

I have scleroderma and need to know more....

## **WHAT IS SCLERODERMA?**

Scleroderma, or systemic sclerosis, is a chronic connective tissue disease generally classified as one of the autoimmune rheumatic diseases. The word “scleroderma” comes from two Greek words: “sclera” meaning hard, and “derma”, meaning skin. Hardening of the skin is one of the most visible manifestations of the disease. The disease has been called “progressive systemic sclerosis,” but the use of that term has been discouraged since it has been found that scleroderma is not necessarily progressive.

Scleroderma is a disease whose systems may be visible, as is the case when the skin is affected, or the symptoms may be invisible, as when internal organs are affected.

## **WHAT SCLERODERMA IS NOT?**

Scleroderma is **not** contagious, it is **not** infectious, it is **not** cancerous or malignant, and it is usually **not** hereditary.

## **HOW SERIOUS IS SCLERODERMA?**

Any chronic disease can be serious. The symptoms of scleroderma vary greatly from individual to individual, and the effects of scleroderma can range from very mild to life-threatening. The seriousness will depend on what parts of the body are affected and the extent to which they are affected. A mild case can become more serious if not properly treated. Prompt and proper diagnosis and treatment by qualified physicians may minimize the symptoms of scleroderma and lessen the chance for irreversible damage.

## **WHAT FACTORS CONTRIBUTE TO SCLERODERMA?**

The cause of scleroderma continues to be unknown. Researchers are working hard to determine what factors contribute to where scleroderma originates.

## **WHERE CAN I SEEK TREATMENT?**

If you are looking for clinical trials or want to seek treatment for your scleroderma please contact:

### **University of Michigan Health System Rheumatology/Scleroderma Clinics**

1500 E. Medical Center Drive  
3rd Level Taubman Center - Clinic Areas A & C  
Ann Arbor, MI 48109-0370

### **Richard Martin MD**

West Michigan Rheumatology  
1155 East Paris Avenue #100  
Grand Rapid, MI 49546-8368  
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**Delfin Santos MD**

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Traverse City, MI 49684-3258  
(231) 935-0550

**John Kolstoe MD**

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East Lansing, MI 48823-7787

**John Howland MD**

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2110 16<sup>th</sup> Street Suite 9  
Bay City, MI 48708-7609  
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## WHO DEVELOPS SCLERODERMA, AND WHEN?

It is estimated that there are over 300,000 persons with scleroderma in the United States, including 80,000 to 100,000 with the systemic form, and the rest with the localized form of scleroderma-spectrum disease features. International incidence is unknown but it has been reported worldwide. About three to four times more women than men develop the disease. Scleroderma can develop and is found in every age group from infants to the elderly but its onset is most frequent between the ages of 25 to 55.

Factors other than sex, such as race and ethnic background, may influence the risk of getting scleroderma, the age of onset, and the pattern or severity of internal organ involvement. The reasons for this are not clear. Although scleroderma is not directly inherited, some scientists feel there is a slight predisposition to it in families with a history of rheumatic disease.

## WHAT CAUSES SCLERODERMA?

The exact cause or causes of scleroderma are still unknown, but scientists and medical investigators in a wide variety of fields are working hard to make those determinations. What is known about the disease process in scleroderma is that it involves three features:

1. an overproduction of collagen

2. an autoimmune process
3. blood vessel damage

Collagen is the major protein portion of the connective tissue of the body, which includes the skin, joints, and tendons, and parts of internal organs. The structure of collagen is made up of tiny fibers, which are woven together much like the threads forming a piece of cloth. When there is an overproduction of collagen, thickening and hardening of the affected area takes place and often interfering with the normal functioning of those parts.

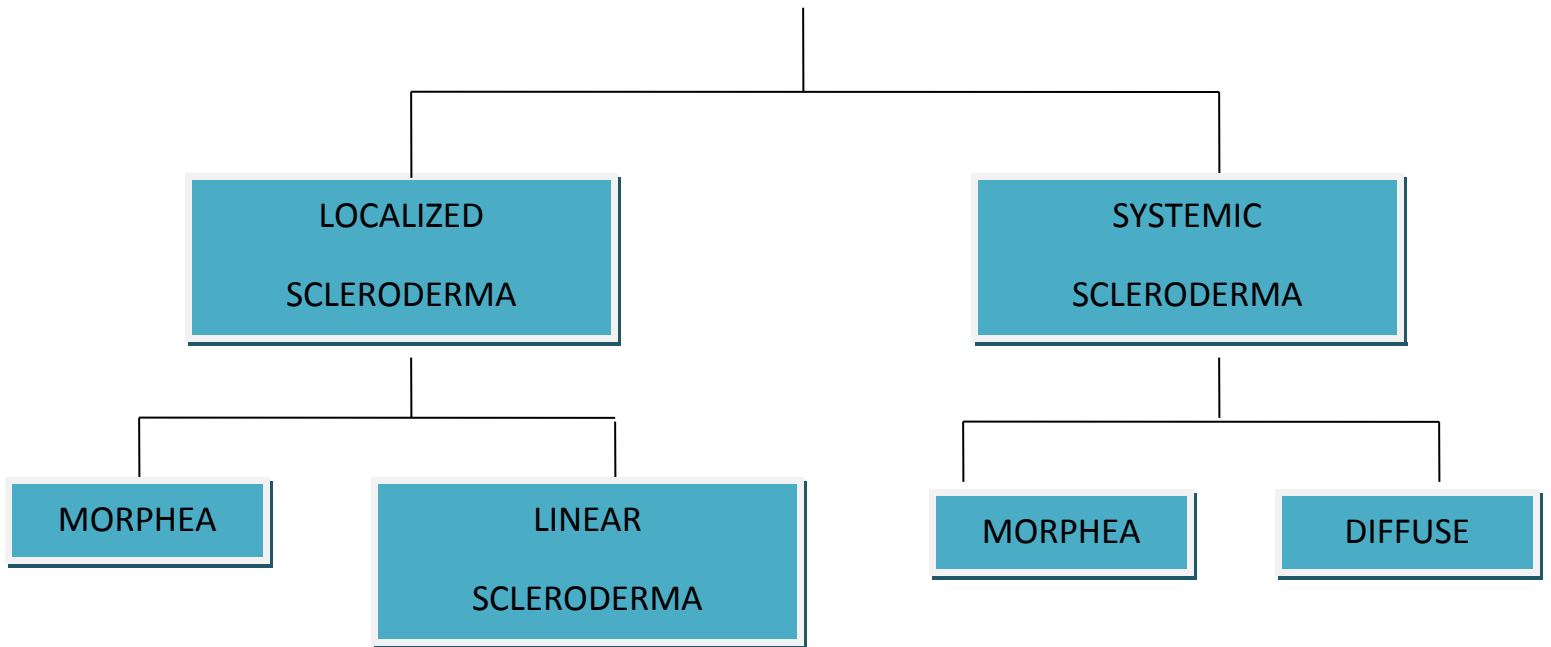
There are several theories about how collagen is overproduced. The “autoimmune theory” suggests that the body’s own immune system plays a part. Normally, the body’s immune system produces chemical signals in the blood called cytokines, which coordinate the body’s defense against bacteria, viruses, and other foreign invaders. In addition, some cytokines help to repair wounds by stimulating collagen production that forms a scar. There are a number of theories on the way in which the immune system is activated inappropriately, causing abnormal levels of cytokines to be produced. These, in turn, mount an attack not against a foreign invader but against the body’s own healthy tissues, stimulating an overproduction of collagen.

Another theory, the “vascular theory” concerns blood vessels. Damage to the blood vessels, especially the small ones, is typical in scleroderma. Injury to blood vessels causes them to constrict and stiffen and leads them to overreact to cold and stress. These reactions can cause further damage to the vessels themselves and to the organs, which they supply. There may also be a connection between the buildup of excess collagen and blood vessels, the processes which take place, and their significance for prevention and treatment.

## **ARE THERE DIFFERENT FORMS OF SCLERODERMA?**

There are two major classifications of scleroderma: **localized scleroderma** and **systemic sclerosis (SSc)**. Other forms or sub classifications, each with its own characteristics and prognosis, may be identified through future research.

SCLERODERMA



## LOCALIZED SCLERODERMA

The changes, which occur in localized scleroderma, are usually found in only a few places on the skin or in the muscles, and only rarely spread elsewhere. Generally, localized scleroderma is relatively mild. The internal organs are usually not affected, and persons with localized scleroderma rarely develop systemic scleroderma. Some laboratory abnormalities commonly seen in systemic scleroderma are frequently absent in the localized form.

## MORPHEA

Morphea is a form of localized scleroderma characterized by waxy patches on the skin of varying sizes, shapes, and coloration. The skin underlying the patches may thicken. The patches may enlarge or shrink and may often disappear spontaneously. Morphea usually appears between the ages of 20 and 50, but is often seen in young children.

## LINEAR SCLERODERMA

Linear scleroderma is a form of localized scleroderma which frequently starts as a streak or line of hardened, waxy skin on an arm or on the forehead. Sometimes it forms a long crease on the head or neck, referred to as **en coup de sabre** because of its resemblance to a saber or sword wound. Linear scleroderma tends to involve deeper layers of the skin as well as the surface layers and sometimes affects the motion of the joints which lie underneath. Linear scleroderma usually develops in childhood. In children the growth of involved limbs may be affected.

## SYSTEMIC SCLERODERMA (SYSTEMIC SCLEROSIS)

The changes occurring in systemic scleroderma may affect the connective tissue in many parts of the body. Systemic scleroderma can involve the skin, esophagus, gastrointestinal tract (stomach and bowels), lungs, kidneys, heart, and other internal organs. It can also affect blood vessels, muscles, and joints. The tissues of involved organs become hard and fibrous, causing them to function less efficiently. The term **systemic sclerosis** indicates that “sclerosis” may occur in the internal systems of the body. There are two major recognized patterns that the illness can take **diffuse** or **limited** disease. In **diffuse scleroderma**, skin thickening occurs more rapidly and involves more skin areas than in limited disease. In addition, people with diffuse scleroderma have a higher risk of developing “sclerosis” or fibrous hardening of the internal organs.

About half of the patients have a slower and more benign illness called **limited scleroderma**. In limited scleroderma, skin thickening is less widespread, typically confined to the fingers, hands, and face, and develops slowly over years. Although internal problems occur, they are less frequent and tend to be less severe than in diffuse scleroderma, and are usually delayed in onset for many years. However, persons with limited scleroderma can develop pulmonary hypertension, a condition in which the blood vessels of the lung become narrow, leading to impaired blood flow through the lungs.

## LIMITED SCLERODERMA

Limited scleroderma is sometimes called the **CREST syndrome**. CREST stands for the initial letters of five features:

Calcinosis

Raynaud’s phenomenon

Esophageal dysfunction

Sclerodactyly

Telangiectasia

To further complicate the terminology, some people with diffuse disease will go on to develop calcinosis and telangiectasias so that they also have the features of CREST.

Although most patients can be classified as having either diffuse or limited disease, different people may have different symptoms and different combination of symptoms of the illness.

## HOW IS SCLERODERMA DIAGNOSED?

Diagnosis of scleroderma may be very difficult, particularly in its early stages. Many of its symptoms are common to, or may overlap with, those of other diseases, especially other autoimmune connective-tissue diseases such as rheumatoid arthritis and lupus. Different symptoms may develop in stages over a very long period of time, and few persons with scleroderma experience exactly the same set of symptoms and effects.

While scleroderma can often be suspected from its more visible symptoms, no single test can prove its presence. The diagnosis is usually made by physicians with extensive experience in the treatment of scleroderma by a combination of the following:

- Medical history including past present symptoms
- Thorough physical examination
- Findings from a variety of laboratory tests and other studies

In making the diagnosis, it is important not only to confirm the presence of scleroderma, but also to determine its extent and severity, particularly with regard to the involvement of internal organs.

Diffuse and limited scleroderma can sometimes be differentiated by the presence of different antibodies, called **anti-nuclear antibodies (ANA)** in the blood. For example, **anti - Scl-70** is frequently associated with diffuse scleroderma whereas **anti-centromere** is usually indicative of limited scleroderma.

## WHAT ARE THE SYMPTOMS OF SYSTEMIC SCLERODERMA?

Scleroderma is a complex disease with many possible symptoms that can affect many parts of the body. Most people develop a few of the symptoms mentioned. Each patient is different in number of symptoms and severity. Typically, the symptoms may also vary over time with periods of improvement and worsening.

Even though scleroderma is not curable, many of the symptoms can be improved with medication or lifestyle changes.

- **Raynaud's phenomenon**- most common early symptom of systemic scleroderma. It is present at one or another in about 90% of patients. It is most obvious in the fingers and toes but can also involve the ears, nose and tip of the tongue. The blood vessels constrict or narrow in response to cold or to emotional upset and stress. The resulting disturbance in circulation of the blood causes a series of color changes in the skin: white, blanched, or pale when circulation is reduced; blue as the affected part loses oxygen from decreased blood flow; and then red or flushed as blood flow returns and the part re-warms. Finally, as the attack subsides and the circulation returns to normal, usual skin color is restored. In the "white" or "blue" stages, sensations such as tingling, numbness and coldness may be felt. In the "red" stage, a feeling of warmth, burning, or throbbing may be noted.
- **Swelling or puffiness of the hands**-swelling is another typical early symptom of scleroderma, and this may be especially noticeable upon awakening because of muscle inactivity overnight. The skin of the fingers may look full and sausage like, making it difficult to close the hand into a fist.

- **Pain and stiffness of the joints**-symptoms of pain, stiffness, swelling, warmth or tenderness may accompany the arthritis-like joint inflammation which frequently occurs in scleroderma. Muscle pain and weakness are other important symptoms.
- **Skin thickening**- Hardening and thickening of the skin give scleroderma its name (“hard skin”). There are no proven treatments as yet to prevent or alter the course of the skin changes in scleroderma. Many medications and treatments are being tested. The skin sometimes softens spontaneously over time.
- **Skin ulcerations**-Sores, especially on the fingertips are a common symptom of systemic scleroderma. They may be very slow or difficult to heal because of poor circulation. These sores or ulcerations may also occur on the knuckles, elbows, toes, or other sites of the body where the skin is especially tight or stretched. The affected area should be kept warm to increase blood flow, and scrupulously clean to avoid infection.
- **Calcinosis**-This condition is characterized by deposits of calcium in the skin which may be painful. The calcium deposits may occur just below the skin surface in the form of hard lumps or nodules. They may break through the skin, becoming visible as chalky white material, and may become infected. Care should be taken not to bump or injure affected areas.
- **Telangiectasis**-This abnormality consists of the dilation of small blood vessels near the surface of the skin, which become visible as small red spots, usually on the fingers, palms, face and lips. The spots usually fade with pressure, but turn red again when the pressure is released. These spots are generally not harmful. Special cosmetics may be used to mask the spots or to reduce their visibility.
- **Dry skin**-Excessive dryness of the skin may lead to skin breakdown and ulcerations. Excessive bathing and handwashing should be avoided, and rubber gloves worn to avoid direct contact with household detergents. Keeping the skin moist and well lubricated is important to avoid complication from dry skin.
- **Itchy skin**-If moisturizing creams do not work; the physician may prescribe a topical cortisone cream to be rubbed on the skin to relieve itching. Antihistamines have been effective for some people.
- **Other skin symptoms**-There may be a decrease in hair over affected areas of the skin, as well as a decrease in the ability to perspire. In addition, there may be an increase in pigment, (which looks like a skin tan) or a spotty loss of pigment.
- **Sclerodactyly and joint contractures**- Sclerodactyly means simply “hard skin of the digits”; that is, of the fingers and toes. It generally occurs after initial swelling has subsided. It is characterized by shiny, tight skin of the fingers. Affected digits may be difficult to move, and they may become fixed in

a bent or flexed position called a “contracture” or a “flexion contracture”. Tightening and hardening of the skin and of the tissues surrounding the joints can cause decreased motion of the wrists, elbows, and other joints.

- **Digestive system and gastrointestinal tract problems-** Persons with systemic scleroderma may develop abnormalities of the digestive and gastrointestinal tract from the mouth to the anal canal. The overproduction of collagen typical of scleroderma can cause thickening and fibrosis (or scarring) of the tissues. This can result in weakening muscles and lead to abnormally slow movement of food in the digestive process.
- **Esophageal dysfunction-** Food travels from the mouth and throat into the stomach through a tube called the esophagus. Normally, the lower esophageal sphincter or valve acts as a gate which opens to permit food from coming back up. In systemic scleroderma, the gate does not close properly and the result is a backwash of acid and a burning sensation (“heartburn”) as food and acid return into the esophagus. The acid may also injure the lining of the lower portion of the esophagus, causing scarring and a narrowing (“stricture”) of the tube.
- **Swallowing difficulties-** Abnormally slow movement of food and narrowing of the esophagus may cause swallowing difficulties. Eating slowly and chewing thoroughly are important. Swallowing and digesting are made easier by eating softer foods or by preparing foods in a blender. Avoid foods which tend to stick in your throat.
- **Diarrhea-** In systemic scleroderma, there can be damage to the muscles of the small bowel (small intestine). The weakened muscles do not work effectively to push food through the bowel. Simply put, things “sit” rather than move well. One consequence can be an overgrowth of bacteria, leading to diarrhea. There may also be a bloated, distended feeling and some pain if the bowel is stretched.
- **Constipation-**Weak or scarred muscles in the colon wall make it difficult for the bowel to work well, resulting in constipation or other abnormalities of the colon. Maintaining a diet high in fiber, and drinking at least six to eight glasses of fluid daily, especially water will prevent constipation. Fresh fruits and vegetables are natural laxatives.
- **Sjogren’s syndrome-** Sjogren’s syndrome is characterized by a decrease in secretions of the tear glands and the salivary glands, which provide lubrication for the eyes and mouth. The unusual dryness of the eyes resulting from this condition can lead to serious irritation and inflammation. Excessive dryness of the mouth may lead to difficulties in swallowing and in speaking, a pronounced increased in tooth decay and cavities, and a reduced sense of taste. The lack of secretions in Sjogren’s syndrome may also involve the vagina and other areas of the body. Regular visits to the ophthalmologist are important and the mouth should be kept as well lubricated as possible by sipping fluids throughout the day. Chewing sugar free gum or sucking sugar free candy to stimulate salivary activity can be helpful. It is also a good idea to keep a water bottle handy during the day.

- **Lung involvement-** Lung involvement in systemic scleroderma may be caused by multiple factors. Build up of collagen thickens lung tissues and causes fibrosis or scarring, making the transport of oxygen into the bloodstream more difficult. Pulmonary arterial hypertension (PAH) a state of increased resistance to blood flow through the lungs, can result from damage to blood vessels and may lead to additional strain on the heart resulting in heart failure. Respiratory muscle weakness may decrease lung function. Symptoms of lung involvement include:
  - **Shortness of breath**
  - **A decreased tolerance for exercise**
  - **A persistent cough**

It is important for the person with scleroderma to take whatever measures are within his or her control to avoid further damage to the lungs. **It is essential to avoid smoking, a major cause of lung disease.**

- **Heart involvement-** If the heart muscle becomes thickened and fibrous scar tissue accumulates, the force of heart contractions may be decreased, which may ultimately result in heart failure. Spasm of the coronary arteries may cause chest pain and, rarely lead to a heart attack. The spasm appears similar to that involving the fingers in Raynaud's phenomenon. Inflammation of the outer heart lining may cause pain and accumulation of fluid around the heart. An irregular heartbeat may also be experienced.
- **Oral, facial and dental problems-** Persons with scleroderma may experience a general tightening of skin over the face. The opening of the mouth may be decreased in size, making lip and mouth movements as well as oral hygiene difficult. The best approach to treatment is by means of facial grimacing and mouth stretching exercised, including the use of "oral augmentation" props inserted between the upper and lower teeth. Take caution to avoid further damage when using these maneuvers.

Preventive dental care through regular flossing and brushing of the teeth and gums is very important, as they are regular dental visits for oral health and for early detection and prompt correction of any abnormalities.

- **Kidney involvement-** Kidney or renal involvement in systemic scleroderma may be mild or very serious in nature. Early signs of kidney involvement may include mild hypertension (high blood pressure), protein in the urine, and blood test abnormalities, "Renal crisis," a highly dangerous complication of systemic scleroderma, may occur quite suddenly. Its most important warning signal is an abrupt rise in blood pressure. Other symptoms are:
  - **Headache**
  - **Visual disturbances**
  - **Shortness of breath**
  - **Chest pain or discomfort**
  - **Mental confusion**

Unless treated promptly, renal crisis leads to kidney failure, a condition in which the kidneys lose their ability to eliminate waste products from the body.

## HINTS

- Decrease the amount of sodium in your diet. Stay away from salts, soy sauces, commercially canned goods like soups, beans and sauces. You can still add flavor to your foods by using herbs and (salt-free) spices.
- Make sure you get adequate fluids, especially water, throughout the day.

**Non-specific symptoms-** The person with systemic scleroderma may experience a variety of non-specific symptoms including:

- Fatigue –ranging from mild to severe
- Lack of energy
- Generalized weakness
- Weight loss
- Vague aching of muscles, joints or bones
- Treatments or medications recommended by the physician will depend on his/her evaluation of the causes of these symptoms.

## HOW DO I COPE WITH MY WITH MY DIAGNOSIS?

Any chronic disease is life changing. Symptoms demand your attention. You have to adjust your schedule to accommodate medications, doctor appointments and treatments of various kinds. You need to arrange for rest and relaxation and do things more slowly than you use to. You need to balance work and rest. You will have to confront changes in your body and the way you see yourself as well as the way others are used to seeing you. The people around you need to adjust as well. They live with the changes of your disease, too even though they don't experience your symptoms.

## HINTS TO MAKE LIFE EASIER

- Take notes about what your doctor tell you. If you have questions later after you have absorbed the information, you have absorbed the information, you can refer to those notes and ask questions. If you can take a friend, a family member and/or a tape recorder along so the information can be reviewed later. This reduces the chance that worry will inhibit hearing the information. It may also help your family member understand the disease.
- Share your worry with people you trust, if this method of reducing anxiety works for you. Spouses and partners can be especially eager to know how you are being affected.
- Recognize that this can be a difficult time. Just knowing that fact might help you balance some of the challenges. Being proactive in doing things can channel stress and anxiety in positive action.

- It has been reported that people who are struggling with a set of problems new to them often drop fun activities in their lives because they don't have the energy. Remember to retain the activities you enjoy and are capable of doing. Learn to delegate other activities to those who care.

## **THE EMOTIONS**

### **Denial**

Once testing has occurred and a diagnosis is made the next step is to determine what you want to do about it. Sometimes when people get upsetting news they feel disbelief and refuse to acknowledge that anything is seriously wrong. This can make people feel better for a short time, but the reality of the situation will eventually become undeniable.

When you keep denying that something is wrong, you cannot move forward into helpful activity. One way to absorb and deal with the news of your diagnosis is to give yourself some time to think and accept what has happened. Listen to your favorite music, watch videos that make you feel good, bask in the sun in a garden, or lie in a warm tub and think.

## **HINTS**

- Think about your resources-people who care about you, your talents and abilities, available information that can help you understand what is happening to you, crises you've confronted in the past and conquered.
- Think about your options: learn about your illness and how you can live and function with it, meet others with scleroderma who are functioning well, talk to someone who can help you confront your illness and support you while you do it.
- Think about the possible consequence of not tending to yourself-increasing worry, pain, disability and depression.

## **ANGER AND DEPRESSION**

After you've had time to understand the implication of the diagnosis, you may get angry and depressed at the same time. This is the next phase of moving toward an acknowledgement that changes are occurring. This isn't an easy phase to move through-not for you, and not for those close to you. But it is necessary, and as long as you know you're going through it, go through it with your eyes open.

Anger can be a very energizing emotion. It's during this time that patients typically do the following:

- Find and join a support group- MI Chapter has several all over the state (check our web site for locations)
- Seek out a physician who knows how to treat scleroderma
- Search libraries and the Internet for information they feel might be helpful (be careful to seek accurate information)

Depression can slow people down. Patients describe feeling negative about everything, an increase in physical symptoms and helpless/hopeless feelings, as well as a decrease in energy.

Fearfulness is also common. This is the phase during which a counselor could be useful. Select someone who has experience helping people with chronic disease. This person can be objective and help you to get angry/depressed feelings out in the open in a safe place-not on the job, directed at your loved ones or

yourself. Your physician might also prescribe a medication that will help your energy level and even treat the physical pain.

## **BARGAINING AND ACCEPTANCE**

While trying to cope with anger and depression, some people find themselves bargaining. This stage may take the form of praying, “God, if you’ll only take this from me, I promise...” or being willing to take medicine with uncomfortable side effects if only it might “cure” the disease. Some go to herbalists, chiropractors and acupuncturists in hopes of a “cure”. These are all ways of bargaining and another way of denying the reality of the disease.

## **ACCEPTANCE**

So, how does a person come to accept scleroderma? There isn’t any one way that patients reach that stage; some never do and some do so in part. Patients who cope well with their disease have these things in common:

- They have discovered the hope and strengths they have inside
- Have found people to help them along their way (friends, relatives, loved ones, support groups, counselors, faith groups, or health care providers)
- Have learned to help others by getting involved in scleroderma organizations, educating their friends and the public and helping other patients through difficult times

The most important underlying factor seems to be that patients who successfully cope with scleroderma have discovered that their lives are worth the fight.

Resources used:

*Successful Living with Scleroderma: A Balance Strategy Workbook*, by Robert Phillips, PhD., Director Center for Coping

*Understanding and Managing Scleroderma* by Maureen D. Mayes, M.D. , M.P.H. and Khanh T. Ho M.D.