



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. DEB can be mild, causing little more than minor inconvenience, but it can also 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.

Dystrophic epidermolysis bullosa (DEB) is divided into two major types depending on inheritance pattern: **recessive** dystrophic epidermolysis bullosa (RDEB) and **dominant** dystrophic epidermolysis bullosa (DDEB). Each type is further subdivided into multiple clinical subtypes.

DEB is different from the other forms of epidermolysis bullosa (EB), which include epidermolysis bullosa simplex, junctional epidermolysis bullosa and Kindler Syndrome. Individuals who have DEB will not develop one of the other types of epidermolysis bullosa 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 (mutation) in the structure of type 7 collagen, one of the most important of these proteins. A number of such mutations have been identified in DEB; some are common

but others are only found in individual families. The mutations lead to reduced, altered or absent type 7 collagen which is unable to join the epidermis and dermis together as firmly as it should. When the skin in people with DEB is rubbed, even slightly, the two layers of skin separate and fluid accumulates in the gap between them, forming a blister.

Is dystrophic epidermolysis bullosa hereditary?

Yes. DEB may be inherited in a recessive or dominant way. Both males and females can be affected.

Everyone has two genes which control 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 themselves 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 even rarer.

In order to know the exact DEB type detailed genetic testing is usually 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, reduced growth of hair on the scalp, some restriction of mouth opening and tongue protrusion, tooth decay, and difficulty straightening the fingers fully due to scar tissue formation after repeated blistering. 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. Growth can be affected in severely affected individuals, and some individuals develop anaemia, eye problems and osteoporosis.

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?

The extent and severity of skin involvement depends on the type of DEB. Blisters vary in size. They are fragile and easily damaged leaving raw moist areas which require dressings. Healed blisters leave scars and milia (small pin-head sized white-coloured bumps). The nails may become unsightly (thickened and discoloured) or even permanently lost.

How will dystrophic epidermolysis bullosa be diagnosed?

Specialised investigations are usually necessary to make the diagnosis of DEB. Different types of EB may look very similar during the early months of life. If there is a family history of the condition, the diagnosis is usually straightforward.

Investigations usually involve removing a small piece of skin from the affected person. This is a simple procedure involving an injection of local anaesthetic into the skin. The skin sample will then undergo a number of detailed tests.

Blood samples will be taken from the affected person and if possible from both parents for genetic analysis. Early pregnancy prenatal diagnosis of DEB, at 8-10 weeks gestation is possible for families in whom the causative genes have been identified.

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?

Specialist teams including doctors, nurses, dentists and physiotherapists work together to help those affected by DEB. 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 reduce infection. Often, special 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. Adequate pain relief is important.

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.

Although not yet available in routine practice, current research work indicates that gene therapy could be a realistic goal for the treatment of severe DEB in the future.

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: it is important to warn the surgical team that their usual dressings and skin care regimes may be unsuitable for DEB sufferers. Careful handling and anaesthetic precautions may be required.
- It can be helpful to carry a special wallet-sized information card giving details of DEB and the relevant precautions to be taken should you or your child need help in an emergency. These cards are available from DEBRA.

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 organisation 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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Telephone: 01344 771961
Fax: 01344 762661

Website (UK): www.debra.org.uk

Website (international): www.debra-international.org

Other useful websites:

<http://ghr.nlm.nih.gov/condition/dystrophic-epidermolysis-bullosa>

<http://www.ncbi.nlm.nih.gov/books/NBK1304/>

For details of source materials used please contact the Clinical Standards Unit (clinicalstandards@bad.org.uk).

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: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel

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