Ichthyosis Support Group

People who care about ichthyosis

X linked ichthyosis

What is X-linked Ichthyosis (XLI)?

'Ichthyosis' means dry, scaly skin, which may occur as a result of an inherited genetic condition. X-linked ichthyosis (XLI) is the second most common form of ichthyosis; the most common type of dry skin is called ichthyosis vulgaris. XLI affects approximately 1 in 6000 boys/men. It only usually occurs in males because it is caused by a genetic change on the X chromosome (see below "how is the condition inherited").

What are the signs?

The severity of XLI varies. Babies with XLI may show no problem with their skin at birth but they may develop areas of scaling within the first few days, weeks or months of life. Most patients develop dry scaly skin by the age of 1 year and have the condition for the whole of their life.

The scales in XLI tend to be brownish in colour. They are up to a few millimetres in size and may occur all over a patient's body, particularly on the trunk, on skin at the back of the neck and on the outer parts of the arms and legs. XLI patients may find that the skin scaling is worse in winter than in summer because cold and dry weather tends to aggravate the condition.

Sometimes XLI is associated with eye changes (corneal opacities) or testicular problems (undescended testicles) or rarely a delay in intellectual development.

Ladies who are carriers of XLI do not have skin problems but they may have a long or difficult labour during childbirth.

How is the condition inherited?

RXLI is an inherited (i.e genetic) disorder so it runs in families. As the name suggests, RXLI is linked to the X chromosome (sex chromosome). It is a recessive condition. This means that males may develop the disease but females are carriers of the disease. Men only have one X chromosome, so a baby boy with only one copy of the faulty gene will develop XLI. Women have two X chromosomes, so if a baby girl has one faulty copy of the XLI gene she is a carrier, but she is protected from developing the condition because her other X chromosome is normal.

A mother with one copy of the faulty gene is a carrier of RXLI and she can give birth to an affected son. Some (approximately 1 in 2) of her female babies will also be carriers. If the father has XLI, he cannot pass this on to his son, but his daughters will be carriers of XLI. Families affected by XLI may wish to discuss these chances of passing on the condition with their doctor or a specialist before expanding their family.

What genes are affected?

XLI is caused by changes in the *STS* gene which codes for an enzyme called steroid sulphatase. XLI patients have a lower level of STS in cells throughout their body. This is particularly important in the skin, where STS regulates the chemical changes that allow dead skin to be shed from the body. When this enzyme is lacking, the dead skin cells build up as brown scales.

Low STS in the placenta is thought to cause long, difficult labour in some ladies who carry an *STS* mutation.

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How is the condition diagnosed?

A doctor, for example a skin specialist doctor (dermatologist), will look for the characteristic skin changes seen in XLI.

Blood tests may be used to confirm the diagnosis.

- A blood test can be taken to check the level of steroid sulphatase (STS). The STS level is very low or completely absent in a boy or man who has XLI. Ladies carrying XLI also have lower STS levels than normal.
- A blood test can also be taken for genetic analysis at a national reference laboratory to check for changes in the *STS* gene which lead to XLI.
- Prenatal diagnosis of XLI is now available so that changes in *STS* can be detected very early in pregnancy. However these procedures are not routinely provided, so families affected by XLI are advised to discuss any concerns with their doctor.

Why is X-linked Ichthyosis sometimes misdiagnosed?

It is quite common for newborn babies with XLI to have healthy-looking skin at birth, making immediate diagnosis of XLI difficult. Mild scaling does generally appear within the first few days following birth, but these signs may fade within a few weeks and may not appear again as true ichthyosis until later in life. Dry skin can also be a symptom of many other skin conditions, such as the more common ichthyosis vulgaris, or eczema or, rarely, an immune deficiency disease.

Looking after X-linked Ichthyosis

As yet, there is no cure for XLI but continual lifelong care with moisturisers and other creams/ointments should help keep the skin as healthy as possible. Success of the treatments will depend on individuals and what works for one person may not work for another. Patients will need to consider different options to find the best for them.

Moisturising creams and skin treatment

It is very important that the skin is kept moisturised and that suitable emollients are applied frequently throughout the day. More greasy treatments, such as ointments, may be needed in the winter as this is when the skin tends to be at its driest.

Creams or ointments containing lactic acid or urea may help to dissolve the visible scales on the skin of patients with XLI. However, these products can cause a baby's or child's skin to sting and so should be used with caution. The use of bath oils is important as bathing does not then cause drying or irritation of the skin and these are suitable for long-term use. Soaking in the bath for a period of time also tends to be more beneficial than a shower.

In most cases, XLI is not considered severe enough to prescribe retinoid tablets as these treatments can have side effects. It is important to seek advice from a specialist dermatologist before using any new treatment for XLI to check that it is safe.

More specific information on caring for the skin in XLI and other forms of ichthyosis can be found on the Ichthyosis Support Group website at www.ichthyosis.org.uk.

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Treatment of associated conditions

About half of males with XLI and some females who carry the condition develop cloudy spots in their eyes called “corneal opacities.” These are usually identified by an ophthalmologist (eye specialist) but they do not affect eyesight.

STS plays an important role in the placenta, so some female carriers of XLI experience difficulties in labour, such as a slowed birth. Carriers of the condition should let their obstetrician know beforehand so they can be prepared for the possibility of a prolonged labour.

In rare cases, the genetic change that results in XLI may affect other genes nearby, if a large section of DNA is deleted. Delayed growth, delayed intellectual development and delayed puberty have all been associated with XLI. If you have concerns about these conditions with XLI, the patient should be assessed by a specialist doctor.

Personal care

Although scaling in XLI is usually less severe than some forms of ichthyosis, the brown appearance of these areas can cause a child’s skin to look “unwashed.” This may lead to teasing or bullying by other children during school years if they do not understand the condition and can make it hard for the patient to build self-confidence. Consequently, extra support may be needed and general practitioners need to understand the psychological impact of the condition and provide adequate support to the family. This may require the involvement of other healthcare professionals.

Further help

There are a number of online forums about Ichthyosis where individuals can share their experiences and detail their own treatment recommendations. Not everything will work for everyone but these forums are a good place both to receive and offer support.

Contact the Ichthyosis Support Group for 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:

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PO Box 1242, Yateley GU47 7FL

Facebook:

facebook.com/ichthyosissupportgroup

Twitter:

twitter.com/ISG_Charity