What is scleroderma?

Scleroderma is typically described as a rheumatic disease of the connective tissues. It is a chronic, degenerative disorder accompanied by widespread vascular deterioration and tissue loss. Scleroderma can be disfiguring, debilitating and deadly. In the most serious cases, the disease causes severe damage and severe complications for the body's digestive, respiratory and circulatory systems.

Pain, ranging in severity from uncomfortable to debilitating, is a common characteristic of the disease. Other common symptoms can include, but are not limited to the following:

- General fatigue
- Joint or bone aching
- Stiffness of hands and feet
- Skin discoloration
- Swallowing difficulties
- Skin thickening or tightness
- Dry mucus membranes
- Calcium deposits under the skin
- Unexplained ulcers on fingers or toes
- Raynaud's phenomenon (described below)

Scleroderma is not a single disease, but a set of related disorders involving a similar set of symptoms. This makes a concise definition – and diagnosis – difficult. All forms of scleroderma do, however, share the basic characteristics of fibrosis (an excessive accumulation of tissue) and inflammation.

Recent research suggests that scleroderma is not only a rheumatic disease, but also has a blood vessel component. Inflammation in the body's blood vessels leads to their narrowing. Further damage from the inflammation and impact of the increased blood pressure can lead to destruction of smaller arteries. As a result, or perhaps as a parallel disease process, fibrosis takes place. This process can have devastating implications and the heart and lungs can suffer a significant loss in efficiency. Pulmonary arterial hypertension (PAH) is the leading cause of death in scleroderma patients.
The first documented account of scleroderma was by Dr. Carlo Curzio of Naples in 1754, who described his patient as a 17-year-old Neapolitan woman afflicted by a disease of “extraordinary nature.” “Her complaint,” wrote Dr. Curzio, “was an excessive tension and hardness of her skin over all her body, by which she found herself so bound and straightened that she could hardly move her limbs.” Nor could she fully close her eyelids or open her mouth, due to “the firmness of the skin and membrane.”

The fibrosis witnessed by Dr. Curzio is the most noticeable feature of the disease. In fact, the word scleroderma is Greek for “hard skin,” although it is much more than that, often affecting the internal organs with life-threatening consequences.

Hundreds of thousands of people suffer from the various types of scleroderma, and as many as 10,000 patients die every year from the most serious form of the disease. Eighty percent of those afflicted with the disease are women, though scleroderma also strikes men and children across all ages and ethnic boundaries. Unfortunately, the disease typically strikes in the prime of patients' lives, when they are 30-50 years old.

The cause of scleroderma is unknown. It is not contagious and it is rare for direct family members to manifest the disease. However, there is evidence of a genetic predisposition to scleroderma. Some scleroderma cases have been linked to environmental factors such as silica dust, organic solvents, and certain drugs, though the cause for most cases of scleroderma is still not known.

Significant progress has been made in managing the symptoms and some of the most serious complication of scleroderma, so that people with the disease are living longer, fuller lives. Still, no therapies yet exist to halt or reverse the disease process. Patients continue to experience pain, deterioration and debilitation – and many patients die.

The Scleroderma Research Foundation is devoting every possible resource to solving the mysteries of scleroderma. A collaborative approach to research is opening doors to therapeutic development and ultimately, a cure.

Types of scleroderma
There are two major types of scleroderma—localized and systemic. Localized scleroderma affects only the skin and is, generally, not as destructive or debilitating as systemic scleroderma, which is a multi-system disease often involving the skin, blood vessels, heart, lungs, kidneys and other major organs.

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**Localized Scleroderma**

The skin is typically the only organ involved in localized scleroderma. In extreme cases, however, the muscle below is also affected by a hardening of the tissues. Because internal organs are not affected by localized scleroderma, it is not a life-threatening illness and can be considered a less serious health threat than other forms of the disease. Symptoms of localized scleroderma will, in many cases, improve or go away on their own over time, but there is frequently permanent damage in the form of residual scar tissue. For some patients, localized scleroderma can be disfiguring and painful. In rare cases, it can be disabling. There are three major sub-types of localized scleroderma:

**Localized Morphea**

Morphea (mor-EE-ah) is derived from the Greek word “morfi,” meaning shape or structure. With localized morphea, the body shows one or several patches of scleroderma. Symptoms include reddish patches of inflamed and discolored skin, usually on the chest or back, but sometimes on the face, arms and legs. These oval-shaped patches are limited to certain areas on the body and range in size from a half-inch to approximately 12 inches in diameter. Localized morphea is further pronounced by the absence of sweat glands and very little, if any, hair growth in affected areas.

**Generalized Morphea**

Generalized morphea is a more severe, although less common, condition than localized morphea. Large dark patches of thick, tight skin form on the trunk, arms and legs, and can be so extensive that they merge into one another. These widespread plaques closely resemble oversized patches of localized morphea, but are usually ivory-colored and sometimes have a distinct violet-colored border. Patients with generalized morphea do not suffer from the internal life-threatening features of systemic sclerosis, but the disease can result in severe scars and physical disability.
Generalized morphea has been known to appear on areas of the skin treated by radiation therapy.

In many cases, both localized and generalized morphea improve spontaneously over time (typically 3 through 5 years); however, patients are often left with patches of darkened or discolored skin and, in rare cases, muscle damage.

**Linear Scleroderma**

Linear scleroderma, also known as bilateral scleroderma, is characterized by a highly visible band or bands of thick, tight skin on the arms, legs, chest, stomach, back, buttocks or face. It is often considered a pediatric disease as it usually manifests itself within the first 10 years of life.

When affecting a limb, linear scleroderma can cause arms and legs to develop at noticeably different rates. It is not uncommon for the disease to extend down to the bones and muscles underneath it, thus damaging their structure, limiting motion of affected joints, and interfering with normal growth.

When linear scleroderma occurs on the face, it is often referred to as en coup de saber (en-KOO-de-sob), a French term for “a strike of the sword,” due to its appearance. Linear scleroderma may be disfiguring, but is not life threatening and rarely progresses to systemic scleroderma.

**Systemic Scleroderma (Systemic Sclerosis)**

Systemic scleroderma, also known as systemic sclerosis, is a multi-system disease with numerous manifestations that can affect not only the skin, but also underlying blood vessels, muscles and joints, as well as the gastrointestinal tract, kidneys, lungs and heart. There are two subsets of systemic scleroderma, limited and diffuse – both are characterized by vascular injury, as well as severe inflammation and fibrosis.

Systemic scleroderma causes a loss of circulation that can result in painful skin ulcers occurring on fingers, toes and other extremities. Joint pain caused by inflammation and swelling can be chronic. Loss of flexibility in the hands and severe skin tightening can impair the use of fingers and toes. The skin of the face may also tighten and harden, especially around the mouth,
making eating and drinking difficult.

Systemic scleroderma (both limited and diffuse) is the most dangerous form of the disease and the primary focus of research funded by the Scleroderma Research Foundation.

It is estimated that approximately 90% of systemic scleroderma patients experience Raynaud's phenomenon as a primary symptom. Named after the French physician who discovered it more than a century ago, Raynaud's is a condition in which the small blood vessels of the hands and, sometimes, feet contract in response to cold or stress. The process makes the hands/feet extremely sensitive to cold and causes their pigmentation to blanch from white to blue. As blood flow returns, they become red. With scleroderma patients, tissues in fingertips, especially the capillaries, may suffer damage. This damage can lead to ulcers, scars, or, in severe cases, gangrene.

It should be noted that Raynaud's phenomenon is quite common, affecting approximately 1 of every 20 people in the United States. Research suggests that only 5 – 20% of people with Raynaud's phenomenon will go on to develop a form of systemic scleroderma.

**Limited Scleroderma**

Systemic limited scleroderma affects approximately two-thirds of the systemic population. Although the progression is typically more slow-moving, the disease can still be life-threatening. It is imperative to treat these symptoms and carefully monitor for end-organ damage. Chronic pain, loss of mobility and disfigurement are possible. There is also the potential for serious complications related to the esophagus and other parts of the gastrointestinal tract, as well as pulmonary complications leading in some cases to dangerously high blood pressure. Systemic-limited scleroderma is often referred to as CREST, an acronym for several of the most prominent symptoms:

- **Calcinosi**s: Hard, painful calcium deposits in the skin
- Raynaud's phenomenon: Blanching of feet and hands from white, to blue to red in response to cold or stress
- **Esophageal dysfunction**: Problems with swallowing caused by internal scarring. The scarring is caused by the collagen build-up.
- **Sclerodactyly**: Tightening of the skin on the fingers and toes, progressing to the hands and face.
- **Telangiectasia**: Small red spots on the fingers, palms, forearms, face and lips

**Diffuse Scleroderma**
Diffuse scleroderma is typically characterized by an early internal inflammatory phase. Also, unlike limited scleroderma, the onset of symptoms that significantly impact quality of life can be rapid and severe. In most cases of diffuse scleroderma, there is extensive skin fibrosis covering large parts of the body and extremities. Even cases without significant skin involvement can be life-threatening, depending on the type and magnitude of organ damage.

Problems of the digestive tract can make eating, nutrient absorption and elimination difficult. The most serious complications of diffuse scleroderma involve the kidneys, lungs and heart and may include renal failure, pulmonary arterial hypertension (PAH) and the potential for heart failure. PAH is the leading cause of death among scleroderma patients.

People with diffuse scleroderma are often chronically tired, have a diminished appetite, show weight loss, and suffer from joint swelling and pain. Changes in the skin can include swelling, tightening and chronic itchiness.

It is not uncommon for people with diffuse scleroderma to enter a phase where disease progression stabilizes for varying lengths of time. This stabilizing phase often begins within three to five years after onset and can last for varying amounts of time.

As with all forms of the disease, there is no cure for diffuse scleroderma. Although the disease may continue to lie dormant for several years, there can be no certainty as to whether symptoms will improve, remain stagnant or worsen.

Early diagnosis, symptomatic treatment and frequent visits to a physician for monitoring are critically important for diffuse scleroderma patients.

**Sine Sclerosis**

Sine (SEEN-ay) sclerosis is a term used by many physicians to describe systemic scleroderma without the characteristic skin involvement. In fact, the word “sine” is Latin for “without.” Patients with sine sclerosis may have symptoms of either limited or diffuse scleroderma, causing damage to the internal organs. Because there are no external manifestations of the disease, it can be difficult to diagnose. As with other forms of systemic sclerosis, however, the presence of Raynaud’s phenomenon and positive autoantibodies can be useful indicators. Complications of the internal organ systems can be serious and often life threatening.