



Bullous Ichthyosis

(also called bullous ichthyosiform erythroderma (BIE) or epidermolytic hyperkeratosis)

What is it?

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.



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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.

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

Treatment

Moisturising creams and a bath oil. It may be helpful to use a bath oil with an antiseptic from time to time.

Antibiotics when necessary.

Metronidazole gel is often used for weeping skin in the flexures to treat “anaerobic” infections.

Acitretin or neotigason is a retinoid drug taken by mouth which is related to vitamin A and can be very helpful for this condition. It is best given at a low dose. It helps by lessening the thickened skin, but care needs to be taken in getting the right dose for a particular patient as it can sometimes worsen the blistering.

Psychological support/counselling. It is essential to have a sympathetic GP and dermatologist.



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October 1998

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