



ICHTHYOSIS

What are the aims of this leaflet?

This leaflet has been written to help you understand more about ichthyosis (pronounced ick-thee-o-sis). It will tell you what it is, the types of ichthyosis, what can be done about it, and where you can get more information about it.

What is ichthyosis?

Ichthyosis is the term used to describe continual and widespread scaling of the skin. It may be inherited (genetic) or acquired during life. The inherited forms are rare, generally present from infancy, and are usually lifelong conditions. Acquired ichthyosis can develop at any age due to a number of medical problems, such as kidney disease.

The commoner forms of inherited ichthyosis are mild and do improve during warmer weather. There are a number of very rare conditions where ichthyosis occurs with problems in other systems of the body. Each of the major types of ichthyosis will be discussed briefly, followed by an outline of the management.

What is inherited ichthyosis?

Inherited ichthyosis is due to a single genetic trait which is passed on either from one or both parents, or develops as a new error in the gene very early in foetal life. It can be mild as with ichthyosis vulgaris, or severe.

Ichthyosis vulgaris

This is the commonest form of inherited ichthyosis, affecting 1 in every 250 people. Ichthyosis vulgaris has been found to be due to a gene defect in filaggrin, which is a protein in the skin that impairs the skin barrier formation and the natural moisturising factors that are key to keeping the skin hydrated. It is usually quite mild and develops in early childhood with fine, light grey

scales and roughness on the upper and lower limbs, but sparing the folds of the arms and legs. It may be more widespread and is more obvious in the winter time. It is sometimes associated with atopic or childhood allergic eczema and may cause an increased wrinkling of the palms and soles.

It can be treated with regular application of moisturisers. It improves in adult life and may be passed on to children whereby there is a 50:50 chance of each child having the disease (“autosomal dominant transmission”).

X-linked ichthyosis

This condition occurs in males only and develops in infancy with tan or grey scales on the limbs and across the trunk. It may affect the ears and face and the scales appear to be stuck on like stamps. It varies in its severity and improves in fine or sunny weather. It changes very little with age.

X-linked ichthyosis is due to lower levels of an enzyme known as steroid sulphatase. This condition is passed on by a mother, who is a carrier of the abnormal gene, to her son with a 50% risk for each son. A carrier mother shows no evidence of the condition. The gene that causes this condition has been identified and, very rarely, a similar fault can affect adjacent genes on the same chromosome, causing a variety of other problems for an affected male, for instance, bony defects or lack of the sense of smell.

A mother who is carrying an affected male baby may have a prolonged and consequently difficult labour. A small number of affected boys have poorly descended or undescended testicles; it is usual to check this aspect of development in affected families. The child's growth should be monitored from time to time. It is quite common to have asymptomatic specks in the cornea that do not interfere with vision which are identified by an eye specialist.

Autosomal recessive congenital ichthyoses

There are three types of ichthyoses in this category: Congenital ichthyosiform erythroderma, Lamellar ichthyosis and Harlequin ichthyosis. These three conditions are passed on by parents with normal skin who both carry the abnormal gene (autosomal recessive) and the risk of a further child being affected is 1 in 4.

Congenital ichthyosiform erythroderma

This type of ichthyosis generally show signs at birth with the appearance of a collodion membrane on the newborn baby. This is a shiny yellow film

stretched across the skin like a sausage skin. It dries out and gradually sheds within the first week of life. The "collodion baby" is nursed in a humidified incubator until the skin settles down. The majority of collodion babies will develop congenital ichthyosiform erythroderma. However, a small number of these babies will have normal skin once the membrane is shed and, therefore, it is not possible to accurately predict the outcome in the early stages.

Most collodion babies develop non-bullous ichthyosiform erythroderma, which literally means inflamed, scaly skin, without blisters. It affects 1 in every 300,000 births and so is very rare. Once the collodion membrane has shed, the skin remains red and has fine, white scales affecting the entire skin surface. In severely affected children the eyelids may be pulled outwards and there may be some mild scalp hair loss and tightness of the fingers. Because the skin is inflamed it will feel hot, even if the child is cold. Most children with this condition do not sweat normally and may overheat in hot weather, when exercising or with a fever. The palms and soles are sometimes thickened and scaly. Otherwise a child's health is normal. They may suffer cosmetically and this is especially important when a child starts school, or in the teenage years.

Lamellar ichthyosis

Lamellar ichthyosis is a rare form occurring in 1 in 200,000 live births. Newborns usually present with a collodion membrane but the skin is different in that it is less red but the scaling is larger, perhaps darker and more adherent, or stuck down.

Harlequin ichthyosis

Harlequin ichthyosis is a very severe, but extremely rare type of inherited ichthyosis (approximately 5 per year in the UK). It is evident at birth due to the very thick scaling all over. Intensive care is required and detailed information on the condition will be needed for the parents and staff.

Bullous ichthyosis

Bullous ichthyosis also known as bullous ichthyosiform erythroderma, is another rare inherited ichthyosis. At birth the baby's skin seems to be fragile and may show blisters, without much scaling. This causes severe problems for the young infant and intensive care is sometimes necessary in the first few weeks of life. A skin biopsy will be required early on to confirm the diagnosis. During the first year or two of life, the blistering tendency reduces but widespread redness, scaling and thickening of the skin becomes more

obvious through childhood. This produces warty skin changes around the creases of the joints. Skin infections are quite common and can lead to a characteristic odour. There may be a reduction in sweating in childhood, which improves later in life. This is a troublesome and distressing condition for the child and the family.

It is transmitted as an autosomal dominant disorder, which means that one of the parents may be affected. However, in at least half of affected children, neither parent is affected; therefore the child has developed a new gene fault while growing in the womb. This means that there is no risk above the ordinary for further pregnancies in that family although the child may pass on the condition onto the next generation. Detailed genetic counselling is necessary.

Netherton's syndrome

The incidence of this condition is not known but it is probably in the region of 1 in each 200,000 births in the UK. The newborn child is very red and has scaly or peeling skin. The infant is often underweight and slow to grow and this problem continues for the first year or two of life. The affected child may need prolonged hospital treatment until both the skin and the nutrition improve. A characteristic feature of Netherton's syndrome is thin fragile scalp hair in the baby. Later it is spiky and this is an important clue to the diagnosis. In many affected children the skin improves in childhood, although it can flare up without warning.

Netherton's syndrome is an autosomal recessive disorder where both parents are carriers and show no sign of the condition. There is however a risk to further babies of the order of 25%.

There are a number of other genetic or inherited forms of ichthyosis where there are other medical problems but these are so uncommon that they are not discussed here.

What are the treatments for ichthyosis?

In the milder types of ichthyosis the main treatment is regular application of moisturisers or emollients. A very wide selection of emollients is available (creams, ointments, lotions, bath oils) and a person with ichthyosis should try several different types before deciding on which suits them best. These moisturisers are most effective when applied on wet skin within a couple of minutes of having a shower or bath. Parents will need to help young children with regular moisturisers, so that they get into the habit of applying them.

Peeling creams (keratolytics, e.g. salicylic acid) are sometimes used but they can irritate the skin and can be helpful on the palms and soles.

CAUTION: This leaflet mentions 'emollients' (moisturisers). When paraffin-containing emollient products get in contact with dressings, clothing, bed linen or hair, there is a danger that a naked flame or cigarette smoking could cause these to catch fire. To reduce the fire risk, patients using paraffin-containing skincare or haircare products are advised to avoid naked flames completely, including smoking cigarettes and being near people who are smoking or using naked flames. It is also advisable to wash clothing and bed linen regularly, preferably daily.

The rarer types of ichthyosis with inflamed skin in the infant require intensive medical and nursing care. For children and adults with these severe types of ichthyosis, a trial of retinoid (synthetic vitamin A) tablet treatment may be suggested. Retinoids reduce the growth of overactive scaly skin and can improve its appearance. They have little influence on the inflammation or redness of severely inflamed ichthyosis. Regular blood tests and checks on growth will be needed. As retinoid treatment can severely damage an unborn baby, pregnancy is to be avoided during treatment and for few months after stopping retinoids. Antibiotics or antiseptics may be prescribed for skin infections but steroid treatments are not effective in ichthyosis.

The genetics of ichthyosis

There are so many types of inherited ichthyosis that have different patterns of inheritance. The parents of an affected child and, when appropriate, the affected person, may need expert genetic counselling to help them understand the genetics of the disorder.

Research in ichthyosis

There have been dramatic advances in the understanding of the causes of certain inherited ichthyosis, especially ichthyosis vulgaris, X-linked recessive ichthyosis and bullous ichthyosis. Intensive research is ongoing and in time will lead to early diagnosis and better treatments.

Where can I find out more about ichthyosis?

Web links to detailed leaflets:

<http://emedicine.medscape.com/article/1112753-overview>
<http://dermnetnz.org/scaly/ichthyosis.html>

Links to patient support groups:

Ichthyosis Support Group

PO Box 1404

Bagshot, GU22 2LS

Email: isg@ichthyosis.org.uk

Web: <http://www.ichthyosis.org.uk/leaflets/>

For details of source materials used please contact the Clinical Standards Unit (clinicalstandards@bad.org.uk).

This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel

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