

# MILD DYSTROPHIC EPIDERMOLYSIS BULLOSA

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## **What is Epidermolysis Bullosa (EB)?**

EB is a group of inherited disorders in which the skin blisters extremely easily. There are three 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 or junctional). Dystrophic EB is so called because of the tendency to heal with scarring.

## **What is the cause?**

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.

## **How do you get it?**

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 on to 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 of the gene.

This short information leaflet will describe the care and management of DDEB and mild RDEB. DebRA produces a booklet outlining the care of children with a more severe form of RDEB.

## 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.

## Management

- Skin

Blisters should be lanced with a sterile needle in order to prevent them from enlarging. The roof should be left on the blister and the area dusted with cornflour to help it to dry up.

Where a knock or fall has removed the skin leaving an open wound, a dressing needs to be applied. We recommend Mepitel (Mölnlycke) directly to the skin, this should then be covered with Mepilex (Mölnlycke) where padding is required for protection, or Mepilex Transfer (Mölnlycke) on areas that require a thinner, more conformable dressing. These dressings are secured with a tubular bandage such as Tubifast.

Wounds on knees and elbows can be dressed with Mepilex Border, which is self-fixating and does not require a retention bandage.

All the described dressings are designed to be left in place for several days to allow healing to take place.

Mepilex, Mepilex Transfer and Mepilex Border can also be used to pad vulnerable areas such as ankles, knees and elbows.

- 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 as described above.

- The mouth and throat

Blisters are often seen in the mouth in those with mild DEB, but rarely cause problems with eating and drinking.

There is a small risk that blisters in the oesophagus (swallowing tube) can develop and these may heal with a scar that 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 (very common in babies and young children) the skin lining the tube is protected from the acid.

- 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 pass a stool. Constipation then develops because the child is frightened to open their bowels in case it hurts. A good diet, high in fibre with plenty of fluids will help. Sometimes medicines are prescribed to help soften the stool or stimulate the bowel.

What help is available?

DebRA is the charity which supports people living with EB, by providing information, practical help and professional advice through our nursing and social care teams.

DebRA keeps you informed through a twice yearly newsletter and annual conference open to anyone affected by EB.

Children with dystrophic EB are usually successful in claiming Disability Living Allowance. The Social Care Team can help you apply for this.

Many schools have not come across the condition so DebRA has produced a booklet for teachers and the EB nurses will visit schools if necessary.

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