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SCLERODERMA

What is it?
Scleroderma is a Greek word that can be translated as "hard skin". In this disease, the skin becomes shiny and hard. There is a wide variety of diseases in which skin hardening is the most prominent feature and there are two different types of Scleroderma, localised and systemic scleroderma.
In **localised scleroderma** the disease is limited to skin and the tissues located under the affected skin. It can be in patches (morphea), or occur as a tight band (linear scleroderma).
In **systemic scleroderma** (or systemic sclerosis) the process is wide-spread and involves not only the skin, but also the internal organs of the body. This can lead to different symptoms, including heartburn, difficulty in breathing, and high blood pressure.

How common is it?
Scleroderma is a rare disease. Estimations of its frequency never exceed three new cases in 100,000 children every year. Localised scleroderma is the most common form in children and predominantly affects girls. Only about 10%, or less, of all scleroderma in children are systemic sclerosis.

What are the causes of the disease?
Scleroderma is an inflammatory disease, but the reason for the inflammation has not yet been discovered. It is probably an autoimmune disease, which means that the immune system of the child reacts against itself. The inflammation causes swelling, heat, and the production of too much fibrous tissue.

Is it inherited?
No, there is no evidence of a genetic link for scleroderma so far, though there are a few reports of the disease occurring in more than one member of the same family.

Can it be prevented?
There is no known prevention for this condition.

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

A) Localised scleroderma

1) How is localised scleroderma diagnosed?
The appearance of hard skin is suggestive. Often there is a red or purplish rim to the patch in the early stages. This reflects inflammation in the skin. In later stages, the skin

becomes brown and then white in Caucasian people. In non-Caucasian people, this can look like a bruise in the early stages, before it turns white.
The diagnosis is made on typical skin appearances.
Linear scleroderma appears as a linear streak on the arm or leg.
The process may affect the tissue under the skin, including the muscle and bone. Sometimes linear scleroderma can affect the face and scalp. Blood tests are usually normal. Significant internal organ involvement does not occur in localised scleroderma.

2) What is the treatment for localised scleroderma?
Treatment is aimed at stopping the inflammation as early as possible. Such treatment has very little effect on the established fibrous tissue. Once the inflammation has gone, the body is capable of reabsorbing some of the fibrous tissue and the skin can become softer again.
Medication varies from no treatment to the use of steroids and methotrexate. There are no studies that have clearly demonstrated the effectiveness of treatments in localised scleroderma. These treatments must be supervised and prescribed by a paediatric rheumatologist and/or a paediatric dermatologist.
The process usually resolves on its own, but may take a few years and it can recur.
In linear scleroderma, more aggressive treatment may be necessary.
Physiotherapy is 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, applying 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 avoid all these effects.
Massage of the lesions with moisturizing creams helps to slow down skin hardening.
Skin camouflage can help with unsightly appearances, especially on the face. In Caucasian people, the skin should be protect from the sun by sunblock so that the morphea (which cannot tan) will not be so obvious.

B) Systemic sclerosis

1) How is systemic sclerosis diagnosed? What are the main symptoms?
The early signs are changes in colour of the fingers and toes, with changes in temperatures from hot to cold (Raynaud's Phenomenon), chilblains and ulcers on the fingertips. The skin of the finger tips and toes often harden fast and become shiny, as does the skin over the nose. The hard skin then spreads and can eventually affect all of the body. Swollen fingers and sore joints can occur early in the illness.
During the course of the disease, internal organs may become affected and long term prognosis (prediction of disease course) depends on the type and severity of internal organ involvement. It is important that all the internal organs are assessed for disease involvement and other types of tests should be undertaken to monitor the function of each organ.
However, there is no specific blood test for scleroderma.
The esophagus is involved in the majority of children, often quite early in the disease course. This may cause heartburn, which is due to stomach acid coming in to the esophagus. Later on the entire gastrointestinal tract may become affected, with abdominal

distension and poor food digestion. Involvement of the lung is frequent and is a major determinant of the long-term prognosis. The involvement of other organs, such as the heart and the kidneys, is also very important for prognosis.

2) What is the treatment of systemic sclerosis in children?

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. Steroids are used, as well as methotrexate, or penicillamine. Where there is lung or kidney involvement, cyclophosphamide is often used. For Raynaud's phenomenon, 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 is needed. There is no therapy that has been shown to be clearly effective in systemic sclerosis. Other treatments are currently under investigation and there is the concrete hope that more effective therapy will be found in the next few years.

Physiotherapy and hard skin care are needed during the illness to keep the joints and chest walls moving.

What kind of periodic check-ups are necessary?

Periodic check-ups are needed to assess disease progression and to modify treatment. As important internal organs can be involved (lungs, gastrointestinal tract, kidneys, heart), regular assessment of organ function is necessary for early detection of their possible impairment.

When certain drugs are used, their possible side effects have to be monitored as well, with periodic controls.

How long will the disease last for?

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

What is the long-term prognosis of the disease?

Morphea usually leaves only cosmetic skin defects. Linear scleroderma can leave the affected child with severe problems due to loss of muscle and decreased bone growth, as well as causing stiff and deformed joints.

Systemic sclerosis is potentially a life-threatening disease. The degree of internal organ involvement (cardiac, renal and pulmonary system) varies among patients and is the major determinant of long-term prognosis. The disease may stabilize, in some patients, for long periods of time.

Is it possible to recover completely?

Children with localised scleroderma recover. After some time, even the hard skin may soften and appear normal. Recovery from systemic sclerosis is much less probable, but significant improvements, or at least disease stabilization, may be achieved.