



Localized Scleroderma

INTRODUCTION

Scleroderma means “hard skin.” The term “localized scleroderma” as a primary diagnosis refers to the fact that this group of conditions is “localized” to the skin, with no internal organ involvement. Sometimes, the term “morphea” is used interchangeably with localized scleroderma, creating some confusion. Morphea, in fact, is only one particular type of localized scleroderma. It is important to understand that localized scleroderma is different from the form of scleroderma which affects internal organs, called systemic sclerosis or, often incorrectly stated, as systemic scleroderma. Localized scleroderma typically only affects the skin, although in some cases the underlying muscle and tissue may be involved (subcutaneous morphea). Localized scleroderma is not a fatal disease, but quality of life is often adversely affected because of changes in the appearance of the skin, the occurrence of joint contractures that affect movement, and, rarely, serious deformities of the face and extremities.

The goal of this pamphlet is to inform you about the nature, progress and complications of localized scleroderma, and to discuss current treatment options. Because localized scleroderma often affects children, a major goal of the pamphlet is to also alleviate fears about this condition and to explain how it differs from systemic sclerosis, which is quite different and affects internal organs and the skin.

What is Localized Scleroderma?

Localized scleroderma is characterized by inflammation and thickening of the skin from excessive collagen deposition. Collagen is a protein normally present in our skin. It provides structural support. However, when too much

collagen is made, the skin becomes stiff and hard.

What Causes Localized Scleroderma?

The cause is unknown. It is not infectious and cannot be spread by touch or contact with the patient. It is not hereditary; however, in rare instances similar problems may be present in relatives. It is thought to be an autoimmune disease but, other than the presence of blood autoantibodies (confusingly similar to those with some internal diseases), patients have no other known or profound defect in the immune system.

Types of Localized Scleroderma

Names and terminology are widely varied and cause a great deal of confusion in localized scleroderma. Patients are often told they have “scleroderma,” which may frighten them. A newly diagnosed patient may think they have systemic sclerosis and will develop internal organ involvement. This is not true.

There are four main types of localized scleroderma. Each type is characterized by the shape and amount of affected skin. The four types are: A) morphea B) generalized morphea C) linear scleroderma and D) en coup de sabre.

A) Morphea – This is the most common type of localized scleroderma. It presents as one or a few (3-4) patches of skin thickening with different degrees of pigment changes. Some areas are dark while others are lighter than the surrounding normal skin. Often, the skin lesion is not quite hard to the touch. It is generally painless, but pruritus (itching) may be present. A violet-colored border may be seen when the lesions are still very active and extending. Sometimes, doctors will classify morphea further into other sub-types, according to the shape

or depth of the lesions. For example, “guttate” morphea refers to “drop-like” shaped areas of skin involvement, whereas “subcutaneous” morphea indicates a substantial involvement of deeper tissues with relative sparing of the overlying skin. The subcutaneous type may extend deep into muscle tissues in very rare instances,

B) Generalized Morphea – Generalized morphea involves larger skin patches than morphea, often including more of the body surface. Rarely, most of the body may be involved. Some patients with generalized morphea also have a band of thickening on an arm or leg as seen in linear scleroderma, another type of localized scleroderma (see below). Moreover, individual patches of morphea are common in linear scleroderma. Therefore, although one type of localized scleroderma usually predominates, patients may have a combination of different types of skin involvement. Patients with generalized morphea, because of the extensive surface area involved, may encounter considerable cosmetic disability resulting from the appearance of the problem (many dark and light areas of skin). Also, because of skin thickening over the joints, patients may have limited joint function.

C) Linear Scleroderma – Linear scleroderma, as the name implies, shows a band or line of skin thickening. It may extend deep into the skin and even involve the underlying muscle. The band of skin thickening is more common on the legs and arms and, when crossing the joints, may prevent proper joint motion. On rare occasions linear scleroderma can be a serious problem in children, especially when it extends deep into the skin. Sometimes, for reasons we do not yet understand, linear scleroderma delays growth of the underlying bones in children who are still in an active growth phase.

D) En Coup de Sabre – An unusual form of linear scleroderma on the face or scalp may appear as a white line referred to as “en coup de sabre.” This is a French term meaning “cut from a sword,” because of the way it looks. Some people think it may be completely different from linear scleroderma. En coup de sabre can be very destructive, as when it results in atrophy (loss

of tissue) of the face in children; this process may involve the tongue and mouth. Rarely, the condition is associated with abnormalities in the growth of facial bones, which can potentially lead to considerable deformities. There may be some overlap between en coup de sabre and a rare atrophy in the face, known as Parry-Romberg syndrome.

How is Localized Scleroderma Diagnosed?

Doctors who are familiar with scleroderma, or who are experts at examining the skin, can arrive at the diagnosis without much difficulty after a careful examination. In some cases, further tests may be needed to confirm the diagnosis. These tests may include taking a small sample of the skin (a biopsy) and some blood samples. It’s important that the entire skin surface be examined, so that a complete record is made of what is present at first (baseline record). Photographic documentation is also valuable.

Prognosis: What Will Happen?

Morphea occurs in all age groups, and tends to be more common in women.

Most patients develop only one or two patches of thickening that are frequently darker or lighter than the surrounding skin. A yellow discoloration also may occur. The changes in skin color may last for years, in spite of improvement and softening of the skin. In general, morphea tends to involve the superficial layers of the skin. Patients with morphea do quite well, but periodic follow-up is recommended. Rarely, patients will continue to develop new spots and essentially develop into generalized morphea. This evolution to generalized morphea is more common in women.

Generalized morphea occurs in all age groups, and patients with this type are expected to have more extensive and prolonged periods when the disease is active, in most cases lasting several years. Generalized morphea represents more than just a greater number of morphea lesions, but rather a greater tendency for the lesions to merge together. The areas of skin thickening in generalized morphea tend to become confluent (joined together), occasionally involving most of the body surface, and the depth of involvement

is generally greater than in morphea. Periodic follow-up is recommended.

Depending on the degree of skin involvement, patients with generalized morphea may have severe disfigurement from the changes caused by extensive skin thickening. Eventually, as with morphea, softening of the skin is expected, but the skin discoloration may last for many years and could remain permanent. This is because the increased pigment persists in the deeper portions of the skin and cannot be removed easily by bleaching agents.

Linear scleroderma is more common in children and adolescents. About 80 percent of patients diagnosed with linear scleroderma are younger than 20. Women are affected about four times more than men. Linear scleroderma has the potential to cause serious complications. The linear areas of the skin thickening may extend to the underlying tissue and muscle in children, which may impair growth in the affected leg or arm. The surface changes in the skin are similar to those described for morphea and generalized morphea. Extensive lesions of linear scleroderma, when cross joint lines, can impair motion of that particular joint. Unless continued efforts are made to maintain a full range of motion to the affected joint with physical therapy, this complication may be permanent and result in the affected area (for example, the elbow, arm, finger, etc.) being in a fixed position (contracture). Many patients with linear scleroderma, especially if older at the age of onset of the disease, will have only minor skin changes and minimal skin thickening. Linear scleroderma remains active for two to five years, but can last longer in some cases. Sometimes patients develop recurrences after a period of what was thought to be inactive disease. This “recurrence” is more frequent in patients with “en coup de sabre”.

En coup de sabre is potentially the most disfiguring form of localized scleroderma, because it affects the face and scalp. It can be mild, with only slight atrophy of the skin. However, depending on its locations on the face, it can lead to considerable problems, especially in children. It is possible that it is an entity by itself, and not truly a type of linear scleroderma. If located on the scalp, it can cause

varied degrees of hair loss. When involving the face, it can lead to indentations or depressions of the skin surface, especially on the forehead. The process can extend to the underlying bone. Recurrences can occur, even when it seems the disease has gone into remission.

Does Localized Scleroderma Go Away?

As a general rule, localized scleroderma is a self-limiting problem, at least in terms of activity of the process; the color changes are likely to remain. Sometimes, new lesions may appear for a few years, but eventually, the process of developing new areas of involvement will subside. The one possible exception to this statement is en coup de sabre, which may run an unpredictable course and become active again, even many years or decades after it first appeared.

Are There Other Complications of Localized Scleroderma?

Some patients with localized scleroderma, an estimated 10 to 20 percent, develop joint pain (arthralgia) during the course of their disease. The pain is not limited to joints beneath the involved skin, but may involve various joints, such as the knees, wrists and even the spine.

Occasionally, this complication occurs before the appearance of lesions in the skin, causing some confusion with rheumatoid arthritis for a short period of time. Eventually, the joint pains subside, even in the face of new skin involvement but an association with spine arthritis has been reported. Along the same lines, it is highly unlikely that patients with localized scleroderma progress and develop severe systemic (internal) conditions such as rheumatoid arthritis, or lupus erythematosus, or systemic sclerosis (the internal form of scleroderma). Still, it is important to recognize that morphea and other types of localized scleroderma may be present in several of these systemic conditions, including systemic lupus erythematosus, systemic sclerosis, and blistering diseases. An association has been found with a parasitic disease (toxoplasma). While this does not imply that patients with localized scleroderma evolve to develop these conditions, it does emphasize the need for follow-up and periodic evaluations,

especially in patients who continue to have active skin problems or develop new issues.

Are There Any Tests to Prove the Diagnosis of Localized Scleroderma?

The diagnosis of localized scleroderma is mainly by visual recognition, though a biopsy often may be done to show increased collagen deposition and other suggestive changes upon microscopic analysis of the skin. The biopsy may also help determine whether the disease is still active. However, skin lesions may extend even when the biopsy does not show any significant changes. The process of taking a skin biopsy involves first numbing the skin with an injection containing a numbing medicine (for example, lidocaine), and then taking a small (3-4 mm or about 0.15 inches) circular area of skin with a special “punch biopsy” device. Generally, unless one is dealing with a much deeper involvement of the skin, larger biopsies are not necessary. Stitches are optional when the biopsy site is very small. Any biopsy will result in a small scar.

It's important to look for physical signs of the internal type of scleroderma (systemic sclerosis), such as thickening of the skin on all the fingers (sclerodactyly), and thickening of the skin on the face (without color changes). There are other clues that can be identified during the initial examination. For example, patients with localized scleroderma do not have the typical changes at the base of the fingernails that are common in the internal (systemic sclerosis) disease: tiny blood vessel changes that require a special magnifying lens for proper detection. Another important question is whether patients have what is called Raynaud's phenomenon. This refers to the inability of the patient's hands to withstand cold exposure, so that the fingers may become pale, or even experience numbness and pain, and/or develop a temporary bluish appearance with even mild cold temperatures, or when touching a cold beverage, or getting some item from the freezer. Patients with localized scleroderma generally do not have Raynaud's phenomenon, which is present in the majority of patients with systemic sclerosis.

Several blood tests may be performed, which, when abnormal, help to determine how active the disease is and how extensive or prolonged

it may become. These blood tests include the number of blood eosinophils (cell normally present in our blood), the level of blood immune proteins (immunoglobulins), and various blood auto-antibodies: antinuclear antibodies (ANA), antibodies to single-stranded DNA (ssDNA) and antihistones antibodies. These tests are not specific for localized scleroderma and may be abnormal in other conditions, including the internal type of scleroderma. However, specific antibodies that are common in systemic sclerosis (typically anti-centromere antibodies or anti-topoisomerase or SCL70) are expected to be negative in patients with localized scleroderma

What Medicines Are Available?

There is no absolute cure for localized scleroderma. An absolute cure could be defined as not only complete stabilization of the skin lesions, but also disappearance of the changes that have taken place in the skin, under the skin, and any associated cosmetic or functional deformity. However, stabilization (inactive disease) of localized scleroderma is indeed possible. The outlook and prognosis of localized scleroderma is generally quite favorable. First, the disease is not fatal, does not appear to decrease lifespan and, except for extremely rare situations, is not associated with another disease that has internal organ involvement. Secondly, in most instances the condition is self-limited, and this definitely applies to individual areas of morphea. In those cases, no treatment is required or advisable, except for some emollients or, in some cases, mild topical steroids. Therefore, systemic treatment with a medication or other treatment interventions (for example, ultraviolet light), are reserved for more extensive forms of localized scleroderma. There are many publications that reference favorable results when using certain medications, but these have truly not been proven to work. Examples of these medications include: phenytoin (Dilantin), systemic corticosteroids (cortisone), antimalarial drugs (plaquenil, chloroquine), cyclosporine, d-penicillamine, antibiotics (presumably to treat an underlying Lyme disease problem) and vitamin E. A previously used vitamin D-based therapy (with calcitriol) has now been proven not to work. There is some recent good news for patients with extensive disease who require

proven medicines and other interventions. Methotrexate (as an anti-inflammatory drug) has proved effective in children with active and extensive (and deep) morphea. It has some side effects and should only be used when truly necessary. Ultraviolet light (there are different forms of this therapy, depending on the light wavelength being used) has also shown considerable promise in patients with active disease. In other situations or when these treatments fail, there are other medicines that can be tried in spite of the fact that they have not been proven to work yet in careful clinical research studies. There is definite agreement that physical therapy to preserve full motion of the affected joints is important.

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Please note that this brochure is provided for educational purposes only. It is not intended to substitute for informed medical advice.

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