

Understanding

A group of autoimmune diseases that affects the skin, with some types also a threat to major organs, scleroderma is difficult to diagnose due to symptoms identical to those of other autoimmune diseases.

By Jim Trageser

First described in ancient Greece by Hippocrates (who called it thickened skin), scleroderma was not given its present name until 1836 by Giovambattista Fantonetti. And it was only toward the end of World War II, in 1945, that Dr. Robert H. Goetz described it as a systemic disease.¹ Today, it is classified as a group of chronic connective tissue autoimmune diseases that affects the skin, joints and, all too often, major organs.

Yet, despite two millennia of awareness of scleroderma, its often disfiguring nature can still cause discomfort — as witnessed this past summer when Facebook declined an advertisement that

featured a photograph of a patient's face. Lisa Goodman-Helfand of Highland Park, Ill., tried to purchase a Facebook ad to promote a website that shares her story of dealing with scleroderma.² Goodman-Helfand's scleroderma has affected the skin on her face, and her photos — taken without the makeup she usually wears in public to hide the signs of the disease — showed her skin covered with bright red and purple splotches. After rejecting the advertisement a second time — and after Goodman-Helfand blogged about the situation — Facebook relented, but not without very effectively highlighting the struggles with which scleroderma patients often are confronted.

What Is Scleroderma?

Scleroderma is broken down into two different classifications; however, both are characterized by an excessive production of collagen that creates a thickening of tissue.³ It is believed this is caused due to a breakdown of the body's immune system, with the body's cells reacting as if there is tissue damage that needs repairing even though there is not.



Scleroderma

Localized scleroderma affects the skin only, and usually just a small portion of the skin — often on the hands or face.⁴ While it can be severe and may leave permanent damage to the skin, it can also go away on its own.

Systemic scleroderma affects not only the skin, but the circulatory system and, often, major organs. It has elements of both rheumatic disease and connective tissue disease.⁶ It may affect the heart, lungs or kidneys, with excessive collagen deposits impacting the ability of the body to function normally. In addition, if the muscles in the abdomen are impacted by fibrosis (the formation of excess connective tissue, often resulting in scarring) and unable to move food through the small intestine normally, the ability of the body to properly extract nutrients can be compromised.⁶

What Causes Scleroderma?

The cause of scleroderma is unknown. It is not contagious and is not believed to be hereditary, although it may be at least partly genetic.⁵ It is classified as an autoimmune disease because doctors and researchers believe the immune system plays a role, but that is not definitively known. It is thought that a triggering event — an injury or infection — causes the onset of biological reactions that lead to scleroderma.⁷ Again, this process is not yet fully understood by doctors or researchers.

For unknown reasons, women are more prone to develop scleroderma than are men (researchers are studying whether hormonal differences between the sexes play a role). Localized scleroderma is more common in people of European ancestry, while systemic forms are seen more often among people of African descent.⁶

In addition, localized scleroderma tends to manifest between the ages of 20 years and 40 years, while systemic scleroderma most often shows up between 30 years and 50 years.⁶ Still, the disease is seen in all age groups and all ethnicities.

Symptoms of Scleroderma

Scleroderma symptoms, and whether it is localized or systemic, vary from patient to patient but nearly every case involves a thickening or hardening of an area of skin. These patches vary in size and shape, as well as number and location.⁸ They are often marked by a white area in the middle, with purple around the edges, and might appear shiny from the

tightening of the skin due to thickening.⁴ For those with localized scleroderma, these will likely be the only symptoms.

Those with systemic scleroderma may also experience Raynaud's phenomenon, which is marked by numbness, pain or change of color in the toes or fingers in response to cold temperatures or extreme emotions.⁸ Hands and feet can also become stiff or swollen.³ As systemic scleroderma continues, it can manifest in other ways, including difficulty swallowing (from scarring of the esophagus due to fibrosis), tightening of the skin on fingers and toes, swollen blood vessels on the face and/or extremities, and even seizures and headaches.⁴ Fatigue and loss of appetite are other common symptoms.

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Diagnosing Scleroderma

Because the above symptoms are also associated with other conditions, it can be difficult to make a diagnosis of scleroderma. In fact, just estimating the number of people who have scleroderma is apparently a challenge, with numbers ranging from 49,000 (the National Institutes of Health) to 300,000 (the Scleroderma Foundation).

A definitive diagnosis will generally be made only after the general practitioner has consulted with rheumatologists, pulmonologists, orthopedists and/or dermatologists. In addition to a full medical history and detailed physical examination, a doctor may also order blood tests⁹ and a skin biopsy.⁵ Calcium deposits on the joints, changes in the capillaries at the base of the fingernails or the appearance of certain antibodies in a blood sample can all be clues that a patient has a type of scleroderma.

Treating Scleroderma

There is no cure for scleroderma, nor a method to prevent it. Treatment will depend on the type of scleroderma and the severity of the case.

Localized scleroderma (the type confined to the skin) is further divided into two distinct types: morphea and linear. Morphea is characterized by waxy, shiny patches of thickened skin, which can grow in size or shrink, and often disappear with no treatment. Linear scleroderma manifests as a line of hardened, waxy skin, often resembling a scar from a severe cut. (These are sometimes referred to as *en coup de sabre* due to their resemblance to a sword wound.)

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While there is not currently a way to cure the thickened skin of localized scleroderma, doctors do recommend regular exercise to keep the skin as limber as possible. Nicotine is known to make scleroderma worse, so eliminating smoking or chewing tobacco is important. And moisturizers and analgesic creams can help relieve pain, swelling and stiffness in the affected areas.⁴ Sunscreen can help prevent further damage from exposure to the sun.

Systemic scleroderma, affecting more parts of the body than localized, often requires more detailed treatment plans to relieve symptoms and reduce physical damage from the disease.

Systemic scleroderma is also broken down into different subtypes: limited cutaneous systemic sclerosis and diffuse cutaneous systemic sclerosis. Limited cutaneous systemic sclerosis affects larger portions of the skin than does localized scleroderma, but it does not affect major internal organs (although the esophagus can still be affected). It tends to manifest with painful calcium deposits on the joints, swallowing difficulties, tightening of the skin on the hands and feet and swollen blood vessels. Diffuse cutaneous systemic sclerosis can have all the above symptoms, plus include excess collagen on the lungs (the most common complication), heart and/or kidneys.⁴

Treatment for systemic scleroderma is administered with the aim of controlling symptoms and preventing more serious complications. It may include:⁴

- Nonsteroidal anti-inflammatory drugs to control swelling and pain in the joints arising from calcium deposits
- Blood pressure medications, if the heart or lungs are affected
- Medications to reduce stomach acid if the esophagus is affected

- Antirheumatic drugs to reduce joint swelling and damage
- Immunosuppressants to try to slow the body's creation of excess collagen, particularly in those with lung damage

Patient advocates also point out that any time someone is diagnosed with a chronic condition, emotional well-being is key to a successful treatment plan. A newly diagnosed patient will do better when surrounded by family and friends in a support network.¹⁰

Scleroderma Research

Important advances are already being made in treating the symptoms of scleroderma. For instance, cyclophosphamide has recently been found effective in slowing lung fibrosis.⁶ And, this summer, the U.S. Food and Drug Administration approved a clinical trial of Actemra (tocilizumab), a drug being developed by Genentech that slows the progress of fibrosis.¹¹ If successful, this could provide physicians another tool to help treat the symptoms of scleroderma.

Among the most promising research is the use of intravenous immune globulin (IVIG) in patients who have not responded to immunosuppressants.¹² Researchers in several studies have reported that patients receiving a regimen of IVIG have shorter outbreaks of skin lesions and improved joint function with less pain. Studies in mice indicate that IVIG treatment lessens the production of collagen and lowers the incidence of fibrosis. Georgetown University is currently conducting a clinical trial of 24 subjects to test the effect of IVIG on the skin, muscles, joints, gastrointestinal system and lung involvement.¹³ That study will conclude at the end of 2015, with results to be published in 2016.

But research into a cure for scleroderma is proceeding more slowly. The recent discovery of a gene linked to scleroderma among members of the Choctaw Nation has geneticists re-examining the link between genes and the disease. Researchers are also examining the genetic blueprint to see if they can predict risk factors so doctors might know which type of scleroderma is most likely to develop after an initial diagnosis.⁶ Others are looking at how and why the immune system in women works differently than in men, as well as the biochemistry behind fibrosis.¹⁴

Because the specific mechanisms that cause scleroderma are not fully understood, an immunization against scleroderma is not likely any time soon.

Scleroderma Outlook

Since there is no cure or any preventive treatment, the prognosis

for those diagnosed with scleroderma varies according to the severity of each case. Patients diagnosed with localized scleroderma should see no difference in their lifespan based on the disease, although their quality of life may be diminished depending on its severity and how they respond emotionally. Those with systemic scleroderma may face more serious consequences, depending on what organs are affected and how severely.

The overall five-year survival rate for those with diffuse cutaneous disease is about 80 percent, and for those with limited cutaneous systemic sclerosis, it is roughly 90 percent.¹⁵ Those numbers can vary widely, however, based on whether there are cardiac, pulmonary or renal complications. Younger patients also tend to face more serious prognoses, as do those of African descent. In addition, the more widespread the skin lesions, the more severe the prognosis.

On an optimistic note, all survival rates associated with scleroderma have shown marked growth over the past few decades as our ability to treat the symptoms and reduce organ damage has improved.¹⁵ ■

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