



DYSTROPHIC EPIDERMOLYSIS BULLOSA

What are the aims of this leaflet?

This leaflet has been written to help you understand more about dystrophic epidermolysis bullosa. It tells you what it is, what causes it, what can be done about it, and where you can find out more about the condition.

What is dystrophic epidermolysis bullosa?

Dystrophic epidermolysis bullosa (DEB) is a rare inherited skin disorder.

The skin of those who have DEB is more fragile than normal. Minor injury causes blisters which often leave scars when they heal. Usually DEB is fairly mild, causing little more than minor inconvenience, but occasionally it can be severe, affecting the mouth, gullet and eyes in addition to the skin. DEB is not an infection, it is not contagious and it is not due to an allergy.

There are other types of epidermolysis bullosa (EB) - the simplex and junctional forms - but if you have DEB, you will *not* develop another type of EB at a later date.

What causes dystrophic epidermolysis bullosa?

The two outermost layers of skin, the epidermis and the dermis, are held together by a variety of proteins. In DEB, there is a genetic fault in the structure of type 7 collagen, one of the most important of these proteins. Weakened type 7 collagen is unable to join the epidermis and dermis together as firmly as it should. When the skin in people with DEB is rubbed, the two layers of skin separate, and fluid accumulates in the gap between them, forming a blister.

Is dystrophic epidermolysis bullosa hereditary?

Yes. Faults in the structure of type 7 collagen are caused by mutations (spontaneous changes) in the gene which carries the instructions for its assembly. A number of such mutations (some *dominant*, some *recessive*) have been identified in DEB: some are common but others are only found in individual families. Both males and females can be affected.

Everyone has two genes controlling the production of type 7 collagen, one inherited from each parent. In the *dominant* type of DEB, only one of the two genes is faulty. Anyone who has dominantly-inherited DEB, male or female, can pass the condition on to his or her children. There is a 50% (1 in 2) chance that each child of an affected parent will inherit the blistering tendency.

Less frequently, DEB is inherited in a *recessive* fashion. The parents of a person with recessively-inherited DEB usually have normal skin. Each unaffected parent has one normal and one abnormal type 7 collagen gene, and is able to make enough normal type 7 collagen to compensate for the weakening effect of the faulty gene. A child with recessive DEB has two abnormal type 7 collagen genes, one inherited from each parent, and is unable to make any normal type 7 collagen. Each time the parents of a child who has recessively-inherited DEB have a further child, there is a 1 in 4 chance that the new baby will also have the condition.

It is unlikely that anyone suffering from recessive DEB will have affected children. This could only happen if the sufferer's partner also carried a faulty type 7 collagen gene: as DEB is rare, the risk of this occurring is very small indeed.

As there are other, more unusual, inheritance possibilities, detailed genetic testing may be necessary before appropriate genetic counselling can be offered.

What are the symptoms of dystrophic epidermolysis bullosa?

Minor trauma causes painful blisters which tend to leave scars as they heal. The most vulnerable sites are the knees, ankles, elbows and knuckles. In dominant DEB, blisters sometimes occur at or shortly after birth, but usually they appear for the first time in early childhood. The normal rough and tumble of childhood often causes blistering, but in adults with dominant DEB blisters are usually infrequent. Most adults affected by dominant DEB have thickened great toe nails and sometimes other nails are also affected. In those who are more severely affected, finger and toe nails may be permanently lost. If blisters occur within the mouth, brushing teeth can be painful, leading to poor dental hygiene and dental caries. Constipation is quite common.

A variety of other problems are seen mainly in recessively-inherited DEB, but occasionally in those with DEB of dominant inheritance. These problems may include difficulty with swallowing, some restriction of mouth opening and tongue protrusion, and difficulty straightening the fingers fully. Those with recessive DEB usually have more fragile skin than sufferers of dominant DEB, and may develop severe scarring. Blisters can be induced by even the most gentle skin contact: affected babies need careful handling, and special feeding techniques may be necessary.

Skin cancer arising in areas of scarred skin is a possibility, especially in those individuals who have the most severe form of DEB. Regular examinations by a

dermatologist are important to ensure that skin cancer is detected and treated at an early stage.

What does dystrophic epidermolysis bullosa look like?

Blisters vary in size. They are fragile and easily damaged leaving raw moist areas which require dressings. Healed blisters leave pinkish- purple scars (which gradually become fainter) and *milia* (small pin-head sized creamy-coloured bumps). The nails may become unsightly (thickened and discoloured) or even permanently lost.

How will dystrophic epidermolysis bullosa be diagnosed?

If there is a family history of the condition, the diagnosis is usually straightforward. When the diagnosis is not clear, it may be appropriate to remove a small piece of skin (a biopsy) for detailed testing. For some individuals, it may be possible to distinguish dominant and recessive inheritance patterns by genetic analysis, requiring a blood test.

Can dystrophic epidermolysis bullosa be cured?

In the past 20 years, there has been rapid progress in our understanding of DEB, but at the moment it cannot be cured. Several laboratories around the world are exploring strategies which hopefully will ultimately lead to a cure, although this research may take many more years to come to fruition.

How can dystrophic epidermolysis bullosa be treated?

Careful choices of clothing and lifestyle, so as to reduce friction and protect vulnerable areas of skin, will reduce the number of new blisters, but, inevitably, some blisters will still occur. It is usually recommended that blisters are burst with a sterile needle. Antiseptic soaks and creams should be used to prevent infection. Often, dressings are necessary. Ordinary sticking plasters should be avoided as their removal usually tears the skin. Dressings with a silicone skin contact are usually best as they are easy to remove without damaging fragile skin.

If the eyes are affected, simple lubricant ointments are helpful. Good oral hygiene and regular dental examinations are important to help prevent dental decay. Plenty of fruit and vegetables will help prevent constipation, but sometimes gentle laxatives are necessary. Those who are more severely affected should be advised by a dietician.

In severe DEB, physiotherapy can help prevent restriction of movements, but sometimes surgery to free fingers encased in scar tissue may be worthwhile. If a child finds swallowing difficult, a minor operation called a *gastrostomy* (which involves making a passage to the stomach through the skin) can allow delivery of nutrients directly into the stomach. In adults, simple stretching of the oesophagus (gullet) is usually sufficient to improve swallowing.

What can I do?

A healthy diet, regular dental checks as soon as the first teeth appear and careful skin care are important. If you have an affected child, tell the teachers about DEB and make sure they understand that your child may not be able to take part in some of the more physical activities of the school curriculum.

If any form of surgery is required, it is important to inform the surgical team of the diagnosis of DEB well before the date of the planned operation so that they can discuss the condition with your dermatologist or with the DebRA nurses: special precautions to care for the skin might be appropriate during anaesthesia and surgery, and the usual surgical dressings may not be suitable.

Where can I get more information about dystrophic epidermolysis bullosa?

Advice and practical support for EB sufferers is available from DebRA. This is a charitable organization that also funds EB research projects and produces a regular magazine with up-to-date information about new developments. The DebRA specialist nurses can visit people in their own homes to demonstrate skin care regimes and they are available for telephone advice. If necessary, the nurses will visit schools to talk to staff and students. DebRA staff can also offer advice and practical support to those applying for a disability living allowance and help with mobility.

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This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: its contents, however, may occasionally differ from the advice given to you by your doctor.

BRITISH ASSOCIATION OF DERMATOLOGISTS

PATIENT INFORMATION LEAFLET

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