



Great Ormond Street Hospital for Children NHS Foundation Trust: Information for Families

Mild dystrophic epidermolysis bullosa (DEB)

This leaflet explains about mild dystrophic epidermolysis bullosa (DEB) and how it can be managed. It also contains suggestions for making everyday life more comfortable and contact details for a support organisation.

What is epidermolysis bullosa (EB)?

EB is a group of inherited disorders in which the skin blisters extremely easily. There are four main types of EB. Each is a quite distinct disorder. If you have dystrophic EB then you cannot later develop one of the other forms of EB (simplex, junctional or Kindler syndrome). Dystrophic EB is so called because of the tendency to heal with scarring.

What caused it?

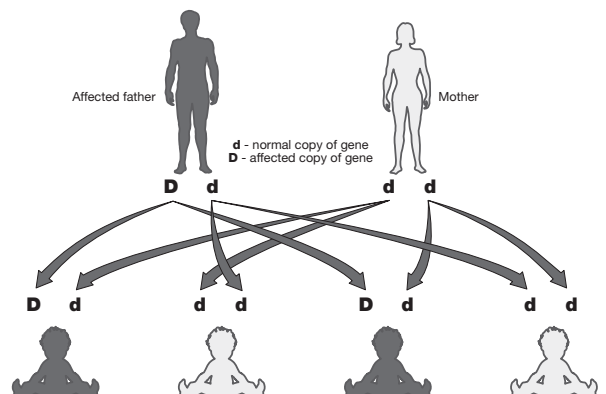
The problem lies in the genes that hold the instructions necessary for production of certain proteins in the skin. These instructions have a fault, rather like a typing error, with the result that the proteins are incorrectly formed, and unable to fulfil their role in attaching the layers of skin together.

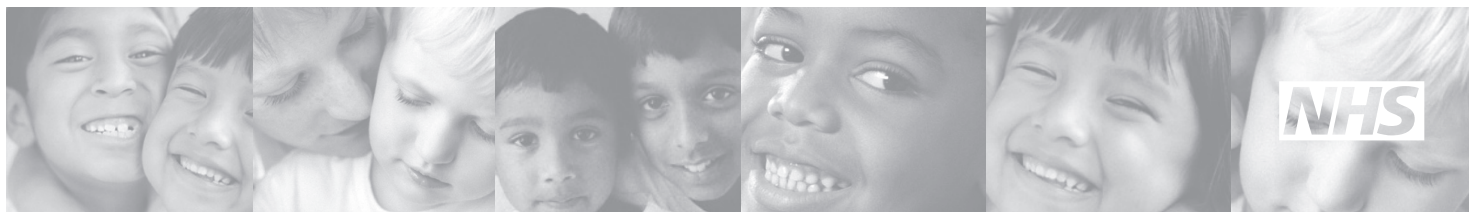
What are the different types of Dystrophic EB (DEB)?

Dystrophic EB (DEB) is found equally in males and females. There are two types of inheritance in dystrophic EB. These are called dominant and recessive:

Dominant dystrophic EB (DDEB)

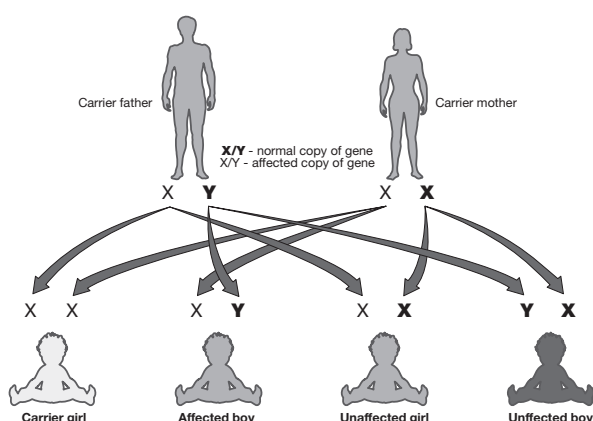
Everyone has two copies of every gene, one inherited from their mother and one from their father. In DDEB a defect in one of those genes can lead to fragile skin and blistering, even though the other gene is normal. This means anyone who has DDEB can pass the condition onto his or her children. Each time a pregnancy occurs there is a 1 in 2 chance that the child will inherit DDEB, if one parent is affected. However, DDEB can sometimes be seen as a 'new mutation' when there is no family history.





Recessive dystrophic EB (RDEB)

In RDEB, both copies of the gene have to be defective in order for the person to have fragile skin and blisters. A person with one defective copy of the gene is healthy and is said to be a carrier of the disorder. However, if two such people who carry a defective copy have children, there is a 1 in 4 risk that the child will inherit both defective copies of the skin and will have fragile skin and blisters. RDEB varies tremendously in severity as the gene responsible for the condition is very large, and the defect can occur on any part.



This short information leaflet will describe the care and management of DDEB and mild RDEB. Further information is available in a separate leaflet for children with severe forms of recessive dystrophic EB.

Is there a cure?

Not yet, but research continues. There is still a long way to go, but an effective treatment to prevent the skin problems may ultimately be possible.

How are DDEB and mild RDEB managed?

Skin

Blisters should be lanced with a sterile needle in order to prevent them from enlarging. The roof should be left on the blister. Where a knock or fall has removed the skin leaving an open wound, a dressing needs to be applied. There are several dressings which are suitable for children with mild dystrophic EB. Dressings can be secured using tubular or wrap around bandages or a silicone based tape. If a dressing with a sticky border is used an adhesive remover can be used to prevent damage to the skin.

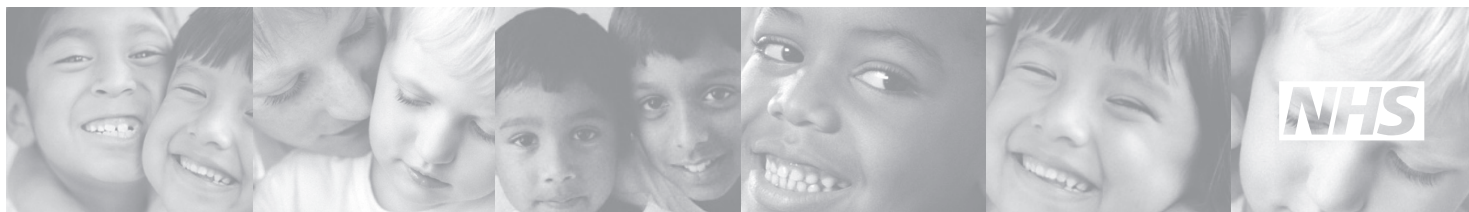
Wounds can become infected and so we recommend using an antiseptic cream or ointment if the wound is oozing pus or if the surrounding skin is red or swollen. If pain, redness and swelling persist, please see your family doctor (GP) as treatment with an antibiotic may be necessary.

Scarring

In those with DEB, the wounds and blisters tend to heal with a scar. Often little white raised spots are seen in the scar tissue. These are called milia and they eventually disappear and do not cause any problems. The scar tissue is fragile and for this reason we suggest padding vulnerable areas of skin.

Nails

Finger and toe nails can become thickened and difficult to cut. This happens when they become knocked and blister forms underneath the nail. We can recommend some creams to soften the nail and make it easier to cut. Sometimes



toenails come off following trauma; either immediately or some time later. They will usually grow back but will be thickened and misshapen.

The mouth and throat

Blisters are often seen in the mouth in those with mild DEB, but rarely cause problems with eating and drinking. The teeth are formed normally but the gums are fragile and can bleed when the teeth are brushed. We recommend using a soft toothbrush and seeing your dentist regularly.

There is a small risk that blisters in the oesophagus (food pipe) can develop and these may heal with a scar, which causes a narrowing or stricture. In order to prevent this, your doctor may prescribe medicines to neutralise the acid in the stomach. This means if stomach contents are refluxed up the oesophagus, which is very common in babies and young children, the skin lining the food pipe is protected from the acid.

Pain

Blisters and wounds can be painful. We recommend giving paracetamol (Calpol® or equivalents) or ibuprofen (Nurofen® or equivalents) for the control of mild to moderate pain. For more severe pain which is not helped by these medicines your EB nurses and doctors will recommend some stronger painkillers which can be prescribed by your GP.

Constipation

This is a very common problem in all types of EB; even those with mild DEB. Blisters around the anal region (bottom) can make it painful to open their bowels. Constipation then develops because the

child is frightened to poo in case it hurts. A good diet, high in fibre with plenty of fluids will help. Sometimes medicines are prescribed to help soften the faeces (poo) or stimulate the bowel.

Suitable dressings and recommended products

The EB Team produce a list of suitable dressings and recommended products, which is updated frequently or as new products become available.

What happens next?

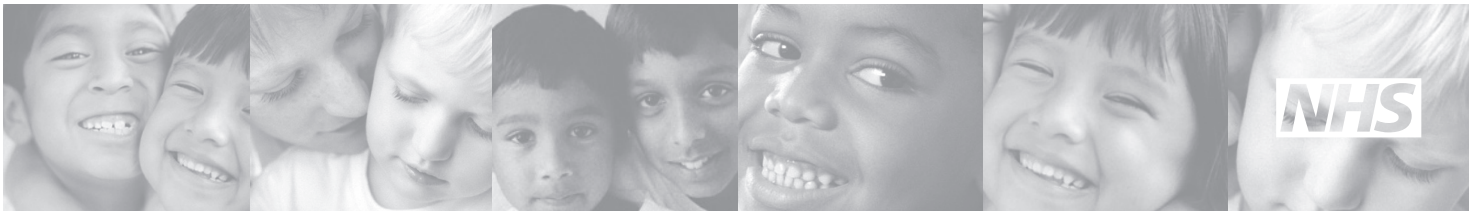
Children with Dystrophic EB will need to attend outpatient appointments at their EB specialist centre and then move on to adult services at around 16 years of age.

Further information and support

EB team at Great Ormond Street Hospital (GOSH) – 020 7829 7808

DEBRA is the national charity that supports people living and working with Epidermolysis Bullosa (EB) – a genetic condition which causes the skin to blister and shear at the slightest friction or even spontaneously.

DEBRA provides information, practical help and professional advice to anybody living or working with EB, including individuals, families, carers and healthcare professionals, and funds research into the condition. To find out more about how DEBRA can support you, please visit www.debra.org.uk.



Notes



Compiled by the Clinical Nurse Specialist for Epidermolysis Bullosa and DEBRA
in collaboration with the Child and Family Information Group at GOSH.

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www.gosh.nhs.uk