August 2016

Handout on Health: Scleroderma

This publication is for people who have scleroderma, as well as for their family members, friends, and others who want to find out more about the disease. This publication describes the different forms of scleroderma and provides information on their symptoms, diagnosis, and treatment, including what patients can do to help manage their disease and the problems associated with it. This publication also highlights research efforts into the understanding and treatment of scleroderma, many of which are supported by the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) and other components of the U.S. Department of Health and Human Services’ National Institutes of Health (NIH). If you have further questions after reading this publication, you may wish to discuss them with your doctor.

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What Is Scleroderma?
Derived from the Greek words “sklerosis,” meaning hardness, and “derma,” meaning skin, scleroderma literally means “hard skin.” Although 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. 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, such as the heart, lungs, and kidneys.

Scleroderma is called both a rheumatic (roo-MA-tik) disease and a connective tissue disease. The term rheumatic disease refers to a group of conditions characterized by inflammation or pain in the muscles, joints, or fibrous tissue. A connective tissue disease is one that affects tissues such as skin, tendons, and cartilage.

Scleroderma is also believed to be an autoimmune disease. In autoimmune diseases, the body's immune system turns against and damages its own tissues.

What Are the Different Types of Scleroderma?

The group of diseases we call scleroderma falls into two main classes: localized scleroderma and systemic sclerosis. (Localized diseases affect only certain parts of the body; systemic diseases can affect the whole body.) Both groups include subgroups (see chart). Although there are different ways these groups and subgroups may be broken down or referred to (and your doctor may use different terms from what you see here), the following is a common way of classifying these diseases:

**Types of Scleroderma**

![Diagram of Types of Scleroderma]

**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 refers to local patches of scleroderma. The first signs of the disease are reddish patches of skin that thicken into firm, oval-shaped areas. 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. 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 is characterized by a single line or band of thickened or abnormally colored skin. Usually, the line runs down an arm or leg, but in some people it runs down the forehead.

**Systemic Scleroderma (also known as Systemic Sclerosis)**

This is the term for the form of the disease that not only includes the skin, but also involves the tissues beneath, the blood vessels, and the major organs. Systemic sclerosis is typically broken down into *limited cutaneous scleroderma* and *diffuse cutaneous scleroderma*.

**Limited cutaneous scleroderma:** Limited cutaneous scleroderma typically comes on gradually and affects the skin only in certain areas: the fingers, hands, face, lower arms, and legs. People with limited disease often have all or some of the symptoms that some
doctors call CREST, which stands for the following:

- **Calcinosis (KAL-sin-OH-sis):** The formation of calcium deposits in the connective tissues, which can be detected by x ray.

- **Raynaud’s (ray-NOHZ) phenomenon:** A condition in which the small blood vessels of the hands or feet contract in response to cold or anxiety.

- **Esophageal (eh-SOFF-uh-GEE-ul) 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.

- **Sclerodactyly (SKLER-oh-DAK-till-ee):** Thick and tight skin on the fingers, resulting from deposits of excess collagen within skin layers.

- **Telangiectasia (tel-AN-jee-ek-TAY-zee-uh):** A condition caused by the swelling of tiny blood vessels, in which small red spots appear on the hands and face.

**Diffuse cutaneous scleroderma:** This condition typically comes on suddenly. Skin thickening begins in the hands and spreads 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, this condition can damage key organs such as the intestines, lungs, heart, and kidneys.

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 severe 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. 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.

**Genetic makeup:** Although 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:** Researchers suspect that exposure to some environmental factors may trigger scleroderma.

**Hormones:** Women develop most types of scleroderma more often than men. Scientists suspect that hormonal differences between women and men might play a part in the disease.

**Who Gets Scleroderma?**

Although scleroderma is more common in women, the disease also occurs in men and children. It affects people of all races and ethnic groups. However, there are some patterns by disease type. For example:

- **Localized forms** of scleroderma are more common in people of European descent than in African Americans. Morphea usually appears between the ages of 20 and 40, and linear scleroderma usually occurs in children or teenagers.

- **Systemic scleroderma,** whether limited or diffuse, typically occurs in people from 30 to 50 years old. It affects more women of African American than European descent.

Because scleroderma can be hard to diagnose and it overlaps with or resembles other diseases, scientists can only estimate how many cases there actually are. It is estimated that 49,000 adults in the United States have systemic sclerosis.¹


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 Is Scleroderma Diagnosed?**

Depending on your particular symptoms, a diagnosis of scleroderma may be made by:

- A **general internist**.
- A **dermatologist**, who specializes in treating diseases of the skin, hair, and nails.
- An **orthopaedist**, who treats bone and joint disorders.
- A **pulmonologist**, who is trained to treat lung problems.
- A **rheumatologist**, who specializes in treating musculoskeletal disorders and rheumatic diseases.

A diagnosis of scleroderma is based largely on the medical history and findings from the physical exam, questions about what has happened to you over time, and about any symptoms you may be experiencing. Once your doctor has taken a thorough medical history, he or she will perform a physical exam, which can help the doctor diagnose a certain form of scleroderma.

Finally, your doctor may order lab tests to help confirm a suspected diagnosis. The presence of certain antibodies is common in people with scleroderma, but having these antibodies does not confirm the presence of the disease.

In some cases, your doctor may order a skin biopsy (the surgical removal of a small sample of skin for microscopic examination) to aid in or help confirm a diagnosis. However, skin biopsies also have their limitations: biopsy results cannot distinguish between localized and systemic disease, for example.

Diagnosing scleroderma is easiest when a person has typical symptoms and rapid skin thickening. In other cases, a diagnosis may
take months, or even years, as the disease unfolds and reveals itself and as the doctor is able to rule out some other potential causes of the symptoms.

**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 doctor specializing in treatment of musculoskeletal disorders and rheumatic diseases). Your rheumatologist may refer you to other specialists, depending on the specific problems you are having. For example, you may see 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 such as 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 doctor. Others are things you can do on your own.

**Stiff, painful joints:** In diffuse systemic sclerosis, hand joints can stiffen because of hardened skin around the joints or inflammation within them. Other joints can also become stiff and swollen.

- Stretching exercises under the direction of a physical or occupational therapist are extremely important to prevent loss of joint motion.
- 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.

- Use medication as recommended by your doctor to help relieve joint or muscle pain.\(^2\) 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.

\(^2\) All medicines can have side effects. Some medicines and side effects are mentioned in this publication. Some side effects may be more severe than others. You should review the package insert that comes with your medicine and ask your health care provider or pharmacist if you have any questions about the possible side effects.

**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, 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 from 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. Otherwise, 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 caused by salivary gland damage speeds up tooth decay.

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 doctor about a saliva substitute or medications that can stimulate the flow of saliva.

**Gastrointestinal (GI) problems:** Systemic sclerosis 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.
- To keep stomach contents from backing up into the esophagus, stand or sit for at least an hour (preferably 2
or 3 hours) after eating.

- When it is time to sleep, keep the head of your bed raised using blocks.
- Avoid late-night meals, spicy or fatty foods, alcohol, and caffeine, which can aggravate GI distress.
- Eat moist, soft foods, and chew them well. 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 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, which can be a cause of diarrhea in some people with systemic sclerosis.

**Lung damage:** Virtually all people with systemic sclerosis have some loss of lung function. Some 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). Treatment for the two conditions is different:

- Pulmonary fibrosis may be treated with drugs that suppress the immune system, along with low doses of corticosteroids.
- Pulmonary hypertension may be treated with drugs that dilate the blood vessels or with newer medications that are prescribed specifically for treating pulmonary hypertension.

Regardless of your particular lung problem or its medical treatment, your role in the treatment process is essentially the same. To minimize lung complications, work closely with your medical team. Do the following:

- 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:** Common among people with scleroderma, heart problems include scarring and weakening of the heart (cardiomyopathy), inflamed heart muscle (myocarditis), and abnormal heartbeat (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:** Renal crisis is an uncommon but serious complication in patients with systemic sclerosis. Renal crisis results in severe uncontrolled high blood pressure, which can quickly lead to kidney failure. It’s very important that you take measures to identify and treat the hypertension as soon as it occurs. These are things you can do:

- Check your blood pressure regularly. You should also check it if you have any new or different symptoms such as a headache or shortness of breath. If your blood pressure is higher than usual, 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 have made scleroderma-related kidney failure a less threatening problem than it used to be. But for these drugs to work, you must take them as soon as the hypertension is present.

**Cosmetic problems:** Even if scleroderma doesn’t cause any lasting physical disability, its effects on the skin’s appearance—particularly on the face—can take their toll on your self-esteem. Fortunately, there are procedures to correct some of the cosmetic
problems scleroderma causes:

- The appearance of telangiectasias—small red spots on the hands and face caused by swelling of tiny blood vessels beneath the skin—may be reduced or even eliminated with the use of guided lasers.
- Facial changes of localized scleroderma—such as the *en coup de sabre* that may run down the forehead in people with linear scleroderma—may be corrected through cosmetic surgery. (However, such surgery is not appropriate for areas of the skin where the disease is active.)

Is Research Close to Finding a Cure?

No one can say for sure when—or if—a cure will be found. But research is providing the next best thing: better ways to treat symptoms, prevent organ damage, and improve the quality of life for people with scleroderma. Multidisciplinary research has also provided new clues for understanding the disease, which is an important step toward prevention and cure.

Studies of the immune system, genetics, cell biology, and molecular biology have helped reveal the causes of scleroderma, improve existing treatment, and create entirely new treatment approaches.

Some advances in the understanding or treatment of scleroderma include the following:

- Building on research that identified a gene associated with scleroderma in Oklahoma Choctaw Native Americans, scientists are using new technology to look for other genes associated with the disease's development and severity.
- The drug cyclophosphamide has been found effective in treating lung fibrosis. One study suggested that treating lung problems early with this immunosuppressive drug may help prevent further damage and increase chances of survival. Clinical trials assessing the effectiveness of other medications for lung fibrosis are
ongoing.

- ACE inhibitors are used increasingly for scleroderma-related kidney problems. ACE inhibitors have greatly reduced the risk of kidney failure in people with scleroderma.
- Several drugs are now available to treat pulmonary hypertension. Previously, pulmonary hypertension was associated with a poor outcome, but medications have increased the quality of life and life expectancy for people with this dangerous form of lung damage.

Other studies are examining the following:

- **The immune system:** Researchers are looking at the complex immune system trying to determine why the body creates antibodies against its own tissues. Identifying where and when the antibodies are created and how they function could lead researchers to find targets for new therapies. Researchers are already trying new therapies, including those that suppress specific parts of the immune system.

- **Genes:** Studies continue to find the genes that predispose a person to getting scleroderma. Those genes may be targets for future therapies. Researchers are also trying to find genes (and other indicators) that could tell doctors who will develop what type of scleroderma. This could someday help doctors identify who might benefit from specific therapies. Additionally, since genes are not the only factor involved in scleroderma, researchers are looking for environmental factors that might trigger the disease in those who are genetically predisposed.

- **Drug therapies:** Through basic studies of the molecular and genetic basis of scleroderma, scientists have indentified a large number of potential therapeutic targets. Other researchers are testing existing drugs to see if they are succesful in delaying cardiovascular changes.

- **Animal models:** Experiments using mice have proved valuable in past
research on scleroderma. Further research using existing strains of mice continues.

- **The fibrosis process:** Researchers are looking into the many factors that lead to fibrosis to see if they can block its development.

- **Cytokines:** Studies have shown that certain chemicals called cytokines, made from cells in the body, contribute to inflammation. Researchers are trying to determine how cytokines contribute to the disease process.

More information on research is available from the following websites:

- **NIH Clinical Research Trials and You** was designed to help people learn more about clinical trials, why they matter, and how to participate. Visitors to the website will find information about the basics of participating in a clinical trial, first-hand stories from clinical trial volunteers, explanations from researchers, and links on how to search for a trial or enroll in a research-matching program.

- **ClinicalTrials.gov** offers up-to-date information for locating federally and privately supported clinical trials for a wide range of diseases and conditions.

- **NIH RePORTER** is an electronic tool that allows users to search a repository of both intramural and extramural NIH-funded research projects from the past 25 years and access publications (since 1985) and patents resulting from NIH funding.

- **PubMed** is a free service of the U.S. National Library of Medicine that lets you search millions of journal citations and abstracts in the fields of medicine, nursing, dentistry, veterinary medicine, the health care system, and preclinical sciences.

**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 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.

For More Information

National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)
Information Clearinghouse
National Institutes of Health

1 AMS Circle
Bethesda, MD 20892-3675
Phone: 301-495-4484
Toll free: 877-22-NIAMS (877-226-4267)
TTY: 301-565-2966
Fax: 301-718-6366
Email: NIAMSinfo@mail.nih.gov
Website: https://www.niams.nih.gov

If you need more information about available resources in your language or another language, please visit our website or contact the NIAMS Information Clearinghouse at NIAMSinfo@mail.nih.gov.

Other Resources

American Academy of Dermatology
Website: http://www.aad.org

American College of Rheumatology
Website: http://www.rheumatology.org

Scleroderma Foundation
Website: http://www.scleroderma.org (con información en español)

Scleroderma Research Foundation
Website: http://www.srfcure.org

Arthritis Foundation
Website: http://www.arthritis.org

Key Words
**Antibodies.** Special proteins produced by the body's immune system. They recognize and help fight infectious agents, such as bacteria and other foreign substances that invade the body. The presence of certain antibodies in the blood can help to diagnose some diseases, including some forms of scleroderma.

**Autoimmune disease.** A disease in which the body's immune system turns against and damages its own tissues.

**Calcinosis.** The formation of calcium deposits in the connective tissues, which can be detected by x-ray. These deposits 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.

**Collagen.** A fabric-like material of fibrous threads that is a key component of the body's connective tissues. In scleroderma, either too much collagen is produced or it is produced in the wrong places, causing stiff and inflamed skin, blood vessels, and internal organs.

**Connective tissue.** Tissues such as skin, tendons, and cartilage that support and hold body parts together. The chief component of connective tissue is collagen.

**CREST syndrome.** An acronym for a collection of symptoms that occur to some degree in all people with systemic sclerosis. The symptoms are calcinosis, Raynaud's phenomenon, esophageal dysfunction, sclerodactyly, and telangiectasia. Because of the predominance of CREST symptoms in people with limited systemic sclerosis, some people use the term CREST syndrome when referring to that form of the disease.

**Fibrosis.** A condition marked by increased fibrous tissue that develops between the cells of various organs or tissues. It is a common feature of scleroderma and some other diseases. Fibrosis causes hardening or stiffening of tissues in the skin, joints, and internal organs.

**Pulmonary fibrosis.** Hardening or scarring of lung tissue because of excess collagen. Pulmonary fibrosis occurs in a small percentage of people with systemic sclerosis.

**Pulmonary hypertension.** Abnormally high
blood pressure in the arteries supplying the lungs that may be caused by a number of factors, including damage from fibrosis.

**Raynaud's phenomenon.** A condition in which the small blood vessels of the hands 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. Fingertip tissues may suffer damage, leading to ulcers, scars, or gangrene.

**Rheumatic.** An adjective used to describe a group of conditions characterized by inflammation or pain in the muscles, joints, and fibrous tissue. Rheumatic diseases or disorders can be related to autoimmunity or other causes.

**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.

**Telangiectasia.** A condition caused by the swelling of tiny blood vessels, in which small red spots appear on the hands and face. Although not painful, these red spots can create cosmetic problems.

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For Your Information

This publication contains information about medications used to treat the health condition discussed here. When this publication was developed, we included the most up-to-date (accurate) information available. Occasionally, new information on medication is released.

For updates and for any questions about any medications you are taking, please contact

U.S. Food and Drug Administration

Toll free: 888-INFO-FDA
(888-463-6332)
Website: http://www.fda.gov

For additional information on specific medications, visit Drugs@FDA at http://www.accessdata.fda.gov/scripts/cder/daf/. Drugs@FDA is a searchable catalog of FDA-approved drug products.

For updates and questions about statistics, please contact

Centers for Disease Control and Prevention, National Center for Health Statistics

Toll free: 800-232-4636
Website: http://www.cdc.gov/nchs

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