PATIENT INFORMATION LEAFLET

DYSTROPHIC EPIDERMOLYSIS BULLOSA

WHAT ARE THE AIMS OF THIS LEAFLET?

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

WHAT IS DYSTROPHIC EPIDERMOLYSIS BULLOSA?

Dystrophic epidermolysis bullosa (DEB) is a rare inherited skin disorder. It is one of the epidermolysis bullosa (EB) group conditions. These include junctional epidermolysis bullosa**,** epidermolysis bullosa simplex and Kindler EB. They are all distinct skin diseases, so people affected by DEB do not develop other types of epidermolysis bullosa at a later date.

DEB is not an infection, it is not contagious, and it is not due to an allergy. The skin of people affected by DEB is more fragile than normal skin. Minor injury causes the skin to blister (water filled bubble under the skin) and this can result in wounds that do not heal as in the normal way (chronic). These wounds usually heal slowly and often leave scars. In different patients DEB symptoms can range from mild to very severe. In severe cases, it affects not only the skin, but also the nails, mouth, oesophagus (food pipe) and eyes.

DEB is divided into two main types, depending on how it is inherited: **recessive** dystrophic epidermolysis bullosa (RDEB), which can be intermediate or severe, and **dominant** dystrophic epidermolysis bullosa (DDEB).

WHAT CAUSES DYSTROPHIC EPIDERMOLYSIS BULLOSA?

Our skin is made up of three different layers. The outer layer of our skin (epidermis) is bound (glued) tightly to the underneath layer (dermis) by several different proteins. One of these proteins, called **type 7 collagen**, makes up tiny rope-like (thread) structures called anchoring fibrils. People affected by DEB do not make enough type 7 collagen in their skin as the gene that controls the production of this protein is faulty (due to a genetic mutation). This makes the skin more fragile, as the layers not held together tightly allow the two layers to separate and slide against each other when the skin is rubbed. The gap between the layers can then become filled with fluid forming a blister, or the upper layer may peel off leaving a raw area of skin (ulcer or erosion). The same process can happen in the mouth, digestive tract, and eyes, leading to ulcers and scars. Children with severe types of DEB are commonly known as 'butterfly babies' because of their fragile skin.

IS DYSTROPHIC EPIDERMOLYSIS BULLOSA HEREDITARY?

Yes. DEB is a genetic skin disorder, and therefore it is inherited, and other family members could be affected, but this not always the case. Everyone has two sets of genes; one set is inherited from their mother and the second from their father. These inherited forms are either autosomal dominant or autosomal recessive.

Dominant dystrophic epidermolysis bullosa (DDEB): Having just one faulty gene will cause a child to develop the disorder. This faulty gene can be inherited from one

of the parents or be acquired “de novo” (newly acquired). If one of the parents is affected, there is 1 in 2 (50%) chance that their new baby will inherit the condition.

Recessive dystrophic epidermolysis bullosa (RDEB): The child will need to have inherited a faulty gene from both parents. If one parent passes on an abnormal gene, the second, good gene from the other parent is usually able to produce enough normal anchoring protein for the skin to be unaffected. The parents of a child affected by RDEB (who will be a ‘carrier’) will usually have one normal and one abnormal gene. This means they will have healthy skin and are usually unaware that they carry a faulty gene until after they have an affected child. Each time parents of an affected child conceive, there is a 1 in 4 (25%) chance that their new baby will have fragile skin. Both males and females can be equally affected.

A specialist doctor (clinical geneticist) can help arrange genetic testing for people affected by DEB and their families, and advise about the chances of passing this on to their children. Sometimes they can offer prenatal testing.

WHAT DOES DYSTROPHIC EPIDERMOLYSIS BULLOSA LOOK AND FEEL LIKE?

DEB is characterised by:

- Fragile skin that can easily be damaged leaving raw wet wounds that require dressings.
- Blisters of various sizes.
- Scars and milia (small pinhead sized white-coloured bumps) can appear when blisters heal.
- Blistering can also occur on the mucous membranes (mouth and eyes) and internally in the oesophagus (food pipe).
- Nails are fragile, and may become thickened and discoloured or even permanently lost.

Minor injuries like rubbing the skin can cause painful blisters which tend to leave scars as they heal. The most vulnerable areas are the knees, feet, elbows and hands. In the case of dominant DEB, blisters may appear at birth or shortly after, but typically they show for the first time in early childhood.

As people with dominant DEB grow older, they tend to have fewer blisters.

Skin wounds can become infected, resulting in an unpleasant smell and pain., It is therefore important to maintain good skin hygiene and manage wounds properly.

Itching is a common issue in DEB, and scratching can cause further skin damage and delay healing. Itching is usually caused by inflammation and dry skin, keeping the skin well-moisturised is key.

Blisters inside the mouth may lead to painful ulcers that interfere with eating and brushing teeth. Poor dental hygiene can also 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 affected too. Severe DEB can lead to permanent loss of nails.

In recessive inherited DEB, as well as occasionally in dominant inherited DEB, individuals may experience various problems. These problems can vary. Some of the common ones include:

- trouble swallowing,
- less hair growth on the scalp,
- difficulty opening the mouth and sticking out the tongue,
- tooth decay,
- trouble fully straightening the fingers due to scars from repeated blisters.

People with the recessive form of DEB usually have more delicate skin and may develop severe scars.
Skin cancer arising in areas of scarred skin is a possibility, especially in those individuals who are affected by the most severe form

of RDEB (recessive dystrophic EB). Regular examinations by a dermatologist are important to ensure that skin cancer is detected and treated at an early stage.

In summary, the extent and severity of skin involvement depends on the type of DEB:

- In the mildest forms of DEB, patients might only have thick, abnormal toenails and mild scarring of the toes and fingers. The nails may become thickened and discoloured or even permanently lost.
- In the severe forms of DEB, skin fragility, scarring, and milia are present from birth or early childhood. The lining of the oesophagus may be affected, resulting in swallowing difficulties. Blistering of the eyes and the mouth can also be present. Severe forms of DEB have a high risk of developing cancer.

HOW WILL DYSTROPHIC EPIDERMOLYSIS BULLOSA BE DIAGNOSED?

Specialised investigations are usually necessary to make the diagnosis of DEB. Different types of EB may look 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 called shave biopsy. This is a simple procedure involving an injection of local anaesthetic into the skin and using a blade to scrape the surface of the skin. The skin sample will then undergo a number of detailed tests trying to identify which skin protein is missing. 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 genes that cause this problem have been identified.

CAN DYSTROPHIC EPIDERMOLYSIS BULLOSA BE CURED?

In the past years, research on the treatment of EB has progressed rapidly, but at the moment there is no cure. Different laboratories around the world are exploring new surgical techniques, new treatments such as cell and gene therapies to help improve symptoms. All these treatments are currently undergoing trials, and some have shown promising results.

HOW CAN DYSTROPHIC EPIDERMOLYSIS BULLOSA BE TREATED?

There are four specialist centres, two for children and two for adults, in the UK. Specialist teams include doctors, nurses, dentists, dietician, occupational therapists, psychologists and physiotherapists who 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 ideas 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. However, inevitably, some blisters will still occur. Schools should be informed of activities that need to be avoided and your EB CNS (clinical nurse specialist) will 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 EB nurses and dermatologists can advise on ways to look after any existing wounds or blisters. Over time, individuals with DEB and their families usually find dressings and techniques that work best for them. Pain control is important when changing dressings and there are many pain relief medications available. Other techniques such as guided imagery and distraction therapies may also be useful, particularly during dressing changes.

When the eyes are affected, lubricant ointments and eye drops applied regularly during the day are helpful.

Mouth and nutrition: Poor appetite, blisters in the mouth, pain on swallowing, oesophageal narrowing (strictures), reduced mouth opening, and tongue movement can cause problems. Good nutrition is highly important to help wounds and blisters heal. Nutritional support and advice from dieticians are essential. Oral hygiene and regular dental examinations are important to help prevent dental decay. If swallowing is severely limited in children affected by a severe form of DEB, sometimes a minor intervention called oesophageal dilatation (a procedure to widen the food pipe) or a surgery procedure called gastrostomy (which involves inserting a tube, often referred to as a "G-tube" through the abdomen into the stomach) can improve nutrition and growth.

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, special measures are needed to protect the skin and mouth of individual. It is therefore extremely important to let the surgical team know you, or your child are affected by DEB. The surgical team can then discuss your case with your dermatology team and/or EB nurses who will advise about the careful handling and anaesthetic precautions which may be required.

Head lice, which are common in school children, can be problematic for children affected by 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 has head lice it may be worth discussing this with an EB nurse or EB centre straightaway.

SELF-CARE (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 in 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 are affected by DEB as far in advance of your surgery date as possible, so they can plan with your dermatologist and/or EB nurse (see above for more details).

WHERE CAN I GET MORE INFORMATION ABOUT DYSTROPHIC EPIDERMOLYSIS BULLOSA?

DEBRA is the national charity and patient support organisation for people living with or directly affected by all types of EB. DEBRA supports the EB community in the following ways:

Specialist healthcare

DEBRA works in partnership with the NHS to deliver an enhanced EB healthcare service which is vital for people living with EB. There are four designated EB centres of excellence around the UK providing expert specialist EB healthcare and support, as well as other hospital locations and regular clinics which aim to provide EB services to people wherever they are located.

Member services

The DEBRA Community Support team works with the EB community, healthcare,

and other professionals to improve quality of life for people living with EB. They offer support, advocacy, information, and practical help at every stage of life. The DEBRA Membership scheme includes holiday home respite, grants, and bespoke events to support people living with EB.

Research to find treatments

DEBRA supports research programmes that aim to find treatments that will slow, stop, or reverse the progression of EB. Repurposing drugs which are already available and successful in treating other inflammatory skin conditions is a key part of its research programme.

For more information or to apply to become a DEBRA member, which is completely free, please visit www.debra.org.uk/support-membership, email membership@debra.org.uk or call us on 01344 771961.

DEBRA House
13 Wellington Business Park
Dukes Ride
Crowthorne
Berkshire
RG45 6LS
Telephone: 01344 771961
Fax: 01344 762661
Website (UK): www.debra.org.uk
Website (international): www.debra-international.org.uk

Link for health care assistant:
https://www.debra.org.uk/specialist-eb-healthcare

Link for clinical guidelines:
https://www.debra.org.uk/clinical-practice-guidelines

There are four designated EB centres of excellence around the UK:

- Great Ormond Street Children's Hospital in London
- Birmingham Women's and Children's Hospital
- Guy's and St Thomas' Hospital in London
- Solihull Hospital

Please note that the British Association of Dermatologists (BAD) provides web links to additional resources to help people access a range of information about their treatment or skin condition. The views expressed in these external resources may not be shared by the BAD or its members. The BAD has no control of and does not endorse the content of external links.

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

BRITISH ASSOCIATION OF DERMATOLOGISTS
PATIENT INFORMATION LEAFLET
PRODUCED | JANUARY 2009
UPDATED | MARCH 2012, APRIL 2015, JUNE 2018, OCTOBER 2023
NEXT REVIEW DATE | OCTOBER 2026