

Rare ichthyoses

What is ichthyosis?

'Ichthyosis' is a group of genetic skin conditions characterised by dry, scaly skin.

There are a number of conditions that as well as having ichthyosis present with other features and some of these very rare forms of ichthyosis are summarised below.

Sjögren-Larsson syndrome

This condition is characterised by three features: a) ichthyosis; b) neurological problems which are often severe and c) mild to moderate mental retardation. The neurological problems can cause weakness and partial paralysis in the legs, but often these children learn to walk unaided or with the help of crutches. It is inherited as autosomal recessive affecting one in four children, assuming both parents are carriers. It is caused by an abnormality in a chemical (fatty alcohols) in the skin and nerves. The gene has been found allowing pre-natal diagnosis early in pregnancy. Treatment includes regular skin care, moisturising creams, bath oil, physiotherapy, as well as support from a number of specialists – dermatologist, neurologist and orthopaedic surgeon. Acitretin or neotigason is a retinoid drug taken by mouth which is related to vitamin A and can be helpful for this condition. It needs careful monitoring under hospital supervision. Orthopaedic surgery, such as the release of tightened tendons, is beneficial.

Conradi-Hünemann syndrome

In this condition ichthyosis is associated with bone abnormalities and cataracts. X rays of the skeleton in babies shows tiny spots at the growing end of the long bones called stippling, which is characteristic for this condition. The ichthyosis or scaling of the skin is often in lines or swirled patterns. It is inherited as a disorder of the X-chromosome and seen in girls. Treatment is along the same lines as the other more severe ichthyoses.

Keratitis-ichthyosis-deafness (or KID) syndrome

This combination is rare and most cases are sporadic with no family history. The skin changes develop in infancy with spiny thickening of the skin, which is prone to becoming infected. They are handicapped by deafness and visual impairment. Treatment is symptomatic and along the same lines as the other more severe ichthyoses.

Refsums disease

This ichthyosis is usually not severe and develops later in childhood with fine white scales like ichthyosis vulgaris. The main problem is neurological with progressive weakness and a serious eye condition called retinitis pigmentosa. It is due to an accumulation of a fat chemical called phytanic acid in the skin and nerves. Dietary restriction of phytanic acid, found particularly in green vegetables, is helpful if started at an early age.

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People who care about ichthyosis

Trichothiodystrophies

The main features are ichthyosis and short brittle hair. It is due to a deficiency of sulphur, which can be demonstrated in the hair. There are a number of different variations depending on other associated symptoms. These are given the names: **IBIDS** (ichthyosis, brittle hair, impaired intelligence, decreased fertility and short stature) and **PIBIDS** which is the same but with photo- (or sun-) sensitivity. Treatment is symptomatic and along the same lines as the other more severe ichthyoses.

Moisturising creams and skin treatment

It is very important that the skin is kept moisturised at all times and that moisturising creams or ointments are applied frequently. Moisturising agents should be perfume-free and without additives to avoid any allergic reactions. The use of bath oils (not bubble bath or soap) is helpful so that bathing does not cause drying or irritation of the skin and soaking in the bath tends to be more beneficial than taking a shower.

Emollients and keratolytic creams which contain urea, and/or lactic acid (see product's ingredients label for these) can be useful to remove scales. However these should be avoided if the skin is particularly itchy or red because they may irritate the skin.

In very severe cases antibiotics can be given to treat secondary infections.

More specific information on caring for the skin can be found on the Ichthyosis Support Group website at www.ichthyosis.org.uk.

Personal care

Scaling and peeling skin, particularly in the most visible areas such as the scalp or face, can be difficult for building self-confidence so individuals may need extra support. General practitioners need to understand the psychological impact of the condition and provide adequate support to the family. This may require the involvement of other healthcare professionals.

Further help

There are a number of online forums about Ichthyosis where individuals can share their experiences and detail their own treatment recommendations. Not everything will work for everyone but these forums are a good place both to receive and offer support.

Contact the Ichthyosis Support Group for information, advice, details on useful products, and to be connected with other people to share experiences and helpful advice.

To find out more about the ISG or become a member please get in touch in one of the following ways:

By Phone or Fax:

Tel: **0845 602 9202** Fax: **0560 343 8046** (on request)

By Email:

isg@ichthyosis.org.uk

By Post:

Ichthyosis Support Group

PO Box 1242, Yateley GU47 7FL

Facebook:

facebook.com/ichthyosissupportgroup

Twitter:

twitter.com/ISG_Charity