

ICHTHYOSIS

What is ichthyosis?

Ichthyosis is a term used to describe continual scaling of the skin. It comes from a Greek word “ichthys” which means fish although not all affected people have fish-like scales. It is quite rare and can be inherited (genetic or congenital) or develop later in life (acquired). The inherited forms of ichthyoses are usually evident at or soon after birth and they tend to persist throughout life although many types improve with age.

The scaling of ichthyosis affects most if not all areas of the skin and is fairly consistent over the years. This is in contrast to other skin disorders such as eczema and psoriasis where scaling affects limited areas of the skin and changes its pattern frequently. However the more common, milder forms of ichthyosis do generally improve in summertime. Several of the rarer types of inherited ichthyosis also cause red inflamed skin and one form produces blistering of the skin in babies.

There are a number of rare genetic diseases which affect several systems of the body and also cause ichthyosis, these are referred to as ichthyosiform syndromes. The main forms of inherited ichthyosis are as follows:-

- **Ichthyosis vulgaris**
The most common form and usually quite mild with fine scaling or roughness on the arms and legs. It is more common in people with atopic (allergic) eczema and improves in warm weather.
- **X-linked recessive ichthyosis**
Occurs in boys and men and causes brownish flat scales most obvious on the arms, legs and tummy. It varies in severity and some affected babies have a difficult delivery. It is passed on through the female line.
- **Non-bullous ichthyosiform erythroderma and Lamellar ichthyosis**

These are very rare, often severe with prominent scaling and redness of most of the skin. Affected babies are often born with a shiny waxy second skin (collodion membrane) which sheds in a few days.

- **Bullous ichthyosis**

This is another rare red ichthyosis which causes blistering, fragile skin early in infancy and thick scaling, especially around the joints later on.

- **Harlequin ichthyosis**

This condition is fortunately an extremely rare but severe ichthyosis which causes thick plates of scale and severe complications at birth. Many affected babies do not survive.

- **Netherton's syndrome**

Is made up of red inflamed scaly skin from birth, fragile spiky hair and difficulty gaining weight in early childhood.

- **Sjogren-Larsson syndrome**

Another condition that also produces scaling and thickening of the skin which may not be obvious until a few months of age. Affected babies also have a type of cerebral palsy.

- **Conradl-Hunermann syndrome, Neutral lipid storage disease and KID syndrome**

These are very rare and associated with other medical problems.

Treatment of ichthyosis

The milder ichthyoses can be well controlled using regular moisturisers, that is bath oils, soap substitutes and moisturising creams. They tend to improve in warm weather and on holiday and may gradually lessen in adult life.

The severe forms may make affected babies quite unwell and treatment in special care baby units will help them get over their problems. Moisturisers are again important as ongoing treatment and antibiotics may be needed from time to time. A group of drugs called retinoids may be tried as they lessen scaling in many forms of ichthyosis. The complicated varieties of ichthyosis will need additional treatments according to their problems.

What causes inherited ichthyosis?

They are due to faulty copies of genes which are passed on from one generation to the next. Each type of ichthyosis is due to a different genetic mutation and the pattern of inheritance varies. Therefore expert genetic advice will be needed by families with affected children.

Recent research had led to the discovery of a particular mutation of the gene which controls a skin protein called keratin in one type, bullous ichthyosis. Other times of

ichthyosis have been shown to be related to abnormalities in the lipid or fat chemicals in the skin (such as X-linked recessive ichthyosis, Refsum's disease and Sjorgen-Larsson syndrome). No doubt further advances will be made and the hope is that it will be possible to improve treatment and prevent the occurrence of the severe form of ichthyosis.