

toes in changes of temperatures from hot to cold (Raynaud's Phenomenon), chilblains and fingertip ulcerations. The skin of the finger tips and toes often are the first to harden and become shiny, so is the skin over the nose. The hard skin then spreads and can possibly affect all the body. Swollen fingers and sore joints can occur in the beginning of the illness. Diagnosis is by skin biopsy. It is important that all the internal organs are assessed for disease with blood tests and other types of tests on the function of each organ. The child should be referred at the earliest opportunity to a paediatric rheumatologist.

#### *What is the treatment of systemic sclerosis in children?*

This varies depending on which organ is affected. For the skin, methotrexate has been proved to be beneficial. Where there are swollen joints and raised inflammation markers on blood tests, then steroids are used as well as methotrexate. For the Raynauds, good care of the circulation by keeping warm all the time is critical to prevent the skin from breaking down, and sometimes medication to dilate the blood vessels are needed. Where there is lung or kidney involvement, cyclophosphamide is often used. The decision as to which treatment is necessary has to be made by a paediatric rheumatologist with experience of scleroderma in conjunction with other specialists looking after specific systems such as the heart and kidneys.

Physiotherapy is needed during the illness to keep the joints and chest walls moving.

#### *What kind of periodic check-ups are necessary?*

The frequency of check-ups depends on the stage of the disease and these may be more frequent at the beginning. Their main aim is to adapt the treatment to the most recent development of the condition in the individual child. When certain drugs are used, their possible side effects have to be monitored as well. In localised scleroderma, sometimes paediatric dermatologists may follow up the children with periodic rheumatology check-ups. Since important internal organs can be involved (lungs, gastrointestinal tract, kidneys, heart) in certain cases of scleroderma, regular check-up of these organs is necessary. Laboratory investigations check the development of inflammatory markers and signs of autoimmunity.

#### *How long will the disease last for?*

The progression of the localised scleroderma is usually limited to several years. Often the skin hardening stops within two years after the start of the disease. Sometimes it can take up to 5 – 6 years, and some patches may become more apparent even after the inflammatory process is over due to the colour changes, or the disease may appear worse due to the unequal growth between the affected and unaffected parts of the body in the case of linear scleroderma. Systemic sclerosis is long-term disease that can be life-long.

#### *What is the long-term evolution of the disease?*

Morphoea usually leaves only cosmetic skin defects. Linear scleroderma can leave the affected child with severe problems due to loss of muscle and bone growth as well as causing a stiff and deformed joint. Recent treatments have minimized this effect in the majority of cases. Fortunately, compared to adults with systemic sclerosis it seems however, that children with systemic sclerosis have a much better outcome. However it is still a serious and potentially life-threatening disease that requires expert treatment. Generally the disease stabilizes after a number of years but long term follow-up even as an adult is recommended in case new problems develop.

#### *Is it possible to recover completely?*

Yes, children with localised scleroderma can recover. After some time even the hard skin may soften and appear normal. Recovery from systemic sclerosis is much less frequent, but significant improvements may be achieved and the disease often stabilizes, particularly in adulthood.

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## Childhood Onset Scleroderma

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## Introduction

Scleroderma is the Greek word that can be translated as “hard skin”. The skin becomes shiny and hard. There is a wide variety of diseases in which skin hardening is the most prominent feature. Some of them are more or less limited to skin (localised scleroderma), and can be in patches (morphoea), or occur as a tight band which can restrict the movement of the joint and cause the joint to become deformed (linear scleroderma, also known as linear morphoea). Sometimes linear scleroderma can affect the face and scalp exclusively and is known as “scleroderma en coup de sabre”. In the beginning, there may be a purplish edge to the patch of abnormal skin, before it becomes hard and shiny. Often there is loss of fatty tissue beneath the abnormal skin and slower growth of the muscle and bone tissues underneath. A minority of children with scleroderma have involvement of other parts of the body (this is termed systemic sclerosis) in addition to the hard skin. Internal organ involvement can lead to symptoms of difficulty in swallowing, often with heartburn. Sometimes the heart, lungs or kidneys can be affected and this may require specific tests and treatment. In systemic sclerosis, poor circulation (Raynaud’s phenomenon) is often also present.

### How common is it?

Scleroderma is a rare disease. Estimations of its frequency never exceed 3 new cases in 100 000 of the population every year. Localised scleroderma is the common form in children and is more frequent in girls than boys. Only about 10% or less of children with scleroderma have the type called systemic sclerosis. It is found in all races but studies of the frequencies in different ethnic groups have not been done. It can appear in every age; systemic sclerosis before puberty occurs equally in boys and girls, but after puberty, it occurs more often in girls.

### What are the causes of the disease?

Scleroderma is an inflammatory disease, but the reason for the inflammation has not been exactly discovered yet. It is probably an autoimmune disease, which means that the immune system of the child reacts against the skin. In systemic sclerosis, other organs of the body are also affected. The inflammation causes swelling and heat, followed by abnormal laying down of excessive

fibrous tissue during the repair phase. The cause is not known, but a number of triggers have been reported such as direct trauma to the skin in the case of localised scleroderma, and rape seed oil in systemic sclerosis.

### Is it inherited?

No, there is no evidence of strong genetic susceptibility for scleroderma so far, though there are some rare case reports of the familiar occurrence of this disease. Studies with large enough numbers of families remain to be done. In adult studies, there appear to be association with genes that are associated with other autoimmune diseases.

### Can it be prevented?

There is no known prevention for this condition.

### Is it contagious?

No. Some infections perhaps may trigger the disease onset, but the condition itself is not infectious and the affected children do not need to be isolated from the others.

## Localised scleroderma

### How is localised scleroderma diagnosed?

The appearance of hard skin is suggestive. Often there is a red or purplish rim to the patch in morphoea in the early stages. This reflects inflammation in the skin. In late stages, the skin becomes brown and then white in white people. In coloured people, this can look like a bruise and often the white patch is the first symptom that attracts the child or parent. The diagnosis is made with a skin biopsy. A test that measures the heat from the skin is often used (thermography). An inflamed patch will be hotter than the surrounding area as well as the same area on the other side of the body. An MRI can be helpful to see if the fat and muscle under the abnormal skin are affected. This is particularly useful for scleroderma en coup de sabre, where sometimes there is inflammation of the eyes, tissue behind the eye and in the brain itself.

### What is the treatment for localised scleroderma?

The current treatment is aimed at reducing inflammation. So this has to be done early in the disease. Such treatment will have very little effect on the fibrous

tissue once it is laid down. However, if inflammation is abolished, then the body is capable of breaking down some of the excess fibrous tissue eventually and the skin will become softer.

In the case of morphoea, a judgement by the doctor as to how inflamed it is and whether the underlying tissues are affected is important. Blood tests for inflammation markers are usually normal. Medication varies from no treatment, to the use of steroids and methotrexate for a period. The process often resolves on its own over a period of a few months to a year but can recur. Limited use of steroid cream (2 weeks) can be helpful in some cases.

In linear scleroderma however, more aggressive treatment is necessary since the fat, muscle and bone growth are all involved. If untreated, the bone stops growing leading to shortening of arms and legs or a small hand for example. There is loss of fat and muscle also under the tight skin. Moreover, the thick skin over a joint will cause stiffness and deformity of the joint. Thermography usually shows inflammation and can be a tool to assess the state of the inflammation during and after treatment. The anti-inflammatory treatment consists of steroids and methotrexate. Steroids may be given as intravenous infusions in the beginning. These treatments must be supervised and prescribed by a paediatric rheumatologist and/or a paediatric dermatologist with an interest in these conditions.

Physiotherapy is extremely important in the case of linear scleroderma. When the tight skin is over a joint, it is important to keep the joint moving with stretches and where appropriate, deep connective tissue massage. In cases where a leg is affected, unequal leg lengths can occur which will cause a limp, putting extra strain on the back, hips and knees. A shoe raise will improve all these effects.

Skin camouflage can help with the unsightly appearances, especially on the face. In white people, the skin should be protected from the sun by sunblock so that the morphoea (which cannot tan) will not be so obvious.

## Systemic sclerosis (systemic scleroderma)

### How is systemic sclerosis diagnosed?

The early signs are changes in colour of the fingers and