EPIDERMOLYSIS BULLOSA SIMPLEX

What are the aims of this leaflet?

This leaflet has been written to help you understand more about epidermolysis bullosa simplex. It tells you what it is, what causes it, what can be done about it, and where you can find out more about it.

What is epidermolysis bullosa simplex?

Epidermolysis bullosa (EB) simplex is a rare inherited disorder in which the skin is fragile and blisters at sites of rubbing. It is mild in the usual form; blisters tend to be confined to the palms and soles, and are most troublesome during warm weather. In other types the blistering may be more generalised and occasionally blisters arise in the mouth. The most severe form is called generalised severe EB simplex (Dowling-Meara) and those affected have more widespread blistering which occurs throughout the year.

EB simplex is different from other types of EB which include junctional, dystrophic and Kindler forms; if you have EB simplex then you will not go on to develop these other types. EB simplex is not an infection, it is not contagious, and it is not due to an allergy.

What causes epidermolysis bullosa simplex?

The top layer of skin (the epidermis) is composed of layers of cells. Each cell has an internal support system formed from proteins (keratins), giving it strength and shape. In EB simplex, there is a weakness in one of these proteins (usually type 5 or type 14 keratin, but very occasionally other proteins called kelch-like family member 24, plectin or exophilin 5). This protein weakness causes affected skin cells to be more easily damaged. When the cells break, they separate from each other, fluid accumulates between them and a blister forms.

The protein weakness is because the DNA in one of the genes is changed (a mutation). Many gene changes can cause EB simplex. Some of these are common and some are only found in one or two families.

Is epidermolysis bullosa simplex hereditary?

Yes. It is usually inherited due to a *dominant* gene. This means that one affected parent can pass this the gene on to children. Sometimes a new change appears in the genes to cause EB simplex so that neither of the parents are affected. Anyone who has a dominant form of EB simplex can pass the condition on to his or her children. There is a one in two chance that the child of an affected parent will inherit EB. EB simplex affects men and women equally. There are also much rarer types of EB simplex due to a type of gene called a recessive gene. In this type neither parent has EB simplex but each carries an affected recessive gene. If both parents have the recessive gene then each child that they have has a one in four chance of having EB simplex if they inherit both affected genes.

What are the symptoms of epidermolysis bullosa simplex?

The main feature of EB simplex is blistering caused by friction. It is often painful and can sometimes significantly affect daily life and interfere with education and work.

The age at which skin blisters develop depends on the type of EB simplex. Most commonly, the blisters occur for the first time during early childhood. However, blisters may start at birth or in adulthood. In early childhood, blisters tend to occur at sites of friction. Friction sites include areas under the elasticated areas of a nappy or the hands and knees during the crawling stage.

Later, painful blisters tend to develop on the soles after walking only short distances. Writing with pens and pencils can cause blisters to appear on fingers, especially in children. Blisters may also appear under close fitting clothing such as waistbands, collars or the ribbing of socks. A few people find that hot food or hot drinks will cause blistering within the mouth.

The formation of blisters is generally worse during warm weather. A few individuals may be free of blisters during the winter. Secondary infection of the blisters can be a common problem.

Although blistering tends to be life long, it sometimes becomes less severe in adult life.

In the generalised severe variant of EB simplex, blisters occur for the first time at an earlier age, usually within a few days of birth. They can occur anywhere on the body and may be very extensive. The infant may be very unwell in the first few months of life but generally becomes much better after this. This form of EB simplex often improves dramatically after early childhood. In some, the blisters may disappear for a short while during a feverish illness.

What does epidermolysis bullosa simplex look like?

The blisters of typical EB simplex look the same as the blisters that anyone can get as the result of friction to the skin. However, although they vary in size, they tend to be small (up to 2 cm) and numerous but heal without leaving any scars. In the generalised severe variant, blisters tend to occur in clusters, often healing to leave residual pigmentation. Thickened skin on the palms and soles can be a feature, especially in the generalised severe type of EB simplex. Thickened deformed nails may also develop.

How will epidermolysis bullosa simplex be diagnosed?

In older children and adults, the diagnosis of EB simplex can usually be made on the basis of the individual's history (especially if other family members are affected) and the distribution of the blisters. Occasionally, if there is uncertainty about the diagnosis, particularly in infants your dermatologist may suggest taking a small sample of skin for more detailed microscopic examination. A blood test may be suggested to look for abnormalities of the genes likely to be involved.

Can epidermolysis bullosa simplex be cured?

Although during the past 20 years there has been rapid progress in the understanding of EB simplex, at the moment it cannot be cured. There is much research worldwide in the hope of leading to an effective treatment.

How can epidermolysis bullosa simplex be treated?

Your dermatologist and specialist nurse will explain the best way to manage your blisters. It is generally recommended that new blisters are burst with a sterile needle, available from your doctor.

In infants careful handling and application of non-sticky dressing will be required. In older children and adults often no dressing is necessary, however if one is required it is important to use a product that will not stick and damage the skin or cause further blistering when it is removed. Dressings with a silicone layer in contact with the skin are usually suitable as they are very easy to remove, and hydrogel dressings are known to help cool the blistered area and have a soothing effect.

Treatments to reduce sweating of palms and soles may be of benefit. Depending on the severity, input may be required from a number of different specialists including nurses, dermatologists, paediatricians, pain specialists, podiatrists and dieticians.

What can I do?

Choose soft, well-fitting, comfortable leather footwear and carry a spare pair of shoes to change into later in the day. Try to keep feet cool and limit walking and manual tasks to what you know from experience are safe. Clothing should not be tight fitting. Use of antiseptic washes may help reduce the risk of secondary infection.

If you have an affected child, tell their school/nursery about EB simplex and make sure they understand that your child may not be able to take part in some physical activities.

If your condition is very disabling, you may be able to obtain further help with mobility by applying for personal independence payments (PIP) (see below for further information). Local authorities can sometimes provide or fund transport to school for badly affected children. If you have a car, consider applying for a disabled person's car badge so that you can park as close as possible to your destination.

Where can I get more information about epidermolysis bullosa simplex?

Advice and practical support for EB sufferers is available from DEBRA.

DEBRA House
13 Wellington Business Park
Dukes Ride
Crowthorne
Berkshire, RG45 6LS

Tel: 01344 771961
Fax: 01344 762661
Web: www.debra.org.uk

Personal Independence Payments (UK Government)
Web:
www.gov.uk/pip
www.gov.uk/dla-disability-living-allowance-benefit

For details of source materials used please contact the Clinical Standards Unit (clinicalstandards@bad.org.uk).

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

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