www.printo.it/pediatric-rheumatology/IE/intro

Scleroderma

Version of 2016

2. DIFFERENT TYPES OF SCLERODERMA

2.1 Localised scleroderma

2.1.1 How is localised scleroderma diagnosed?

The appearance of hard skin is suggestive of localised scleroderma. Often there is a red/purplish or pale rim to the patch in the early stages. This reflects inflammation in the skin. In late stages, the skin can become brown and then white in Caucasian people. In non-Caucasian people it can look like a bruise in the early stages, before it turns white. The diagnosis is based on typical skin appearances.
Linear scleroderma appears as a linear streak on the arm, leg or trunk. The process may affect the tissues under the skin including the muscle and the bone. Sometimes linear scleroderma can affect the face and scalp. Patients with involvement of the skin in the face or scalp have an increased risk of uveitis (inflammation of the eyes). Blood tests are usually normal. Significant internal organ involvement does not occur in localised scleroderma. Often a skin biopsy is performed to help make the diagnosis.

2.1.2 What is the treatment for localised scleroderma?

Treatment is aimed at stopping the inflammation as soon as possible. The available treatments have very little effect on the fibrous (scar) tissue once it is formed. The fibrous tissue is the end stage of the inflammation. The goal of treatment is to control the inflammation to reduce the formation of fibrous tissue. Once the inflammation has gone, the body is capable of reabsorbing some of the fibrous tissue and the

skin can soften again.
Medication varies from no treatment to the use of corticosteroids, methotrexate or other immune-modulating drugs. There are studies demonstrating the beneficial effects (efficacy) as well as safety of these drugs in long-term therapy. The treatment must be supervised and prescribed by a paediatric rheumatologist and/or a paediatric dermatologist.
The inflammatory process may resolve on its own, but it may take a few years. In some individuals, the inflammatory process can persist for many years and in some it may become inactive and then re-occur. In patients with more severe involvement, more aggressive treatment may be necessary.
Physiotherapy is important, especially in linear scleroderma. When the tight skin is over a joint, it is important to keep the joint moving with stretches and, where appropriate, to apply deep connective tissue massage. When a leg is affected, unequal leg length may occur, causing limping and putting extra strain on the back, hips and knees. An insert to be worn inside the shoe of the shorter leg will make the functional length of the legs equal and will avoid any strain when walking, standing or running. Massage of the involved areas with moisturizing creams may be helpful.
Skin camouflage (cosmetics and dyes) can help with the appearances of affected skin (skin pigment changes), especially on the face.

2.1.3 What is the long-term evolution of localised scleroderma?

The progression of localised scleroderma is usually limited to a few years. The skin hardening often stops a few years after the start of the disease, but it can be active for several years. Circumscribed morphea usually leaves only cosmetic skin defects (pigment changes), and after some time the hard skin may even soften and appear normal. Some patches may become more apparent even after the inflammatory process is over, due to the colour changes.
Linear scleroderma may leave the affected child with problems due to unequal growth of the affected and unaffected parts of the body, resulting from loss of muscle and decreased bone growth. A lesion over a joint can cause arthritis and, if uncontrolled, can lead to contractures.

2.2 Systemic sclerosis

2.2.1 How is systemic sclerosis diagnosed? What are the main symptoms?

The diagnosis of systemic sclerosis is made mainly based on the symptoms and examination of the patient. There is no single test that diagnoses this condition. Lab tests are used to eliminate other diseases that are similar, to assess how active scleroderma is and to determine if organs other than the skin are involved. The early signs are changes in the colour of the fingers and toes with changes of temperatures from hot to cold (Raynaud's phenomenon) and ulcers on the fingertips. The skin of the finger tips and toes often hardens fast and becomes shiny. This can also occur in the skin over the nose. The hard skin then spreads and in severe cases may eventually affect the whole body. Swollen fingers and sore joints may occur early in the illness.

During the course of the disease, patients may develop further skin changes, such as visible dilations of small vessels (telangiectasias), wasting of skin and subcutaneous tissue (atrophy) and subcutaneous deposits of calcium (calcifications). The internal organs may be affected and long-term prognosis depends on the type and severity of internal organ involvement. It is important that all the internal organs (lungs, gut, heart, etc.) are assessed for disease involvement and other types of tests of the function of each organ are performed.

The oesophagus (gullet) is involved in the majority of children, often quite early in the disease course. This may cause heartburn, due to stomach acid entering the oesophagus, and difficulty in swallowing certain types of food. Later, more of the intestinal tract may become affected with abdominal distension (bloating) 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 the prognosis. However, there is no specific blood test for scleroderma. The physician managing patients with systemic scleroderma will evaluate organ system function at regular intervals to see if the scleroderma has spread to the organs or if the involvement is worse or better.

2.2.2 What is the treatment of systemic sclerosis in children?

The choice of the most suitable treatment is made by a paediatric rheumatologist with experience in scleroderma, in conjunction with other specialists looking after specific systems such as the heart and kidneys. Corticosteroids are used as well as methotrexate or mycophenolate. In the event of lung or kidney involvement, cyclophosphamide may be used. For Raynaud’s phenomenon, good care of circulation by keeping warm all the time is critical to prevent the skin from breaking down. Sometimes medication to dilate the blood vessels is needed. There is no therapy that has been shown to be clearly effective in all individuals with systemic sclerosis. The most effective treatment program for each individual needs to be determined by the use of medications that have been effective for others with systemic sclerosis to see if they work for that patient. Other treatments are currently under investigation and there is concrete hope that more effective therapies will be found in future. In very severe cases, autologous bone marrow transplantation may be considered. Physiotherapy and skin care are needed during the illness to keep the joints and chest walls moving.

2.2.3 What is the long-term evolution of systemic scleroderma?

Systemic sclerosis is potentially a life-threatening disease. The degree of internal organ involvement (heart, lung, gut and kidney) varies from patient to patient and is the major factor for long-term outcome. The disease may stabilize in some patients for long periods of time.