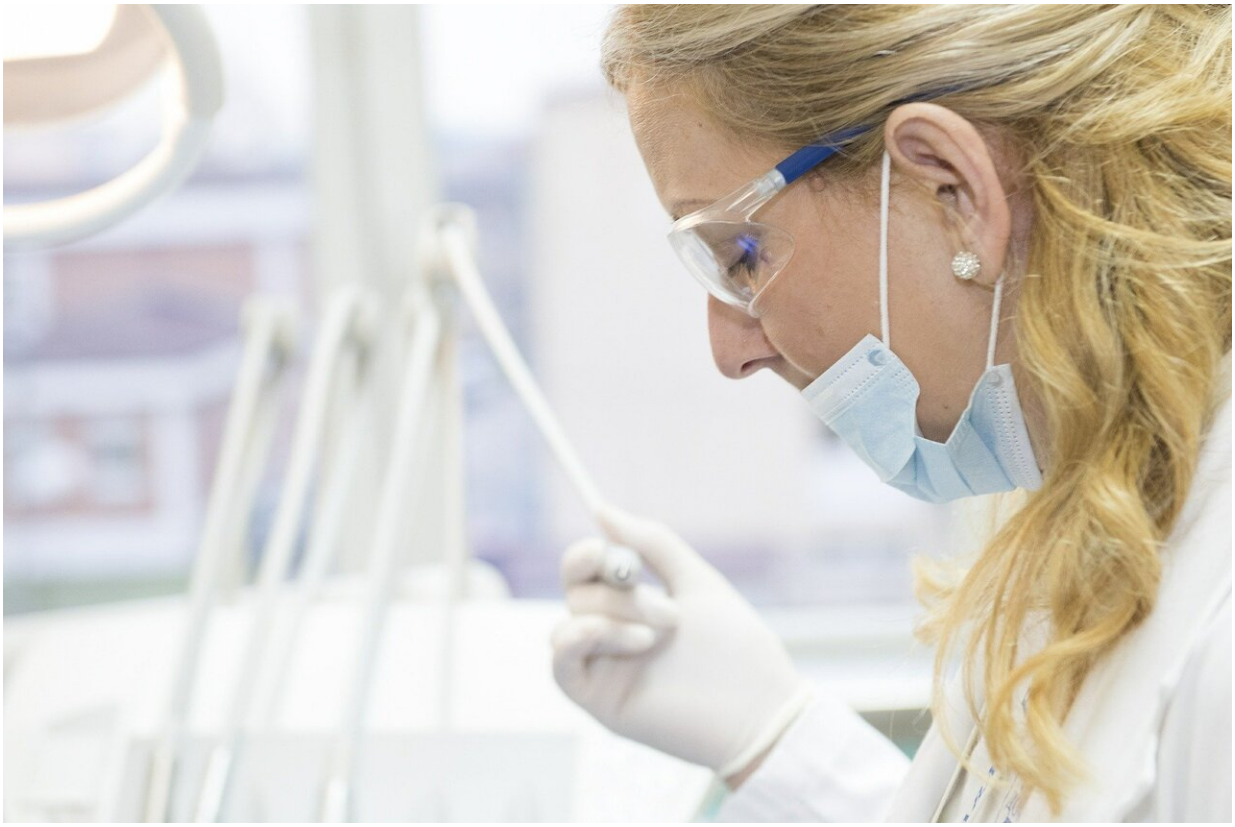

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Like many autoimmune diseases, systemic sclerosis, or scleroderma, can be a challenge to diagnose because its symptoms are varied, can range from mild to debilitating, and often resemble those of other conditions,

such as systemic lupus erythematosus and rheumatoid arthritis.

Although hardening of the skin is the most characteristic sign of scleroderma, what's called "Raynaud's phenomenon" (visible color changes in the fingers and toes in response to cold exposure or stress) is its most frequent symptom. Lung, kidney, and gastrointestinal problems can also develop. Symptoms can come and go, and are easily mistaken for other diseases, prompting many people to go from one specialist to another. Then, when they finally receive a diagnosis, many patients feel frustrated to face a disease that few people have heard of, let alone understand. Even doctors and researchers are trying to better understand it.

"With a disease like scleroderma, I think you need to step into the patient's shoes and see things from their perspective," says Monique Hinchcliff, MD, MS, director of the Yale Medicine Scleroderma Program and clinical director of research for rheumatology at Yale School of Medicine. "I don't ask patients to participate in activities related to research that I wouldn't do myself. I ask patients to donate skin biopsies, and I have donated skin biopsies. I ask patients to undergo bronchoscopy and lung lavage (where fluid is introduced into the lung and then sucked back out to obtain lung cells), and I have undergone bronchoscopy and a lung lavage procedure, so there's a tube of my lung cells in our freezer."

The program has a multidisciplinary team to help scleroderma patients manage the multiple symptoms the disease can cause—and there are treatments for many of those symptoms. For instance, there are now 13 medications approved by the FDA (all becoming available beginning in 1995, says Dr. Hinchcliff) to treat a scleroderma patient with pulmonary arterial hypertension (PAH), a type of high blood pressure in the lungs. "I am dedicated to finding a cure for scleroderma," Dr. Hinchcliff says. "In the meantime, I want to find better treatments for symptoms so

patients can enjoy their lives as fully as possible."

Dr. Hinchcliff answered our questions about scleroderma.

What is scleroderma?

Scleroderma is a chronic connective tissue disease. As many as 300,000 people in the United States have the condition. Four to seven times as many women as men have it, and patients are frequently between ages 25 and 55, although it can occur in adults at any age. The most notable symptom is hardening of the skin, which usually begins on the hands (skin fibroblasts, the cells that help your tissue heal, produce too much collagen). While most patients only experience skin tightening on the hands, face, forearms, and calves—called limited disease—for about a third, the skin tightening spreads to the chest, the upper arms, and legs—called diffuse disease. The esophagus, heart, lungs, kidneys, digestive system, and skin can all be affected in both groups of patients, so a coordinated team approach is required.

How is scleroderma diagnosed?

Scleroderma can be difficult to diagnose because the symptoms resemble other diseases—symptoms across many autoimmune diseases are similar. The symptoms also vary from patient to patient. For instance, a patient might start off having constipation, diarrhea, or heartburn, but then they'll become short of breath. So, a physician might be fooled into thinking that a patient has developed asthma or a more common lung disease when it's really just another manifestation of scleroderma.

Scleroderma is a clinical diagnosis meaning that there is no laboratory or diagnostic test to confirm it. We take into account the patient's history,

laboratory tests, and diagnostic data, as well as a carefully performed physical examination. We look at the results of high-resolution computed tomography of the chest [chest CT], echocardiography, and pulmonary function tests. We may do a skin biopsy. We carefully review these data to determine whether or not the patient has scleroderma. My experience diagnosing and caring for hundreds of patients with scleroderma enables me to make an accurate diagnosis and initiate an individualized plan of care. I partner with patients and their families to find the best solutions to the health issues that they face.

Inflammation plays a role in autoimmune disease. What do you know about its role in scleroderma?

Inflammation tells us that something is awry in our bodies. If we have an infection, inflammation lets us know that something is wrong—the area becomes red and warm and swollen and painful, but when the infection resolves the inflammation subsides. What happens in some diseases like scleroderma is that inflammation sets off a cascade of abnormal tissue responses. The fibroblasts in the tissues remain activated and continue to produce collagen [scar tissue] long after the inflammation has resolved.

You could have two identical twins, and one develops scleroderma and one doesn't. We think one of those twins might have been exposed to something in their environment that "turned on" inflammation, whereas the other twin didn't come into contact with whatever that was. So, I think there's a push for us to understand the toxins in our environment and in our food supply, air, and water, and to commit ourselves to understanding what the environmental triggers are for autoimmune conditions.

What did you learn by undergoing some of the same tests as your patients?

When I underwent a bronchoscopy and lung lavage, I learned a great deal about how we can make our patients more comfortable. For instance, the room where I was seated prior to undergoing bronchoscopy was cold. Patients with scleroderma often have Raynaud's phenomenon that is triggered by cold exposure, so we realized that we needed to provide blankets to patients to prevent Raynaud episodes while they waited for their procedure.

Scleroderma is not a common disease. Do you find many patients get confused by it?

I think patients can become bewildered unless somebody really explains what's going on. Scleroderma is a complex disease. Terms like ILD (interstitial lung disease) and PAH sound like alphabet soup. I often take out a piece of paper and draw the lungs as I explain how there are two main forms of lung disease that tend to affect patients with scleroderma.

There's a lot of misinformation on the internet, and people get depressed and scared when they read things that aren't true. A lot of the first visit is spent reassuring and educating patients. I don't present an overly optimistic picture, but I do give them the facts based on the available data.

What is the treatment for scleroderma?

We don't have a cure at this point, but what I tell patients is that there are only a few diseases that we can cure in medicine. We treat scleroderma as a chronic disease, much like doctors do for patients with diabetes and heart disease. For example, blood vessel spasm [from Raynaud's phenomenon] is common in patients with scleroderma, and there are drugs that can be used to help the blood vessels relax.

Over-the-counter anti-inflammatory drugs and anti-reflux medications can help to reduce other symptoms while immune suppressant medications can help prevent disease progression in the lungs and in the heart. It's important that patients with scleroderma have a specialist who can monitor them closely and manage their condition and any medications they are taking.

What is your approach in the Yale Medicine Scleroderma Program?

The Yale Medicine Scleroderma Program has been designated as a Scleroderma Center of Excellence by the Scleroderma Foundation. This means that we have successfully developed a program that is specifically geared to the special needs of scleroderma patients. We have specialists not only from rheumatology, but also from allergy and immunology, cardiology, dermatology, gastroenterology, nephrology, and pulmonary critical care that are experienced and dedicated to providing top-notch care to scleroderma patients.

Patients with scleroderma lung, gastrointestinal, or heart involvement will find coordinated, individualized care here. We also have specialists who can help patients manage gynecological, nutritional, psychological, urological, and sexual health challenges. The doctors in our program know each other and are used to working with one another, and we work as a team to provide a comprehensive care plan for each patient.

Are you also doing research to learn more about the disease?

We are participating in clinical trials of drugs that have been developed in cooperation with our partners in industry. We're also conducting investigational studies geared towards better understanding the triggers

and drivers of disease in order to identify new treatments. Since my arrival at Yale in the summer of 2018, I've been coordinating scleroderma research with dermatologists, pulmonologists, and cardiologists. We are conducting innovative research to develop new treatments as well as better diagnostic and disease-monitoring tools to propel the field forward.

So, there are many things for patients to think about.

They just know that they feel lousy, and it's up to us to figure out how to make them feel better. We have a dedicated nurse and clinical coordinators who help patients navigate the system and coordinate appointments. If a patient is going to be seen at Yale Medicine for scleroderma, our clinical coordinator will reach out and help them gather all the records from different doctors' offices they have visited. Then, when they come here, we will have the data we need to make an informed medical decision.

Do you find these patients do better with specialty care?

Absolutely. I have three daughters, and I would hope that if they ever developed scleroderma, they would find care in a place that considers the whole person—not just the heart or the skin or the lungs. Physicians need to take a step back and see how the disease is affecting each patient. I think my favorite part of being a specialist in scleroderma is when I see a light bulb go on for the patient, and they finally understand what disease they have, and they feel some level of relief that help is on the way and that it's not going to be so hard for them to get the care that they need.

Is there anything specific that patients with

scleroderma need to know about COVID-19?

Many patients with scleroderma are taking immune suppression medications to control their disease progression. It is likely that these medications may impact their susceptibility to contracting COVID-19. It is especially important for patients with scleroderma to follow all the CDC's social distancing guidelines and hand-washing and mask-wearing recommendations.

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