Mild dystrophic epidermolysis bullosa (DEB): information for families

This information sheet from Great Ormond Street Hospital (GOSH) explains about mild dystrophic epidermolysis bullosa (DEB) and how it can be managed. It also contains suggestions for making everyday life more comfortable. Separate information is available for children with severe forms of recessive dystrophic RB.

Please note: This information sheet suggests some commercial products that can be helpful in managing epidermolysis bullosa – including them in this information sheet does not mean that they are recommended by GOSH and alternative products may be available.

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 causes Dystrophic EB (DEB)?

EB is a genetic condition. Genes determine characteristics such as eye colour and also our health. A genetic mutation means that a change has happened that makes the gene faulty.

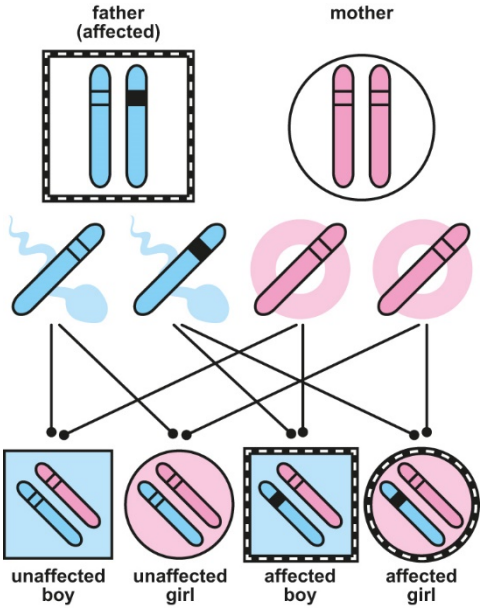
We have two copies of every gene, one from our father and one from our mother. Dystrophic EB (DEB) is found equally in males and females.

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.

There are two types of inheritance in dystrophic EB. These are called dominant and recessive:

Dominant dystrophic EB (DDEB)

In DDEB a defect in one copy of the gene 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 their children.



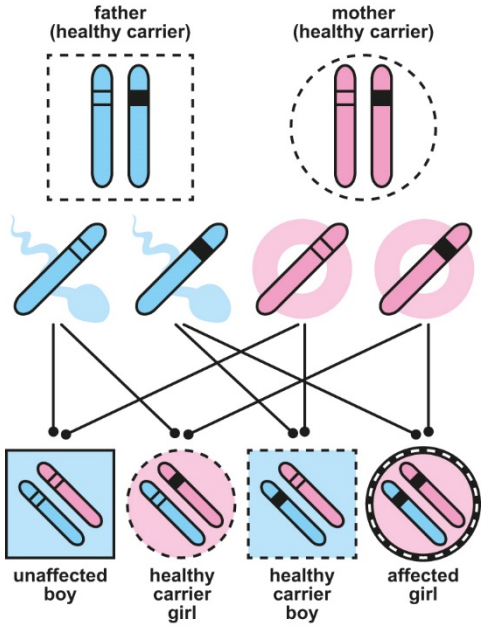
Each time a pregnancy occurs there is a 50 per cent (1 in 2) chance that the child will inherit DDEB, if one parent is affected.

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 people who carry a defective copy have children, there is a 25 per cent (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.



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

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 so 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 sometime later. They will usually grow back but will be thickened and misshapen.

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 or ibuprofen 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 bottom can make it painful to poo. 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.

Is it possible to test for EB before birth?

In most cases, prenatal testing is available for parents who are known to be carriers of junctional EB. The test is called chorionic villous sampling (CVS) and involves a small piece of the placenta being taken for genetic testing after the 11th week of pregnancy. It may also be possible to test before pregnancy is established using IVF techniques and testing the embryos before they are implanted.

Both tests are only possible if specific gene changes have been identified. In a very small minority where there is insufficient information to interpret the test, a later test (after the 15th week of pregnancy) may be offered. This involves analysis of a small piece of skin taken from the baby.

Further information and support

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

DEBRA is the national charity that supports individuals and families affected by Epidermolysis Bullosa (EB) – a painful genetic skin blistering condition which, in the worst cases, can be fatal. 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.