AMERICAN COLLEGE OF RHEUMATOLOGY
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Specialists in Arthritis Care & Research

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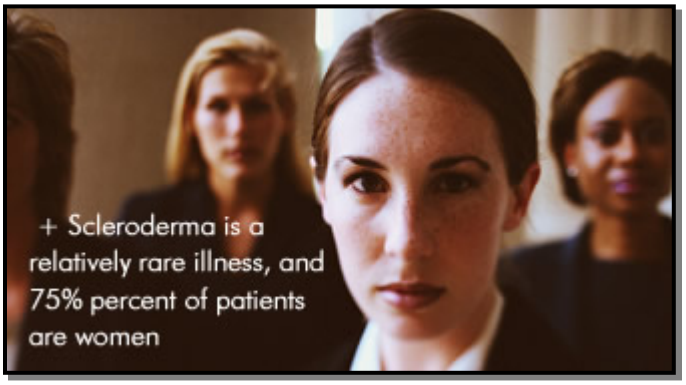

Scleroderma (Systemic Sclerosis)

Scleroderma, or systemic sclerosis, is a disease affecting the skin and other organs that is one of the autoimmune rheumatic diseases. The primary finding in scleroderma is thickening and tightening of the skin. Effective treatments are available for some forms of the disease, although, scleroderma is not yet curable.

Fast Facts

- Scleroderma is a relatively uncommon problem, affecting only 200 to 300 people per million in the U.S. Some 12 to 20 new cases per million are diagnosed annually.
- While scleroderma affects both adults and children, it is most common among adult women.
- Promising research is shedding light on the relationship between the immune system and scleroderma, although the underlying cause is not yet known.
- Medicines traditionally used to treat other autoimmune diseases – such as rheumatoid arthritis and lupus – often have little effect on scleroderma.

What is scleroderma?

Scleroderma, also known as systemic sclerosis, is a chronic disease that causes skin thickening and tightening, a buildup of scar tissue, and damage to internal body organs. There are several types of scleroderma and related diseases with complications ranging from minor to life-threatening. Therefore, the terminology can be confusing.

The two broad categories are “localized scleroderma” which indicates distinct skin lesions, and "systemic sclerosis" (scleroderma) which indicates similar skin symptoms and the potential for internal organ disease.

Other diseases affecting the skin that may be confused with scleroderma include scleredema, scleromyxedema, eosinophilic fasciitis, and nephrogenic systemic fibrosis.

Localized Scleroderma

Localized scleroderma refers to disorders of the skin and sometimes the deeper tissues. The most visible effects are skin lesions or morphea. In some cases, localized scleroderma is just a cosmetic problem. However, for those with widespread skin lesions over their body (generalized morphea or linear

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scleroderma) the thickness and scarring spreads down to the underlying structures including fat, muscle and, on rare occasion, bone. In these instances, the disease can be more serious.

Another pattern of localized scleroderma, called en coup de sabre, usually involves the face.

Localized scleroderma, including deep and extensive lesions, can prevent normal motion of joints and interfere with daily activities. However, this disorder does not affect internal organs of the body.

Systemic Sclerosis (Scleroderma)
Systemic scleroderma can be divided into either "limited" cutaneous systemic sclerosis in which occurs only on the forearms, hands, legs, feet, and face, or "diffuse" cutaneous systemic sclerosis which can affect almost any area of the body.

Cutaneous, or skin, changes are caused by an increase and accumulation of collagen and other proteins that lead to thickening and hardening. This accumulation can extend to other organs including the kidneys, lungs, heart, gastrointestinal tract, and vascular system.

The earliest changes in the body are:

- Immune system problems which may cause tissue damage, particularly in the lungs, as well as inflammation and swelling in the skin. The extent of skin and internal organ involvement dictates whether the diagnosis is labeled as "limited" or "diffuse."
- Damage to blood vessels. For instance, Raynaud's phenomenon, an abnormal reaction of blood vessels to the cold, is caused by structural damage of vessels in the hands, as well as elsewhere in the body. Other blood vessel problems can cause poor blood flow, and lead to severe damage such as finger ulcers or gangrene. These changes also lead to fibrosis (scarring) and other forms of damage in multiple organs.

What causes scleroderma?
The cause of scleroderma is not known. Genetic factors appear to increase a patient's chance of getting the disease. However, some data suggests that exposure to industrial solvents or an environmental agent may play a role in leading to scleroderma. Scleroderma-like syndromes also have been clearly linked to agents as varied as contaminated rapeseed oil, polyvinylchloride, and an impurity in one preparation of L-tryptophan. That said, the vast majority of patients with scleroderma do not have a history of exposure to any suspicious toxins.

Who gets scleroderma?
Scleroderma is a relatively rare illness affecting only 75,000 – 100,000 people in the United States. Of these, 75% percent are women, usually diagnosed between the ages of 30 and 50 years. Twins and family members of patients with scleroderma or other autoimmune connective tissue diseases, such as lupus, appear to be at a slightly increased risk. Children can get scleroderma, although the pattern and extent of disease may be different for children compared to adults.

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How is scleroderma diagnosed?

A diagnosis of scleroderma is based on finding an unusual thickening or swelling of the skin, especially on the hands and extending up the arms. In addition, there may be dilated blood vessels in the face, hands, nail folds and elsewhere. Some patients develop deposits of calcium (calcinosis) in the skin and other organs including the lungs, muscles and the kidneys.

Almost all (more than 90%) of people with scleroderma also have Raynaud's phenomenon. Many also have heartburn and difficulty swallowing. However, since Raynaud's phenomenon and heartburn can be caused by many other conditions, they are not specific disease indicators.

This image shows diffuse soft-tissue swelling of the digits, characteristic of the early edematous phase of scleroderma.

The diagnosis is confirmed by a combination of a person's description of symptoms (history) and physical examination findings. Laboratory tests and x-rays may help, but no one test makes the diagnosis certain. For instance, blood tests for auto-antibodies are often used in helping to make the diagnosis, but the presence or absence of these antibodies is not conclusive.

How is scleroderma treated?

Unfortunately, while some treatments have proven effective, no drug has been found that can arrest or reverse the skin thickening that is the evidence of disease.

For patients with Raynaud's phenomenon, keeping one's fingers and toes warm and adequately protected from exposure to cold, as well as keeping one's core body (trunk) warm has also proven effective in preventing attacks and subsequent damage. Protective measures to avoid trauma to the finger tips are also important.

Early recognition of scleroderma kidney disease is critical. Therefore, patients with diffuse scleroderma should monitor their blood pressure several times a week. Early intervention with a type of blood pressure medication called “angiotensin converting enzyme inhibitors (ACE inhibitors),” has been shown to be extremely effective in treating early scleroderma kidney involvement and helping to prevent renal damage. The use of these drugs has been a major advance for patients with scleroderma.

Glucocorticoids (such as prednisone), intravenous immunoglobin (IVIg), and/or immunosuppressive medications may be effective treatments for those with muscle disease and associated weakness. There are two main types of lung disease that some patients with scleroderma may develop: interstitial lung disease (inflammation and scarring) and pulmonary hypertension (increased pressure in the arteries of the lung).

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Clinical trials have demonstrated that cyclophosphamide is somewhat effective in treating the interstitial lung disease in scleroderma. Clinical trials are underway assessing the effectiveness of several other agents for this condition.

A number of agents have become available in the last 10 years to treat pulmonary hypertension that can occur, including prostacyclin-like drugs (epoprostenol, treprostinol, iloprost) and the endothelin receptor antagonists (bosentan, ambrisentan), and PDE-5 inhibitors (sildenafil, vardenafil, tadalafil). These treatments, and others being tested in current research trials, may provide significant benefit to scleroderma patients with lung disease.

Much research is ongoing into new treatments for scleroderma. Patients and their families should know that experts remain optimistic and take comfort in the fact that work towards a cure will continue.

Broader health impact of scleroderma

Scleroderma can involve almost every organ system including the skin, vascular system, lungs, gastrointestinal tract, heart, joints, muscles, etc. Furthermore, this disease varies greatly from patient to patient, and can dramatically impact someone's life.

Patients should receive care from a specialist (often a team of specialists) with expertise in the management of scleroderma. Support from family and friends are also integral to learning to live with the disease and maintaining a good quality of life.

Living with scleroderma

Living with this condition requires mental and physical changes. First, there is the adjustment to an altered appearance and self-image from the obvious physical feature changes. The psychological component to living with chronic disease, particularly those that are uncommon and currently incurable, should also receive attention.

On a physical level, intestinal involvement may require changes in diet with frequent small meals rather than the usual large ones. Keeping the skin well-lubricated and using appropriate precaution during activities conducive to finger injury (gardening, cooking, opening envelopes) is equally important.

Dressing appropriately to maintain body warmth and protect from blood vessel disease means layering clothing to the point of warmth, as well as adjusting surrounding temperatures where possible. Unfortunately, moving to a warmer climate does not necessarily lead to dramatic improvement. Exercise and/or physical therapy help maintain mobility of joints, which are affected by scarring of the tissues in and around the joints.

Points to remember

- Scleroderma differs from person to person, but can be a very serious disease.
- Physical measures and medications will help control Raynaud's phenomenon and heartburn in many patients.
- Effective treatments are available even for patients with severe disease, including acute kidney disease, pulmonary hypertension, lung inflammation, and gastrointestinal problems.

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- The key is to recognize organ involvement early and treat it before irreversible organ damage occurs.
- Patients with scleroderma should be seen by physicians with specialized expertise in the care of this complex disease.

To find a rheumatologist

For more information about rheumatologists, click here.

For a listing of rheumatologists in your area, click here.

For more information

The American College of Rheumatology has compiled this list to give you a starting point for your own additional research. The ACR does not endorse or maintain these Web sites, and is not responsible for any information or claims provided on them. It is always best to talk with your rheumatologist for more information and before making any decisions about your care.

The Scleroderma Foundation
www.scleroderma.org

International Scleroderma Network
www.sclero.org

Scleroderma Clinical Trials Consortium
www.sctc-online.org

Scleroderma Research Foundation
www.sclerodermaresearch.org

The Arthritis Foundation
www.arthritis.org

National Institute of Arthritis and Musculoskeletal and Skin Diseases Information Clearinghouse
http://www.niams.nih.gov/Health_Info/Scleroderma/default.asp

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Updated by Peter A. Merkel, MD, MPH; and reviewed by the American College of Rheumatology Patient Education Task Force. Fact sheet originally written by Joseph H. Korn, MD and Peter A. Merkel, MD, MPH.

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