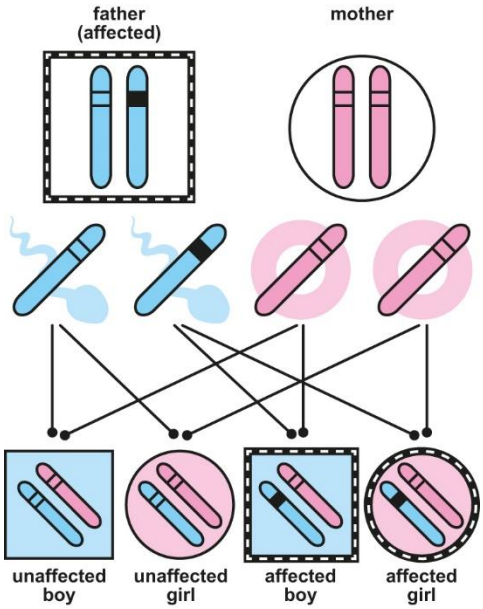
Epidermolysis bullosa simplex: localised and generalised types: information for families

This information sheet from Great Ormond Street Hospital (GOSH) explains about the localised and generalised forms of epidermolysis bullosa simplex and how they can be treated. It also contains suggestions for making everyday life more comfortable. The localised form was previously known as Weber Cockayne epidermolysis bullosa simplex and the generalised form Köbner epidermolysis bullosa simplex.

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 EB simplex then you cannot develop one of the other forms of EB (dystrophic, junctional or Kindler syndrome).



What causes EB simplex?

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. EBS is almost exclusively a dominantly inherited condition seen equally in males and females.

This means anyone who has EBS can pass the condition onto their children. Each time a pregnancy occurs there is a 1 in 2 chance that the child will inherit EBS if one parent is affected.

However, EBS can sometimes be seen as a ‘new mutation’ when there is no family history.

The problem lies in the genes that hold the instructions necessary for production of certain proteins in the top layer of skin. These instructions have a minor fault, rather like a typing error, with the result that the proteins are incorrectly formed, and unable to fulfil their role as scaffolding for the topmost layer of skin. The result is that the top layer of skin does not ‘stick’ securely to the layer beneath it, and where the two layers separate a blister develops.

How is EB diagnosed?

In the majority of cases, the type of EB can be determined by analysis of a skin biopsy (tiny sample of skin). Blood samples are also taken from both child and parents to look for the specific gene changes.

Is there a cure?

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

What are the different types of EBS?

Localised EBS

This is the most common type of EBS. Blistering is localised to the hands and feet. Blisters may not become evident until the child begins to walk.

Generalised EB

Blistering may be obvious from birth, or develop during the first few weeks of life. Occasionally babies are born with raw open wounds which need to be covered with special dressings. Sites of blistering correspond to areas where friction is caused by clothing and frequently appear around the edges of the nappy. Blisters are often seen inside the mouth but do not generally cause a problem during feeding.

EBS generalised severe type

A separate information sheet explaining the management of this type of EB is available.

How is EBS localised and generalised managed?

Management involves identifying new blisters, lancing them with a sterile needle or snipping with a sharp pair of scissors to release the fluid and prevent spread. If compressing the blister to expel the fluid is very painful, then the fluid can be drawn off using a fine needle attached to a syringe.

Many people find dressings unnecessary, preferring to dust the area with cornflour to help dry up the blisters and reduce friction. However, some people find non-stick dressings helpful.

Pain relief

Blisters can be very painful and limit mobility. Some find it helpful to take simple painkillers such as paracetamol and ibuprofen when their feet are especially sore. Longer-term pain management may be necessary – sometimes only in the summer months when blistering is at its most troublesome.

The EB medical team can offer advice on this to your family doctor (GP).

Constipation

This is a very common problem in all types of EB. 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 poo or stimulate the bowel.

Prevention of blisters

There are no measures that will totally stop all blisters forming, however the following actions can be carried out to minimise the risk:

- Heat, especially when associated with high humidity can exacerbate blister formation, so measures taken to keep affected areas cool will often reduce the rate of blistering.
    - Wear natural materials
    - Soak the affected area such as the hands and feet in cool water, especially after a hot day
    - Temperature regulation in house/classrooms

Choice of footwear is very important:

- As stated above, natural materials for shoes are good, such as leather.
- Check the insides of new shoes for excessive seams, as these could cause blisters.
- Children should get their feet measured to ensure good shoe fit.
- It helps to have several pairs of shoes of different styles and to change one pair for another regularly to alter sites of friction. It is helpful to have soft, well ventilated shoes.
- Shoes in line with the uniform policy of schools are often unsuitable and permission needs to be granted to wear more suitable footwear such as black trainers. A wide fit prevents contact of the shoe with the top and sides of the feet.Some people find special socks beneficial in keeping the feet cool. Many of these products can be supplied on prescription from your GP.
- Walking some days will not be a problem but other days can be difficult. Many people ‘save’ their feet to do activities that they enjoy. However, it is important to encourage your child to pace themselves during activities to avoid excess blistering as this will make the following days more difficult.

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.