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 (see bottom of leaflet).

What is dystrophic epidermolysis bullosa?

Dystrophic epidermolysis bullosa (DEB) is a rare inherited skin disorder. The skin of people with DEB is more fragile than normal. Minor injury causes blisters or ulcers that may be slow to heal and often leave scars. DEB can be mild, but it can also be severe, affecting the mouth, oesophagus (gullet), eyes and nails in addition to the skin.
Dystrophic epidermolysis bullosa (DEB) is divided into two main types depending on how it is inherited: **recessive** dystrophic epidermolysis bullosa (RDEB) and **dominant** dystrophic epidermolysis bullosa (DDEB)..

There are other forms of inherited epidermolysis bullosa (EB), including epidermolysis bullosa simplex, junctional epidermolysis bullosa and Kindler Syndrome. These are all distinct skin diseases so people who have DEB do not develop other types of epidermolysis bullosa at a later date.

What causes dystrophic epidermolysis bullosa?

The outer layer of our skin (epidermis) is bound tightly to the underneath layer (dermis) by several different proteins. One of these proteins, called type 7 collagen, makes up tiny rope-like structures called anchoring fibrils. People with DEB do not make enough type 7 collagen in their skin because the gene that controls the production of this protein is faulty (genetic mutation). This makes their skin more fragile, as the epidermis is not held down tightly to the dermis

so the two layers split apart when the skin is rubbed. The gap between the layers can fill with fluid forming a blister, or the upper layer may lift off leaving a raw area of skin (ulcer). The same process can happen in the mouth, digestive tract and eyes, leading to ulcers and scars. Children with severe types of DEB are known as 'butterfly babies' because of their fragile skin.

Is dystrophic epidermolysis bullosa hereditary?

Yes. DEB is an inherited skin disease and there can be other family members affected, but not always. It is not infectious and cannot be caught. It may be inherited in a "recessive" or "dominant" way and affects both males and females.

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 but both carry an abnormal copy of the collagen 7 gene. A child with recessive DEB has two abnormal type 7 collagen genes, one inherited from each parent, and is unable to make any or enough 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.

A specialist doctor (clinical geneticist) can help arrange genetic tests for people with DEB and their families to find out who is carrying abnormal genes and advise about the chances of passing these on to their children.

What are the symptoms of dystrophic epidermolysis bullosa?

Minor trauma such as rubbing the skin 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. People with dominant DEB tend to get fewer blisters as they grow up. Skin wounds can become infected which leads to an unpleasant smell, so good skin hygiene is important.

Itching is common in DEB and this can cause problems, as scratching causes further skin damage and delays healing.

Blisters inside the mouth may lead to painful ulcers that interfere with eating and brushing teeth. Poor dental hygiene can lead to tooth decay. Constipation is quite common particularly if eating is limited or the anus (bottom) is affected.

Most adults with dominant DEB have thick big toenails and sometimes other nails are also affected. Severe DEB can lead to permanent loss of the nails.

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.

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 wounds which require dressings. Healed blisters leave scars and milia (small pin-head sized white-coloured bumps). The nails may become 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. There are many things to consider in the management of DEB and detailed information about this can be found on the DEBRA website (link below). Some of the more common areas include the following:

Practical measures: Careful choices of clothing and activities to reduce friction and protect vulnerable areas of skin, will reduce the number of new blisters, but, inevitably, some blisters will still occur. Schools should be informed of activities that need to be avoided and DEBRA nurses may be able to help and advice or if necessary arrange a visit to school premises.

Skin care: There are lots of different dressings available to use and the DEBRA nurses and dermatologists can advise on ways to look after any existing wounds or blisters. Over time individuals with DEB and their families often find dressings and techniques that work best for them. Pain control is important when changing dressings and there are many pain relief medications available and also other techniques such as guided imagery and distraction therapies may be useful, particularly during dressing changes.

If the eyes are affected, simple lubricant ointments are helpful.

Mouth and nutrition: Poor appetite, blisters in the mouth or pain on swallowing may be problematic, but good nutrition is very important to help wounds and blisters heal. So nutritional support and advice from dieticians can be invaluable. Oral hygiene and regular dental examinations are important to help prevent dental decay. If swallowing is severely limited in children with DEB, 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.

Mobility and complications of scarring: In severe DEB, physiotherapy can help prevent restriction of movements. Hand wrapping and special dressings in the spaces between the fingers may also be helpful to prevent wounds on adjacent fingers fusing together.

Special considerations: If any form of surgery is required, there are special measures that are undertaken to protect the skin and mouth of individuals with DEB; so it is important to let the surgical team know you or your child has DEB as they will then discuss with your dermatology team or DEBRA nurses who will advise the surgical team about careful handling and anaesthetic precautions which may be required.

Head lice, which are common in school children, can be problematic in individuals with DEB. The lice may be more stubborn to clear with conventional treatments, as they may hide under crusts on the scalp. If parents have any concern that their child with DEB has head lice it may be worth discussing with a DEBRA nurse or EB centre early.

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.
- 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.
- If you have any surgical procedures let the medical team know you have DEB as far in advance of your surgery date as possible, so they can plan with your dermatologist or DEBRA nurse (see above for more details)

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

https://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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