What causes scleroderma?

Although much research is focused on this important question, the cause of scleroderma is not known. Most patients do not have any relatives with scleroderma, nor do their children get scleroderma. Research indicates there is a "susceptibility gene" which raises the likelihood of getting scleroderma, but does not cause scleroderma by itself.

Diagnosing scleroderma

An experienced doctor bases a diagnosis of scleroderma primarily on a patient's medical history and a physical examination. The most important finding is hard skin, which gives the disease its name. About 98% of scleroderma patients have hardening or thickening of the skin of the fingers—and often of the hands, forearms, and face.

Raynaud's phenomenon is present in 95%.

A third abnormality found in over 95% of patients is a positive *antinuclear antibody* (ANA) test.

Treating scleroderma

At present there is no cure for scleroderma, but many treatments are available for specific symptoms. Because there is so much variation in the type and severity of symptoms, it is important that each patient receive individualized care from a doctor who is knowledgeable about the disease.

For referrals to knowledgeable health professionals, for information about our nationwide network of Chapters and Support Groups, and to subscribe to our quarterly magazine with the latest scleroderma news, please call toll-free 1-800-722-4673.

The Scleroderma Foundation's mission is three-fold:

- To help patients and their families cope with scleroderma through mutual support programs, peer counseling, physician referrals, and educational information.
- To promote public awareness and education through patient and health professional seminars, literature, and publicity campaigns.
- To stimulate and support research to improve treatment and ultimately find the cause and cure of scleroderma and related diseases.

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Scleroderma: An overview

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What is scleroderma?

Scleroderma is a chronic, often progressive autoimmune disease in which the body's immune system attacks its own tissues. It falls into the same category as rheumatoid arthritis, lupus, Sjögren's syndrome, and multiple sclerosis (MS).

The disease, which literally means "hard skin," can cause thickening and tightening of skin, and in some cases serious damage to internal organs including lungs, heart, kidneys, esophagus, and gastrointestinal (GI) tract.

Scleroderma can vary a great deal in terms of severity. For a few individuals it is merely a nuisance; for others it is a life-threatening illness. For most, it is a disease that affects how they live their daily lives.

Although medicines can sometimes help treat symptoms, there is no cure yet for scleroderma.

Types of scleroderma

There are two basic types of scleroderma: the *systemic* form that affects the internal organs or internal systems of the body, and the *localized* form, that affects a local area of skin.

1. Systemic scleroderma

In *systemic* scleroderma (also called *systemic sclerosis*), the immune system causes damage to two main areas: the small blood vessels, and the collagen-producing cells located in the skin and throughout the body.

In *systemic* scleroderma, the small blood vessels in the fingers tend to narrow, and sometimes the blood channel is completely closed off. As a result, small cuts on the hands are slow to heal, and sometimes ulcers form spontaneously.

Due to this decreased blood supply, scleroderma patients are notoriously cold-sensitive. This vascular aspect of the disease is responsible for *Raynaud's phenomenon* (color changes of the fingers upon exposure to cold), which occurs in about 95% of people with *systemic sclerosis*.

The collagen aspect of the disease is responsible for the thick and tight skin, lung and heart problems, and GI tract features. Under normal conditions, the immune system signals cells to produce collagen to form a scar after an area of injury or infection has been cleared. In scleroderma, this scar tissue is produced for no apparent reason, and builds up in the skin and other organs. Many patients will have trouble only with the GI tract, and have normal lungs, hearts, and kidneys. All combinations of symptoms are seen.

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To further complicate matters, *systemic* scleroderma is divided into two forms: *limited* and *diffuse*, referring to the degree of skin involvement. Both forms are associated with internal organ damage, but the *limited* form tends to have less severe organ problems.

Limited scleroderma often is referred to as the CREST form. (The Scleroderma Foundation offers another brochure specifically on the topic of CREST.) CREST is an acronym that stands for

C alcinosis
R aynaud's phenomenon
E sophageal dysfunction
S clerodactyly
T elangiectasias

Calcinosis refers to calcium deposits in the skin. *Raynaud's phenomenon* refers to color changes (white, purple, or red) in the fingers due to cold exposure. *Esophageal dysfunction* refers to acid in the esophagus, felt as heartburn. *Sclerodactyly* means tight and thick skin of the fingers. *Telangiectasias* is a type of red spots in the skin.

2. Localized scleroderma

Localized scleroderma affects the collagen-producing cells in just some skin areas and usually spares the internal organs and blood vessels.

Localized scleroderma occurs either as patches of thickened skin (*morphea*), or as *linear* scleroderma, a line of thickened skin that may extend down an arm or leg. If this line involves the forehead, it is called *en coup de sabre* or "cut of the saber."

It is easy to confuse the terms *localized* scleroderma (which is one of the two main types of scleroderma) and *limited* scleroderma (one of the two types of *systemic* scleroderma, also called CREST). However, this terminology is widespread and not likely to change.

3. Scleroderma-related disorders

Scleroderma can also occur in patients with other autoimmune disorders, as part of an overlap syndrome. A variety of drugs and chemicals can also produce lesions similar to those seen in scleroderma.

Who gets scleroderma?

Localized scleroderma is more common in children, whereas *systemic* scleroderma is more common in adults. Overall, female patients outnumber males about 4:1, and the average patient is diagnosed in her 40s.

Having said this, it is important to note that scleroderma is a disease of many exceptions.