

Overview of ichthyosis

What is ichthyosis?

Ichthyosis, pronounced Ick-thee-o-sis (which comes from the Greek word meaning 'fish') describes a group of conditions in which the skin is dry and scaly. Lots of people have dry skin conditions (such as eczema or psoriasis) but they tend to be patchy and they come and go. By contrast, in ichthyosis the scaling is continuous and usually affects the whole body. Most types are congenital, meaning that they are present at birth, and inherited, meaning that they result from genetic changes, so they may run in families. More information on this can be found in the 'What is meant by congenital ichthyosis leaflet'.

You are likely to have lots of questions and concerns about ichthyosis which you may wish to discuss with your doctor, but the following section may help to answer some questions which individuals and parents often ask.

What causes the scaling in ichthyosis?

The skin is made up of millions of tiny cells joined together to form our protective covering. Skin is a living organ which has to grow, adapt and respond to damage. It is constantly shedding (or exfoliating) and being replaced, and these processes are controlled by genes. Mistakes ('mutations') in genes cause malfunction. Ichthyosis is caused by mutations in genes that control the formation of skin cells, so they don't function properly. Different types of ichthyosis are caused by mutations in different genes: in some, skin cells are formed at a faster rate than they are needed and they pile up on the skin surface, thickening the skin. In other forms, the cells are produced at the normal rate but instead of brushing off when they reach the surface, they cannot become detached from the cells beneath them and so they build up in layers. Either way, the end result is ichthyosis.

Can ichthyosis be treated?

There is no cure for ichthyosis at present, but it is possible to manage the symptoms.

The main aim of treatment is to improve the condition of the skin (make it less dry and less scaly, for example) and to relieve discomfort. This is primarily achieved through regular, intensive (at least twice daily) use of moisturisers, sometimes with antiseptics or antibiotics, and occasionally with retinoids – a group of drugs that can reduce skin scaling – and other medicines as needed. Treatment is dealt with in more detail in the 'Managing Ichthyosis leaflet'.

There are many different types of ichthyosis. Some are listed in the table below. Please note that this is not an exhaustive list of all forms of ichthyosis and there are a number of other syndromes with ichthyosis. This information focuses on the forms of ichthyosis relevant to the majority of ichthyosis patients. Some of the rarer forms (or related syndromes) are dealt with in the 'Rarer forms of ichthyosis' leaflet. Leaflets about specific conditions can also be found on our website.



Types of congenital (inherited) ichthyosis

Name	Appearance & severity	Frequency	Notes
Ichthyosis vulgaris (IV)	Appears in the first year or two. Usually mild, with fine scaling or roughness on the abdomen, arms and legs and more skin creases on the palms. Knee and elbow flexures usually spared.	Most common form of ichthyosis accounting for 95% of ichthyosis cases; occurs in 1 in 250 to 1 in 100 people.	People with IV are prone to atopic (allergic) eczema, asthma and food allergies. The skin may improve in warm weather.
X-linked (recessive) ichthyosis (XLI)	Appears in the first few months. Varies in severity, with brownish flat scales, most obvious on the arms, legs and tummy. Flexures e.g armpits are usually spared.	Occurs in 1 in 2000 to 1 in 6000 males	Only occurs in males. Mothers of some affected boys have a long and difficult labour. Skin condition may improve in summer.
Bullous ichthyosis/ Bullous congenital ichthyosiform erythroderma (BCIE)/ Epidermolytic hyperkeratosis (EHK)	Appears at or soon after birth with blistering, fragile, red, shiny skin, replaced later by thick scaling especially around the joints. The scale easily lifts off leaving superficial but painful raw areas.	Rare – occurs in 1 in 100,000 babies	Skin infections are quite common and may cause odour. Symptoms are due to skin fragility and overproduction of skin cells
Netherton syndrome (NS)	Red, inflamed, itchy, scaly skin from birth; may be all over or patchy and severity fluctuates. Hair is sparse, brittle and spiky.	Rare – occurs in 1 in 200,000 babies	Patients may be very unwell, with poor weight gain in childhood, and multiple food allergies. Symptoms are due to skin inflammation and impaired protective barrier function.
Non-bullous ichthyosis/Non-bullous congenital ichthyosiform erythroderma (NBCIE)	"Collodion baby" (see <i>Caring for a Collodion baby</i> leaflet) refers to the appearance at birth and is followed by red, itchy skin with fine, white scales affecting the whole body including scalp.	Rare – occurs in 1 in 300,000 babies	Tightness of the skin may prevent the eyes from closing properly, and restrict the movement of fingers.
Lamellar ichthyosis (LI)	"Collodion baby" (see <i>Caring for a Collodion baby</i> leaflet) refers to the appearance at birth and is followed by scaling all over the body and scalp, with particularly large brown scales on the limbs and trunk. The skin is not usually red but may be itchy.	Rare – occurs in 1 in 100,000 babies	Tightness of the skin may prevent the eyes from closing properly, and restrict the movement of fingers.
Harlequin ichthyosis (HI)	Very severe form, apparent at birth with thick plates of scale affecting the whole body including face. Gradually the thick scales are shed leaving an appearance like severe CIE (see above)	Extremely rare – possibly only 3 cases per year in the UK	This type is so severe at birth that sadly in some cases babies may not survive. The thick tight skin can occlude nostrils and ears, and prevents the eyes from shutting. Fingers and toes are tightly constricted.

Autosomal recessive congenital ichthyosis (ARCI) is a collective term applying to Lamellar ichthyosis (LI), Non bullous congenital ichthyosiform erythroderma (NBCIE) and Harlequin ichthyosis (HI).



People who care about ichthyosis

Living with ichthyosis

The congenital (inherited) forms of ichthyosis tend to persist throughout life, although the symptoms may become milder as time goes on. If you have a child with ichthyosis you may need to help them deal with people's reaction to the appearance of ichthyosis, or if you have the condition yourself you may have experienced unhelpful reactions first hand – this can seem hostile and unsympathetic and while it often stems from ignorance, the effect on you or your child should not be underestimated. Two of the most difficult times are when a child starts school, and potentially has to deal with the staring and teasing on their own for the first time, and during the teenage years when it is so important not to be different from your peers. As an adult you may also experience difficult times when starting college, university, a new job or starting new relationships. The ISG has two leaflets – 'Growing up with ichthyosis' (for teenagers and young adults) and 'What's it like to have ichthyosis?' (for younger children) to address some of these issues. There is also a leaflet that you can give to teachers, group leaders and others who may care for your child to help explain a bit about ichthyosis and the things that they need to watch for.

The ISG exists to help people like you and children with ichthyosis, by providing opportunities to meet other families and individuals with the condition to share experiences, and hints and tips for managing the condition. You may also find our leaflets on dealing with healthcare professionals and how to explain the condition to teachers, childminders etc useful.

Please contact the ISG for more 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/ichthyosisupportgroup](https://www.facebook.com/ichthyosisupportgroup)

Twitter:

twitter.com/ISG_Charity