PATIENT INFORMATION LEAFLET

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 explains what it is, the different types, how it can be managed and where you can get more information.

WHAT IS ICHTHYOSIS?

Ichthyosis refers to a group of skin conditions that cause a widespread persistent dry, scaly skin. This can range from mild to severe and is sometimes associated with skin inflammation (red or darker skin). It may be inherited (genetic) or acquired during life. The word 'ichthyosis' comes from the Greek word for fish, 'ichthys,' as in some types of ichthyosis, the skin is said to resemble fish scales.

Types of ichthyosis that are inherited are rare. Generally, these are present from birth and are usually life-long conditions. They are due to a single gene which is passed on either from one or both parents, or it develops as a new mutation (change) in the gene very early in the baby's life. The more common forms of inherited ichthyosis are usually mild. There are a few very rare types that cause problems elsewhere in the body.

Acquired ichthyosis can develop at any age, usually in adulthood, due to an underlying medical problem or medication. This leaflet will focus on the different types of inherited ichthyosis.

The common types of inherited ichthyosis are:

Ichthyosis vulgaris

This is the most common form of inherited ichthyosis, affecting 1 in every 250 births.

Ichthyosis vulgaris is due to a gene change causing loss of function in a skin protein called filaggrin. This protein helps to form an effective barrier to protect the outer skin from irritants and prevent it from drying out. It also forms natural moisturising substances that are key to keeping the skin hydrated. Therefore, when the gene for this protein is altered, the skin barrier doesn't work as well.

Ichthyosis vulgaris is usually quite mild and develops in early childhood. It is characterised by fine, white to grey scales on the abdomen, arms and legs. The face, elbows, armpits and knee creases are usually unaffected. Rough bumps can appear on the skin of the arms and legs. The palms and soles may have increased skin markings. It may be more widespread and obvious in the wintertime. It is sometimes associated with being atopic (overreactive immune system which causes asthma, allergies and eczema) and generally improves in adult life.

X-linked ichthyosis

This condition only affects males. It occurs in about 1 in 6000 births. It can cause generalised peeling of the skin in newborns and then fine scales on the trunk and limbs in infancy. Over time, the scales become thicker with a brown or grey colour. These thicker scales do not peel very easily and appear to be stuck on. It mainly affects the lower legs, upper arms and abdomen. The elbow and knee creases are often not affected. It varies in its severity and generally improves in warmer weather and adult life.

X-linked ichthyosis is due to lower levels of an enzyme known as steroid sulfatase. This

condition is only seen in people born as males. Females may carry the faulty gene. They can pass it on to a son with a 50% risk for each son. In very rare cases a carrier mother may show some mild symptoms of the condition.

Most commonly, X-linked ichthyosis is caused when a part of the X chromosome which includes the steroid sulfatase gene is missing. Boys and men affected in this way may have other conditions such as autism, learning difficulties, abnormal heart rhythms or loss of smell. These can occur depending on what other genes are also "deleted" alongside the ichthyosis gene. In other cases, X-linked ichthyosis results from a mutation within the steroid sulfatase gene - this does not usually have associated problems.

A mother who is carrying an affected male baby may have a prolonged and 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 painless spots on the surface of the eye (the cornea). These spots are called asymptomatic specks and do not interfere with vision.

Rare types:

Autosomal recessive congenital ichthyoses

There are three main types of ichthyoses in this group: 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). The chance of each of their children being affected is 1 in 4.

1. Congenital ichthyosiform erythroderma

This type of ichthyosis generally shows signs at birth. The baby's body will be covered by a collodion membrane. This is a shiny yellow film stretched across the skin which 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 which is termed self-improving collodion ichthyosis.

Congenital ichthyosiform erythroderma causes inflamed, scaly skin, without blisters. It affects 1 in every 200,000 births. Once the collodion membrane has shed, the skin remains red and has fine, white scales covering the entire body 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 affected by 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. The child's health is otherwise normal.

2. *Lamellar ichthyosis*

Lamellar ichthyosis occurs in about 1 in 200,000 births. Newborns usually present with a collodion membrane. Once the membrane is shed in the skin is less inflamed, but the scales are larger and generally darker or brownish in colour. The scales can affect the whole body but may be larger on the legs. The skin on the palms and soles of the feet may be thickened with painful cracks. The skin loses more water causing dehydration and there is a risk of skin infection.

The baby will be nursed in an incubator and have intensive care to cope with all the expected problems. Some children are often admitted to hospital with repeated infections and dehydration.

Some of the skin signs seen in congenital ichthyosiform erythroderma and lamellar ichthyosis can be similar. Alterations in many different genes can result in

autosomal recessive congenital ichthyosis. Bathing suit ichthyosis is a rare form of autosomal recessive congenital ichthyosis. Newborns with this condition often have a collodion membrane. After the membrane is shed and over time, areas affected by large dark-coloured scales become limited to warmer parts of the body such as the scalp and body, rather than the arms and legs.

3. Harlequin ichthyosis

Harlequin ichthyosis is an extremely rare and severe type of inherited ichthyosis (approximately 5 cases per year in the UK). It is evident at birth due to the very tight, thick scaling skin all over the body.

Harlequin ichthyosis causes the skin to split and fluid to leak out of it. This leads to deep cracks, dehydration and thickening of the skin. The thickened skin prevents the production of sweat. In turn, this leads to the body's inability to regulate its temperature. The face looks stretched with turned out lips and eyelids; the ears, hands and feet may be hidden in the scale. The eyes may be invisible because they are temporarily covered by the swollen eyelids. Feeding, and sometimes breathing, is difficult. There is risk of serious infection.

Immediately after birth, the baby will be nursed in the neonatal intensive care unit. They may need antibiotics to prevent skin infections. They will be placed in an incubator and have intensive care to cope with all the expected problems. Skin creams and, in some cases, retinoid medicine will help the scale plates to shed in a few weeks. Then the skin will appear red, tight and scaly all over and this is usually an ongoing problem.

Parents will be taught how to cope with the day-to-day management.

4. Bullous ichthyosiform erythroderma

Epidermolytic ichthyosis 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 may be required early on to confirm the diagnosis. During the first year or two of life, the blistering tendency reduces but widespread redness/darkening, scaling and thickening of the skin becomes more obvious *throughout* childhood. This produces warty, ridged skin changes around the creases of the armpits, elbows and knees. Skin infections are quite common and can lead to a characteristic odour. Sweating may reduce during childhood and improve later in life.

Epidermolytic ichthyosis is inherited 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 only a very small risk above the ordinary for the parents' further pregnancies, although the affected child may pass the condition onto their children. Detailed genetic counselling is necessary.

5. Netherton syndrome

This is a condition where ichthyosis is associated with significant abnormalities elsewhere in the body.

The incidence of this condition is not known but it is probably in the region of 1 in 100-200,000 births in the UK.

The newborn child has very red/darker skin which can be scaly or peeling. The infant is often underweight and slow to grow (sometimes called failure to thrive). This problem continues for the first year or two of life. The affected child may need prolonged hospital treatment until both the skin and their nutrition improve.

A characteristic feature of Netherton syndrome is thin fragile scalp hair in the baby. Later on, the hair becomes 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. Many patients are prone to develop food allergies.

Netherton syndrome is an autosomal recessive disorder where both parents are carriers but show no signs of the condition. There is however a 1 in 4 risk for these parents of their children being affected.

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

HOW IS ICHTHYOSIS TREATED?

For the milder types of ichthyosis, the main treatments are regular bathing and application of moisturisers.

When bathing, it is important to use a moisturiser as a soap substitute or dedicated products such as bath oils and shower lotions. Whilst bathing gentle exfoliation (removing the scales) can be tried using a loofah sponge, bathing mitt or pumice stone.

Moisturising should be done at least twice a day. A person affected by ichthyosis should try several different types of moisturisers before deciding which suits them best. Moisturisers are most effective when applied to wet skin within a couple of minutes of having a shower or bath. Parents will need to help young children with routine regular moisturising, so they get into the habit of applying them. These are the types of moisturisers which can be used:

- Emollients help soften the skin, add moisture and prevent water loss. They come in the form of lotions, creams and ointments. Lotions are lightweight, creams a little thicker and ointments are thick and greasy.
- Humectants contain ingredients like urea or glycerin that can help keep and draw water into the skin.
- Keratolytics contain ingredients such as salicylic acid, lactic acid and urea. These help break down the dry/rough skin but can sometimes cause skin irritation and should be avoided in children under 2 years old.

Infants with rarer and more severe types of ichthyoses require intensive medical and nursing care. Children and adults affected by these severe types may be given a trial of retinoid (synthetic vitamin A) treatment. This can be either in cream or tablet form. Retinoids can help reduce the scaliness of the skin. If taken in tablet form regular blood tests and checks on growth will be needed. Retinoid treatment can damage an unborn baby severely. Pregnancy must be avoided during and after stopping treatment for up to 3 years, depending on the type of retinoid taken.

Antibiotics or antiseptics may be prescribed for skin infections. Steroid treatments are not effective in ichthyosis.

CAUTION:

This leaflet mentions 'emollients' (moisturisers). Emollients, creams, lotions and ointments contain oils. When emollient products get in contact with dressings, clothing, bed linen or hair, there is a danger that they could catch fire more easily. There is still a risk if the emollient products have dried. People using skincare or haircare products should be very careful near naked flames or lit cigarettes. Wash clothing daily and bedlinen frequently, if they are in contact with emollients. This may not remove the risk completely, even at high temperatures. Caution is still needed. More information may be obtained at https://www.gov.uk/guidance/safe-use-of-emollient-skin-creams-to-treat-dry-skin-conditions.

Some people affected by ichthyosis may find they do not sweat effectively. This is due to the thickened skin; some simple measures to avoid overheating can be helpful. These include avoiding very high temperatures and prolonged, direct, strong sunlight. Staying hydrated and wearing a wet t-shirt or using cool packs can also be helpful. Vitamin D levels are often low in individuals with ichthyosis. This is due to difficulty making vitamin D in the skin from sunlight and, sometimes, from reduced skin exposure to sunlight. This should be checked and supplemented if deficient.

Ichthyosis can have a significant psychological and emotional impact on those affected by it. It is important that you speak to the dermatology team so they can direct you or your child to other teams who can support your needs. The Ichthyosis Support Group can also provide you with support and additional information. The genetics of ichthyosis

THE GENETICS OF ICHTHYOSIS

The inherited ichthyoses have different patterns of inheritance. Individuals and parents of an affected child may need expert genetic counselling to help them understand the genetics of the condition.

RESEARCH IN ICHTHYOSIS

There have been dramatic advances in the understanding of the causes of certain inherited ichthyosis. Research is ongoing and in time will lead to earlier diagnosis and better treatments.

WHERE CAN I FIND OUT MORE ABOUT ICHTHYOSIS?

Patient support group providing information:

Ichthyosis Support Group
Email: isg@ichthyosis.or.uk
Web: http://www.ichthyosis.org.uk

Web links to other relevant sources:

NHS*:* Ichthyosis - NHS (www.nhs.uk)
DermNet NZ:
dermnetnz.org/scaly/ichthyosis.html

Primary Care Dermatology Society:
Ichthyosis (pcds.org.uk)

Great Ormond Street Hospital (GOSH):
Ichthyosis | Great Ormond Street Hospital (gosh.nhs.uk)

Ichthyosis Support Group:
www.ichthyosis.org.uk/Pages/FAQs/Category/what-is-ichthyosis
Jargon Buster:

www.skinhealthinfo.org.uk/support-resources/jargon-buster/

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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