Ichthyosis

This information sheet from Great Ormond Street Hospital (GOSH) explains the skin condition ichthyosis – what causes it, its symptoms and treatment and where to get help.

What is ichthyosis?

Ichthyosis is a term used for a group of conditions that affect the skin, making it rough and scaly. The name comes from the Greek for 'fish' as sometimes the skin may look a little bit like fish scales. Normal skin is continuously shed and re-grown to form an effective barrier against infection and other damage. In ichthyosis, this mechanism does not work properly, so the skin does not shed properly, so builds up as thick, rough areas. How much of the body is affected varies depending on the particular type of ichthyosis.

Ichthyosis can feature as part of a syndrome (collection of symptoms often seen together), for instance, Netherton syndrome.

Ichthyosis can be acquired later in life, usually in adulthood, so is not covered in this information sheet.

What causes ichthyosis?

Ichthyosis is caused by a fault on a gene that is responsible for the skin shedding and regrowth process. In some cases, this fault develops sporadically (out of the blue) but in many cases, it is passed on from parents to their child. How the faulty genes are passed on varies depending on the type of ichthyosis.

Ichthyosis vulgaris is passed on in an autosomal dominant manner – this means that a child only has to inherit the faulty gene from one parent to risk having the condition. For each pregnancy, there is a 1 in 2 (50 per cent) chance of the child having ichthyosis vulgaris.

X-linked ichthyosis is passed on in an X-linked manner – this means that the faulty gene is on the X-chromosome. As females have two X-chromosomes, the faulty gene is cancelled out by the other normal one so females cannot have this type of ichthyosis. Males only have one X-chromosome so are affected by this type.

Autosomal recessive congenital ichthyosis including harlequin ichthyosis are passed on in an autosomal recessive manner – this means that a child has to inherit the faulty gene from both parents to risk having the condition. For each pregnancy, there is a 1 in 4 (25 per cent) chance of the child having this type of ichthyosis and a 1 in 2 (50 per cent) they will inherit the faulty gene but not develop the condition – this means they are a 'healthy carrier' so could pass it on to their children in future.

What are the signs and symptoms of ichthyosis?

The main symptom of all types of ichthyosis is rough, scaly skin that is very dry. The specific areas affected and how severely will vary depending on the type of ichthyosis.

In ichthyosis vulgaris, the scaly areas of skin tend to affect only the limbs (arms and legs) but not in the skin folds, such as behind the knees or inside the elbows.

Boys with X-linked ichthyosis have scaly skin on their limbs and torso, as well as their ears and face. It is very variable in severity and in many cases, seems to improve during the summer months.

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Bullous ichthyosis leads to blistering of the skin, which generally improves as the child grows older, but the skin remains red, scaly and thickened.

Some types of ichthyosis present as a 'collodian membrane' – this is a yellowy shiny covering that sheds soon after birth. After this, the specific type of congenital ichthyosis may become evident.

How is ichthyosis diagnosed?

All types of ichthyosis have a characteristic appearance but as it is a rare condition, diagnosis of the precise type of ichthyosis will usually only be possible at a specialist centre. With the more severe types of ichthyosis, a baby may be transferred to a specialist centre soon after birth both for diagnosis and the more intensive support needed. Blood tests may be ordered to identify the gene mutation causing the condition.

How is ichthyosis treated?

Children with the more severe forms of ichthyosis may need intensive support soon after birth as the scaly sore patches of skin can lead to rapid heat loss and infection. There is no cure for any type of ichthyosis but the symptoms can be managed with an intensive daily treatment programme.

What happens next?

Once any problems soon after birth have been overcome, a near-normal day to day life in usually possible. As teenage years approach, appearance becomes more important so additional support to develop coping strategies may be needed. Ichthyosis is a lifelong condition so as a young person grows older, they will need to learn to become more independent before their care is transferred to an adult specialist service.

Further information and support

The Ichthyosis Support Group UK offers support and advice to anyone affected by ichthyosis. Call their helpline on 0845 602 9202 or visit their website at www.ichthyosis.org.uk

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