

Autoimmune Resource & Research



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HANDOUT ON HEALTH - SCLERODERMA

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What Is Scleroderma?

Derived from the Greek words "sklerosis," meaning hardness, and "derma," meaning skin, scleroderma literally means hard skin. Though it is often referred to as if it were a single disease, scleroderma is really a symptom of a group of diseases that involve the abnormal growth of connective tissue, which supports the skin and internal organs. It is sometimes used, therefore, as an umbrella term for these disorders. In some forms of scleroderma, hard, tight skin is the extent of this abnormal process. In other forms, however, the problem goes much deeper, affecting blood vessels and internal organs, and rarely the heart, lungs, and kidneys.

Scleroderma is called both a rheumatic disease and a connective tissue disease. The term rheumatic disease refers to a group of conditions characterized by inflammation and/or pain in the muscles, joints, or fibrous tissue. A connective tissue disease is one that affects the major substances in the skin, tendons, and bones.

What Are the Different Types of Scleroderma?

The group of diseases we call scleroderma falls into two main classes: localized scleroderma and systemic scleroderma. Localized diseases affect only certain parts of the body; systemic diseases can affect the whole body.

Localized Scleroderma

Localized types of scleroderma are those limited to the skin and related tissues and, in some cases, the muscle below. Internal organs are not affected by localized scleroderma, and localized scleroderma can never progress to the systemic form of the disease. Often, localized conditions improve or go away on their own over time, but the skin changes and damage that occur when the disease is active can be permanent. For some people, localized scleroderma is serious and disabling.

There are two generally recognized types of localized scleroderma:

Morphea: Morphea comes from a Greek word that means "form" or "structure." The word refers to local patches of scleroderma. The first signs of the disease are reddish patches of skin that thicken into firm, oval-shaped areas. The center of each patch becomes ivory colored with violet borders. These patches sweat very little and have little hair growth. Patches appear most often on the chest, stomach, and back. Sometimes they appear on the face, arms, and legs.

Morphea can be either localized or generalized. *Localized morphea* limits itself to one or several patches, ranging in size from a half-inch to 12 inches in diameter. The condition sometimes appears on areas treated by radiation therapy. Some people have both morphea and linear scleroderma (described below). The disease is referred to as *generalized morphea* when the skin patches become very hard and dark and spread over larger areas of the body.

Regardless of the type, morphea generally fades out in 3 to 5 years; however, people are often left with darkened skin patches and, in rare cases, muscle weakness.

Linear scleroderma: As suggested by its name, the disease has a single line or band of thickened and/or abnormally colored skin. Usually, the line runs down an arm or leg, but in some people it runs down the forehead. People sometimes use the French term *en coup de sabre*, or "sword stroke," to describe this highly visible line.

Systemic Scleroderma (also known as Systemic Sclerosis)

Systemic scleroderma, or systemic sclerosis, is the term for the disease that not only includes the skin, but also involves the tissues beneath to the blood vessels and major organs. Systemic scleroderma is typically broken down into *diffuse* and *limited* disease. People with systemic scleroderma often have all or some of the symptoms that some doctors call CREST, which stands for the following:

- **Calcinosis:** the formation of calcium deposits in the connective tissues, which can be detected by x ray. They are typically found on the fingers, hands, face, and trunk and on the skin above elbows and knees. When the deposits break through the skin, painful ulcers can result.
- **Raynaud's phenomenon:** a condition in which the small blood vessels of the hands and/or feet contract in response to cold or anxiety. As the vessels contract, the hands or feet turn white and cold, then blue. As blood flow returns, they become red and painful. Fingertip tissues may suffer damage, leading to ulcers, scars, or gangrene.
- **Esophageal dysfunction:** impaired function of the esophagus (the tube connecting the throat and the stomach) that occurs when smooth muscles in the esophagus lose normal movement. In the upper esophagus, the result can be swallowing difficulties; in the lower esophagus, the problem can cause chronic heartburn or inflammation.
- **Sclerodactyly:** thick and tight skin on the fingers, resulting from deposits of excess collagen within skin layers. The condition makes it harder to bend or straighten the fingers. The skin may also appear shiny and darkened, with hair loss.
- **Telangiectasias:** small red spots on the hands and face that are caused by the swelling of tiny blood vessels. While not painful, these red spots can create cosmetic problems.

Limited scleroderma: Limited scleroderma typically comes on gradually and affects the skin only in certain areas: the fingers, hands, face, lower arms, and legs. Many people with limited disease have Raynaud's phenomenon for years before skin thickening starts. Others start out with skin problems over much of the body, which improves over time, leaving only the face and hands with tight, thickened skin.

Telangiectasias and calcinosis often follow. Because of the predominance of CREST in people with limited disease, some doctors refer to limited disease as the CREST syndrome.

Diffuse scleroderma: Diffuse scleroderma typically comes on suddenly. Skin thickening occurs quickly and over much of the body, affecting the hands, face, upper arms, upper legs, chest, and stomach in a symmetrical fashion (for example, if one arm or one side of the trunk is affected, the other is also affected). Some people may have more area of their skin affected than others. Internally, it can damage key organs such as the heart, lungs, and kidneys.

People with diffuse disease are often tired, lose appetite and weight, and have joint swelling and/or pain. Skin changes can cause the skin to swell, appear shiny, and feel tight and itchy.

The damage of diffuse scleroderma typically occurs over a few years. After the first 3 to 5 years, people with diffuse disease often enter a stable phase lasting for varying lengths of time. During this phase, skin thickness and appearance stay about the same. Damage to internal organs progresses little, if at all. Symptoms also subside: joint pain eases, fatigue lessens, and appetite returns.

Gradually, however, the skin starts to change again. Less collagen is made and the body seems to get rid of the excess collagen. This process, called "softening," tends to occur in reverse order of the thickening process: the last areas thickened are the first to begin softening. Some patients' skin returns to a somewhat normal state, while other patients are left with thin, fragile skin without hair or sweat glands. More serious damage to heart, lungs, or kidneys is unlikely to occur unless previous damage leads to more advanced deterioration.

People with diffuse scleroderma face the most serious long-term outlook if they develop severe kidney, lung, digestive, or heart problems. Fortunately, less than one-third of patients with diffuse disease develop these problems. Early diagnosis and continual and careful monitoring are important.

What Causes Scleroderma?

Although scientists don't know exactly what causes scleroderma, they are certain that people cannot catch it from or transmit it to others. Studies of twins suggest it is also not inherited. Scientists suspect that scleroderma comes from several factors that may include:

Abnormal immune or inflammatory activity: Like many other rheumatic disorders, scleroderma is believed to be an autoimmune disease. An autoimmune disease is one in which the immune system, for unknown reasons, turns against one's own body.

In scleroderma, the immune system is thought to stimulate cells called fibroblasts to produce too much collagen. In scleroderma, collagen forms thick connective tissue that builds up around the cells of the skin and internal organs. In milder forms, the effects of this buildup are limited to the skin and blood vessels. In more serious forms, it also can interfere with normal functioning of skin, blood vessels, joints, and internal organs.

Genetic makeup: While genes seem to put certain people at risk for scleroderma and play a role in its course, the disease is not passed from parent to child like some genetic diseases.

Environmental triggers: Research suggests that exposure to some environmental factors may trigger the disease in people who are genetically predisposed to it. Suspected triggers include viral infections, certain adhesive and coating materials, and organic solvents such as vinyl chloride or trichloroethylene. In the past, some people believed that silicone breast implants might have been a factor in developing connective tissue diseases such as scleroderma. But several studies have not shown evidence of a connection.

Hormones: By the middle to late childbearing years (ages 30 to 55), women develop scleroderma at a rate 7 to 12 times higher than men. Because of female predominance at this and all ages, scientists suspect that something distinctly feminine, such as the hormone estrogen, plays a role in the disease. So far, the role of estrogen or other female hormones has not been proven.

Who Gets Scleroderma?

Although scleroderma is more common in women, the disease also occurs in men and children.

For some people, scleroderma (particularly the localized forms) is fairly mild and resolves with time. But for others, living with the disease and its effects day to day has a significant impact on their quality of life.

How Can Scleroderma Affect My Life?

Having a chronic disease can affect almost every aspect of your life, from family relationships to holding a job. For people with scleroderma, there may be other concerns about appearance or even the ability to dress, bathe, or handle the most basic daily tasks. Here are some areas in which scleroderma could intrude.

Appearance and self-esteem: Aside from the initial concerns about health and longevity, one of the first fears people with scleroderma have is how the disease will affect their appearance. Thick, hardened skin can be difficult to accept, particularly on the face. Systemic scleroderma may result in facial changes that eventually cause the opening to the mouth to become smaller and the upper lip to virtually disappear. Linear scleroderma may leave its mark on the forehead. Although these problems can't always be prevented, their effects may be minimized with proper treatment and skin care. Special cosmetics--and in some cases, plastic surgery--can help conceal scleroderma's damage.

Caring for yourself: Tight, hard connective tissue in the hands can make it difficult to do what were once simple tasks, such as brushing your teeth and hair, pouring a cup of coffee, using a knife and fork, unlocking a door, or buttoning a jacket. If you have trouble using your hands, consult an occupational therapist, who can recommend new ways of doing things or devices to make tasks easier. Devices as simple as Velcro* fasteners and built-up brush handles can help you be more independent.

Family relationships: Spouses, children, parents, and siblings may have trouble understanding why you don't have the energy to keep house, drive to soccer practice, prepare meals, and hold a job the way you used to. If your condition isn't that visible, they may even suggest you are just being lazy. On the other hand, they may be overly concerned and eager to help you, not allowing you to do the things you are able to do or giving up their own interests and activities to be with you. It's important to learn as much about your form of the disease as you can and share any information you have

with your family. Involving them in counseling or a support group may also help them better understand the disease and how they can help you.

Sexual relations: Sexual relationships can be affected when systemic scleroderma enters the picture. For men, the disease's effects on the blood vessels can lead to problems achieving an erection. In women, damage to the moisture-producing glands can cause vaginal dryness that makes intercourse painful. People of either sex may find they have difficulty moving the way they once did. They may be self-conscious about their appearance or afraid that their sexual partner will no longer find them attractive. With communication between partners, good medical care, and perhaps counseling, many of these changes can be overcome or at least worked around.

How Is Scleroderma Diagnosed?

To make a diagnosis, your doctor will ask you a lot of questions about what has happened to you over time and about any symptoms you may be experiencing. Are you having a problem with heartburn or swallowing? Are you often tired or achy? Do your hands turn white in response to anxiety or cold temperatures?

Once your doctor has taken a thorough medical history, he or she will perform a physical exam. Finding one or more of the following factors can help the doctor diagnose a certain form of scleroderma:

- Changed skin appearance and texture, including swollen fingers and hands and tight skin around the hands, face, mouth, or elsewhere.
- Calcium deposits developing under the skin.
- Changes in the tiny blood vessels (capillaries) at the base of the fingernails.
- Thickened skin patches.

Finally, your doctor may order lab tests to help confirm a suspected diagnosis. At least two proteins, called antibodies, are commonly found in the blood of people with scleroderma:

- Antitopoisomerase-1 or Anti-Scl-70 antibodies appear in the blood of up to 40 percent of people with diffuse systemic sclerosis.
- Anticentromere antibodies are found in the blood of as many as 90 percent of people with limited systemic sclerosis.

How Is Scleroderma Treated?

Because scleroderma can affect many different organs and organ systems, you may have several different doctors involved in your care. Typically, care will be managed by a rheumatologist, a specialist who treats people with diseases of the joints, bones and muscles, or an immunologist who specializes in diseases of the immune system. They may refer you to other specialists, depending on the specific problems you are having: for example, a dermatologist for the treatment of skin symptoms, a nephrologist for kidney complications, a cardiologist for heart complications, a gastroenterologist for problems of the digestive tract, and a pulmonary specialist for lung involvement.

In addition to doctors, professionals like nurse practitioners, physician assistants, physical or occupational therapists, psychologists, and social workers may play a role in

your care. Dentists, orthodontists, and even speech therapists can treat oral complications that arise from thickening of tissues in and around the mouth and on the face.

Currently, there is no treatment that controls or stops the underlying problem--the overproduction of collagen--in all forms of scleroderma. Thus, treatment and management focus on relieving symptoms and limiting damage. Your treatment will depend on the particular problems you are having. Some treatments will be prescribed or given by your physician. Others are things you can do on your own.

Here are some of the potential problems that can occur in systemic scleroderma and the medical and nonmedical treatments for them.

[Note: This is not a complete listing of problems or their treatments. Different people experience different problems with scleroderma and not all treatments work equally well for all people. Work with your doctor to find the best treatment for your specific symptoms.]

Raynaud's phenomenon: One of the most common problems associated with scleroderma, Raynaud's phenomenon can be uncomfortable and can lead to painful skin ulcers on the fingertips. Smoking makes the condition worse. The following measures may make you more comfortable and help prevent problems:

- Don't smoke! Smoking narrows the blood vessels even more and makes Raynaud's phenomenon worse.
- Dress warmly, with special attention to hands and feet. Dress in layers and try to stay indoors during cold weather.
- Use biofeedback (to control various body processes that are not normally thought of as being under conscious control) and relaxation exercises.
- For severe cases, speak to your doctor about prescribing drugs called calcium channel blockers, which can open up small blood vessels and improve circulation.

Stiff, painful joints: In diffuse systemic scleroderma, hand joints can stiffen because of hardened skin around the joints or inflammation of the joints themselves. Other joints can also become stiff and swollen. The following may help:

- Exercise regularly. Ask your doctor or physical therapist about an exercise plan that will help you increase and maintain range of motion in affected joints. Swimming can help maintain muscle strength, flexibility, and joint mobility.
- Use an over-the-counter or prescription nonsteroidal anti-inflammatory drug, as recommended by your doctor, to help relieve joint or muscle pain. If pain is severe, speak to a rheumatologist about the possibility of prescription-strength drugs to ease pain and inflammation.
- Learn to do things in a new way. A physical or occupational therapist can help you learn to perform daily tasks, such as lifting and carrying objects or opening doors, in ways that will put less stress on tender joints.

Skin problems: When too much collagen builds up in the skin, it crowds out sweat and oil glands, causing the skin to become dry and stiff. If your skin is affected, you may need to see a dermatologist. To ease dry skin, try the following:

- Apply oil-based creams and lotions frequently, and always right after bathing.
- Apply sunscreen before you venture outdoors, to protect against further damage by the sun's rays.
- Use humidifiers to moisten the air in your home in colder winter climates. (Clean humidifiers often to stop bacteria from growing in the water.)
- Avoid very hot baths and showers, as hot water dries the skin.
- Avoid harsh soaps, household cleaners, and caustic chemicals, if at all possible. If that's not possible, be sure to wear rubber gloves when you use such products.
- Exercise regularly. Exercise, especially swimming, stimulates blood circulation to affected areas.

Dry mouth and dental problems: Dental problems are common in people with scleroderma for a number of reasons: tightening facial skin can make the mouth opening smaller and narrower, which makes it hard to care for teeth; dry mouth due to salivary gland damage speeds up tooth decay; and damage to connective tissues in the mouth can lead to loose teeth. You can avoid tooth and gum problems in several ways:

- Brush and floss your teeth regularly. (If hand pain and stiffness make this difficult, consult your doctor or an occupational therapist about specially made toothbrush handles and devices to make flossing easier.)
- Have regular dental checkups. Contact your dentist immediately if you experience mouth sores, mouth pain, or loose teeth.
- If decay is a problem, ask your dentist about fluoride rinses or prescription toothpastes that remineralize and harden tooth enamel.
- Consult a physical therapist about facial exercises to help keep your mouth and face more flexible.
- Keep your mouth moist by drinking plenty of water, sucking ice chips, using sugarless gum and hard candy, and avoiding mouthwashes with alcohol. If dry mouth still bothers you, ask your chemist about a saliva substitute that can stimulate the flow of saliva.

Gastrointestinal (GI) problems: Systemic scleroderma can affect any part of the digestive system. As a result, you may experience problems such as heartburn, difficulty swallowing, early satiety (the feeling of being full after you've barely started eating), or intestinal complaints such as diarrhea, constipation, and gas. In cases where the intestines are damaged, your body may have difficulty absorbing nutrients from food. Although GI problems are diverse, here are some things that might help at least some of the problems you have:

- Eat small, frequent meals.
- Raise the head of your bed with blocks, and stand or sit for at least an hour (preferably two or three) after eating to keep stomach contents from backing up into the esophagus.
- Avoid late-night meals, spicy or fatty foods, and alcohol and caffeine, which can aggravate GI distress.
- Chew foods well and eat moist, soft foods. If you have difficulty swallowing or if your body doesn't absorb nutrients properly, your doctor may prescribe a special diet.
- Ask your doctor about prescription medications for problems such as diarrhea, constipation, and heartburn. Some drugs called proton pump inhibitors are highly effective against heartburn. Oral antibiotics may stop bacterial overgrowth in the bowel that can be a cause of diarrhea in some people with systemic scleroderma.

Lung damage: About 10 to 15 percent of people with systemic scleroderma develop severe lung disease, which comes in two forms: pulmonary fibrosis (hardening or scarring of lung tissue because of excess collagen) and pulmonary hypertension (high blood pressure in the artery that carries blood from the heart to the lungs).

- Watch for signs of lung disease, including fatigue, shortness of breath or difficulty breathing, and swollen feet. Report these symptoms to your doctor.
- Have your lungs closely checked, using standard lung-function tests, during the early stages of skin thickening. These tests, which can find problems at the earliest and most treatable stages, are needed because lung damage can occur even before you notice any symptoms.
- Get regular flu and pneumonia vaccines as recommended by your doctor. Contracting either illness could be dangerous for a person with lung disease.

Heart problems: About 15 to 20 percent of people with systemic scleroderma develop heart problems, including scarring and weakening of the heart (cardiomyopathy), inflamed heart muscle (myocarditis), and abnormal heart beat (arrhythmia). All of these problems can be treated. Treatment ranges from drugs to surgery, and varies depending on the nature of the condition.

Kidney problems: About 15 to 20 percent of people with diffuse systemic scleroderma develop severe kidney problems, including loss of kidney function. Because uncontrolled high blood pressure can quickly lead to kidney failure, it's important that you take measures to minimize the problem. Things you can do:

- Check your blood pressure regularly and, if you find it to be high, call your doctor right away.
- If you have kidney problems, take your prescribed medications faithfully. In the past two decades, drugs known as ACE (angiotensin-converting enzyme) inhibitors, including captopril (Capoten), enalapril (Vasotec), and quinapril (Accupril), have made scleroderma-related kidney failure a less-threatening

problem than it was in the past. But for these drugs to work, you must take them.

How Can I Play a Role in My Health Care?

Although your doctors direct your treatment, you are the one who must take your medicine regularly, follow your doctor's advice, and report any problems promptly. In other words, the relationship between you and your doctors is a partnership, and you are the most important partner. Here's what you can do to make the most of this important role:

- **Educate yourself:** Knowledge is your best defense against this disease. Learn as much as you can about scleroderma, both for your own benefit and to educate the people in your support network (see below).
- **Seek support:** Recruit family members, friends, and coworkers to build a support network. This network will help you get through difficult times: when you are in pain; when you feel angry, sad, or afraid; when you're depressed. Also, look for a scleroderma support group in your community.
- **Assemble a health care team:** You and your doctors will lead the team. Other members may include physical and occupational therapists, a psychologist or social worker, a dentist, and a pharmacist.
- **Be patient:** Understand that a final diagnosis can be difficult and may take a long time. Find a doctor with experience treating people with systemic and localized scleroderma. Then, even if you don't yet have a diagnosis, you will get understanding and the right treatment for your symptoms.
- **Speak up:** When you have problems or notice changes in your condition, don't feel too self-conscious to speak up during your appointment or even call your doctor or another member of your health care team. No problem is too small to inquire about, and early treatment for any problem can make the disease more manageable for you and your health care team. Make a list of questions to ask your doctor so you don't forget anything.
- **Don't accept depression:** While it's understandable that a person with a chronic illness like scleroderma would become depressed, don't accept depression as a normal consequence of your condition. If depression makes it hard for you to function well, don't hesitate to ask your health care team for help. You may benefit from speaking with a psychologist or social worker or from using one of the effective medications on the market.
- **Learn coping skills:** Skills like meditation, calming exercises, and relaxation techniques may help you cope with emotional difficulties as well as help relieve pain and fatigue. Ask a member of your health care team to teach you these skills or to refer you to someone who can.
- **Ask the experts:** If you have problems doing daily activities, from brushing your hair and teeth to driving your car, consult an occupational or physical therapist. They have more helpful hints and devices than you can probably imagine. Social workers can often help resolve financial and insurance matters.

More Questions? Count on More Answers

Scleroderma poses a series of challenges for both patients and their health care teams. The good news is that scientists, doctors, and other health care professionals continue to find new answers--ways to make earlier diagnoses and manage disease better. In addition, active patient support groups share with, care for, and educate each other. The impact of all of this activity is that people with scleroderma do much better and remain active far longer than they did 20 or 30 years ago. As for tomorrow, patients and the medical community will continue to push for longer, healthier, and more active lives for people with the diseases collectively known as scleroderma.

Reviewed by: Dr. Michael Boyle & Dr. Glenn Reeves (29 October 2002)

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