Bullous Ichthyosis
(also called Bullous ichthyosiform erythroderma (BIE), Epidermolytic hyperkeratosis (EHK) or Epidermolytic ichthyosis (EI).

What is ichthyosis?
Ichthyosis describes dry, thickened, scaly or flaky skin. There are at least 28 different ichthyosis subtypes, which are mainly inherited (have a genetic cause).

What is Bullous ichthyosis?
This is a rare genetic skin disorder affecting less than 1 in 100,000 though it is likely that some mild cases are not recognised. It is characterised by blisters, skin fragility and ichthyosis.

What does it look like?
From birth the skin is noted to be fragile with blisters and peeling. Often there is no evidence of ichthyosis at birth and the skin appears red with superficial erosions. From early childhood the skin becomes more scaly and the redness and blistering less noticeable. The skin thickening can affect any part of the body but is most prominent on the scalp, around the neck and in the skin creases of the armpits, elbows and knees. Many patients with this condition develop thickening of the skin of the palms and soles. Older children and adults suffer from repeated skin infections especially in the skin folds.

It is possible that one of the parents may have a dark warty birth mark usually in a line, which may be the only expression of this disorder. A sample of skin examined under the microscope shows the same changes as the full blown condition. It is therefore important that both parents are carefully examined.

What is it caused by?
Bullous ichthyosis is caused by an abnormality of one of the many proteins in the skin, keratin. There are a number of different types of keratin and this condition is related to an abnormality in keratins 1 and 10. The specific gene site has been identified.

How is it inherited?
It is inherited as “autosomal dominant”, which means that there is a 50/50 risk for each child, if one parent is affected. At least half the patients have no family history and represent new mutations, although the parents must be carefully examined to make sure they do not have any sign of the condition however minimal.

Can it be diagnosed pre-natally if I want to have another child?
It may be possible to test the baby using molecular genetic techniques by taking a biopsy from within the uterus early in pregnancy, but this is not routinely available. This would need to be discussed with your dermatologist well in advance.

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What are the complications?
- repeated infections
- an unpleasant odour from the skin
- skin fragility and painful cracks in the skin creases
- thickening of the palms and soles is often a problem
- psychological upset with difficulties at school and work

Managing bullous ichthyosis
As yet, there is no cure but continual lifelong care with moisturisers and anti-infection treatments 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 at all times and that greasy, moisturising creams and ointments are applied frequently throughout the day. Moisturising agents should be perfume-free and without additives to avoid any allergic reactions. Bath oils are important so that bathing does not cause drying or irritation of the skin. Bathing is considered to be more beneficial than showering.

Emollients and keratolytic (anti-scaling) creams which contain urea, and/or lactic acid and propylene glycol (see product’s ingredients label for these) can be used to keep the skin as moist and hydrated as possible, however keratolytics can cause irritation if applied to inflamed and/or broken skin.

Steroid creams should be avoided as these do not help the skin.

Antibiotics are also important to treat secondary skin infections and gentle antiseptics can provide the skin with additional protection. Washing with an emollient containing an antimicrobial agent may help minimise the risk of skin infection.

It is important to check with your doctor or pharmacist before trying any new treatment. It is advisable to patch test any new cream for sensitivity before general use, and even “herbal” and “natural” creams may contain unsuitable ingredients.

Oral treatment (tablets) may be needed in severe ichthyosis. A group of drugs known as retinoids (synthetic vitamin A derivatives) are sometimes used (e.g. acitretin). They can reduce the thickness of the scale and help improve the appearance by reducing the overactive growth of the outer skin layer. Although often effective, they have a number of side-effects which should be fully discussed and considered before starting the tablet. Monitoring blood tests are needed while taking retinoid tablets, and pregnancy must be avoided as the drug can severely damage any unborn baby (teratogenic).

Non adherent dressings can be helpful in the treatment of fragile skin and blisters in Bullous ichthyosis.

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

Eye treatment
Some patients find that using moisturising eye drops can help prevent the eyes from becoming too dry. An ophthalmologist (eye doctor) may be contacted if ectropion (everted eyelids) is preventing the eyelids from closing at night but surgery for this condition is not generally recommended.

Ear treatment
Patients may experience a build up of dead skin in their ears and regular appointments with the Ear, Nose and Throat department at the local hospital should help to prevent problems such as potential impaired hearing.

Physiotherapy
Patients with Bullous ichthyosis may have problems with flexibility in their fingers, known as contractures. This is when the elastic tissue in these appendages is replaced with non-elastic tissue. Some individuals find that physiotherapy and splinting at night can help relieve this problem.

Personal care
Apart from keeping the skin hydrated and moisturised at all times, patients may get tired and overheated quickly when doing active sports (e.g. running, football etc). They may need to rest more than other individuals and teachers should be aware of this so they do not push children to continue participating. Individuals should be given the flexibility to access fluids throughout the day and not just when participating in sporting activities.

Peeling skin, particularly on the face can be difficult for building self-confidence so individuals may need extra support. General practitioners need to understand the psychological impact of the condition and provide adequate support to the family, which may require the involvement of other health professionals.

Individuals may experience thick scaling on the scalp and patients should manage their scalp to lessen the scaling where possible. Please see our leaflet for advice about how to look after your scalp.

Further help
There are a number of online forums about BIE where individuals can share their experiences and detail their own treatment recommendations. Not everything will work for everyone but they can be 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:**
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