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Scleroderma

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1. WHAT IS SCLERODERMA

1.1 What is it?

The name scleroderma is derived from Greek and can be translated as "hard skin". The skin when affected by this disease becomes shiny and hard. There are two different types of scleroderma: localised scleroderma and systemic sclerosis.
In localized scleroderma, the disease is limited to the skin and tissues located underneath but not the internal organs. It can involve the eyes and cause uveitis, it may also involve the joints and cause arthritis. The skin can be affected in oval patches (morphea) or occur as a tight band (linear scleroderma).
In systemic sclerosis, the process is more severe and widespread and involves not only the skin but also some of the internal organs of the body.

1.2 How common is it?

Scleroderma is a rare disease. There is no detailed registry of children in Australia, but international estimations of its frequency never exceed 3 new cases in 100,000 people every year. Localized scleroderma is the most common form in children and affects predominantly girls. Only about 10% or less of children with scleroderma are affected by systemic sclerosis.

1.3 What are the causes of the disease?

Scleroderma is an inflammatory disease but the reason for the

inflammation has not been discovered yet. It is probably an autoimmune disease, which means that the immune system of the child reacts against itself. The inflammation causes swelling, redness or colour change from the inflamed tissues and then an overproduction of thickened (scar) tissue.

1.4 Is it inherited?

No, there is no evidence of a genetic link for scleroderma to date, although there are a few reports of the disease running in families.

1.5 Can it be prevented?

There is no known prevention for this condition. This means that you as a parent or a patient could not have done anything to prevent the onset of the disease.

1.6 Is it infectious?

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

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 localized scleroderma. Often there is a red or purplish or pale rim to the patch in the early stages. This reflects inflammation in the skin. In late stages, the skin becomes 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 (making it thinner) and the bone (affecting its growth). Sometimes linear scleroderma can affect the face and scalp. This can cause a scar

like appearance down the forehead or face called "en coup de sabre", as it may appear like a sabre cut. Patients with involvement of the skin in the face or scalp have an increased risk of uveitis. Blood tests usually yield normal results. 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 tissue once it is formed. The fibrous tissue is the end stage of the inflammation. The aim of treatment is to control the inflammation and hence to minimise 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 according to the stage and severity of the disease. It ranges 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.

In many patients, the inflammatory process resolves 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 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, 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 helps to slow down skin hardening.

Skin make up (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 or oval patches of 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 shortened or smaller bones. If the joint underneath has arthritis, this can lead to contractures – limiting the full movement of that joint.

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 lab test that can be diagnostic of scleroderma. Lab tests are used to rule out 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 and the fingers going purple or white (Raynaud’s phenomenon). Severe lack of blood flow causes 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 and around the mouth. 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 fat tissue (atrophy) and lumps of calcium under the skin (calcifications). The internal organs may be affected and long-term outcome 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 – the tube linking the mouth to the stomach - 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 (swollen belly) 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 lungs, 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 cutting and ulceration, and 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 real hope that more effective therapies will be found in future. In very severe cases, autologous bone marrow transplantation may be considered. Physiotherapy and hard 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 evolution. The disease may stabilize in some patients for long periods of time.

3. EVERYDAY LIFE

3.1 How long will the disease last?

The progression of localised scleroderma is usually limited to a few years. The skin hardening often stops after a few years from the start of the disease. Sometimes it can take up to 5 – 6 years and some patches may even become more visible after the inflammatory process is over due to colour changes, or the disease may appear worse due to unequal growth between affected and unaffected parts of the body. Systemic sclerosis is long-term disease that can last for years. However, an early and appropriate treatment can lessen the impact of the disease.

3.2 Is it possible to recover completely?

Children with localised scleroderma may recover completely. After some point even the hard skin may soften and only darker areas may persist. Complete recovery from systemic sclerosis is much less probable, but significant improvements, or at least stopping or slowing the disease, may be achieved, allowing a good quality of life.

3.3 What about unconventional/complementary therapies?

There are many complementary and alternative therapies available and this can be confusing for patients and their families. Think carefully about the risks and benefits of trying these therapies as there is little proven benefit and they can be costly both in terms of time, burden to the child and money. If you want to explore complementary and alternative therapies, please discuss these options with your paediatric rheumatologist. Some therapies can interact with conventional medications. Most doctors will not be opposed, provided you follow medical advice. It is very important not to stop taking your prescribed

medications. When medications are needed to keep the disease under control, it can be very dangerous to stop taking them if the disease is still active. Please discuss medication concerns with your child’s doctor.

3.4 How could the disease affect the child and the family’s daily life, and what kind of periodic check-ups are necessary?

Like any long-term disease, scleroderma does affect the child and the family’s daily life. If the disease is mild, without major organ involvement, the child and family generally lead a normal life. However, it is important to remember that children with scleroderma may frequently feel tired or be less resistant to fatigue, and they may need to change position frequently because of poor circulation. Periodic check-ups are needed to assess disease progression and of the need for treatment modifications. Because important internal organs (lungs, gastrointestinal tract, kidneys, heart) may be involved at different time points during the course of systemic sclerosis, regular assessment of organ function is necessary for early detection of possible impairment. Certain drugs need monitoring for side-effects which may include regular blood tests.

3.5 What about school?

It is essential to continue education in children with chronic diseases. There are a few factors that may cause problems for school attendance and it is therefore important to explain the child’s possible needs to teachers. Whenever possible, patients should take part in physical education; in this case, the same considerations discussed below with respect to sports should be taken into account. Once the disease is well-controlled, as is generally the case using the currently available medications, the child should have no problems whatsoever in participating in all the same activities as other children. School for children is what work is for adults: a place where they learn how to become independent and productive individuals. Parents and teachers should do whatever they can to allow the child participate in school activities in a normal way, in order not only for the child to be successful academically but also to be accepted and appreciated by both peers and adults.

3.6 What about sports?

Playing sports is an essential aspect of the everyday life of any child. One of the aims of therapy is to allow children to conduct a normal life as much as possible and to consider themselves not different from their friends. Therefore, the general recommendation is to allow patients to participate in the sports they choose and to trust that they will stop if there are limitations due to pain or discomfort. This choice is part of a more general attitude that tends to psychologically encourage the child to be autonomous and able to cope by himself with the limits imposed by the disease.

3.7 What about diet?

There is no evidence that diet can influence the disease. In general, the child should observe a balanced, normal diet for his/her age. A healthy, well-balanced diet with sufficient protein, calcium and vitamins is recommended for a growing child. Overeating should be avoided in patients taking corticosteroids because these drugs may increase appetite and lead to excessive weight gain.

3.8 Can climate influence the course of the disease?

There is no evidence that climate can affect the disease course.

3.9 Can the child be vaccinated?

Scleroderma patients should always consult their physician before receiving a vaccination of any kind. The physician will decide which vaccines the child can receive, considering case by case. Overall, vaccinations do not appear to increase the disease activity and do not cause severe adverse events in scleroderma patients.

3.10 What about sexual life, pregnancy, birth control?

There are no restrictions on sexual activity or pregnancy due to the disease. Nevertheless, many medications used to treat scleroderma may have adverse effects on a foetus. Sexually active patients are advised to use contraception and to discuss issues of contraception and

pregnancy with their doctor.