PATIENT INFORMATION LEAFLET

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 explains what it is, what causes it, what can be done about it, and where you can find more information.

WHAT IS EPIDERMOLYSIS BULLOSA SIMPLEX?

Epidermolysis bullosa simplex (EBS) is a rare inherited disorder in which the skin is fragile and blisters easily in places where it is rubbed or scratched. The mild form of EBS tends to cause blisters only on the palms of the hands and soles. They are most troublesome during warm weather. In other rare types of EBS, the areas of blisters may be more generalised and occasionally develop in the mouth. The most severe form is called "severe EB simplex" (previously known as Dowling-Meara). People affected by this subtype have more widespread blistering which occurs throughout the year.

EBS is different from other types of EB which include junctional epidermolysis bullosa, dystrophic epidermolysis bullosa and Kindler-EB. If you are affected by EBS, you will not go on to develop these other types. EBS is not caused by an infection, it is not contagious, and it is not due to an allergy.

WHAT CAUSES EPIDERMOLYSIS BULLOSA SIMPLEX?

The top layer of the skin (the epidermis) is made up of several layers of cells. Each layer has an internal support system formed from proteins (keratins), giving the skin strength and shape. In EBS, there is a reduction 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 reduction leads to skin weakness resulting in affected skin to be more easily damaged. When the cells break, they separate from each other, fluid builds between them, and a blister forms.

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

IS EPIDERMOLYSIS BULLOSA SIMPLEX HEREDITARY?

Yes, it can be inherited in two ways, as an autosomal dominant or as an autosomal recessive way. It is usually inherited from an autosomal dominant gene change. An autosomal dominant gene change means that one parent affected (has skin changes) by EBS can pass their altered gene on to their children who will then be affected by EBS. Either parent who has a dominant form of EBS 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 EBS with every pregnancy.

An autosomal recessive gene change means that both parents carry the gene change that causes EB simplex but have no symptoms of EBS. When both parents have the recessive gene change then each child will have a one in four chance of being affected by EBS. To be affected they would have to receive the affected gene change from each parent.

Sometimes a new change appears in the genes that cause EBS. This means that neither of the parents are affected or carry a gene change. This is called a new mutation.

Men and women are affected equally.

WHAT ARE THE SYMPTOMS OF EPIDERMOLYSIS BULLOSA SIMPLEX?

The main feature of EBS is blistering caused by friction (rubbing or scratching). 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 EBS. 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 in areas of friction. Friction areas may include the skin 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 of the feet 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 rubbing of socks. A few people find that hot food or hot drinks will cause blistering in 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 (infection with bacteria) can be a common problem.

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

In the severe type of EBS, 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 become much better after this. Severe EBS often improves 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 EBS 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 (usually less than 1 cm) and numerous but heal without leaving any scars. In severe EBS, blisters tend to occur in clusters, often healing to leave residual pigmentation (a dark area of skin). The skin on the palms and soles may be thicker, especially in the 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, a dermatologist may suggest taking a small sample of skin for more detailed examination under a microscope. A blood test may also be suggested to look for changes (mutations) 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?

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

In infants careful handling and application of non-sticky dressings 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.

Depending on the severity, treatment and advice may be required from a number of different specialists including nurses, dermatologists, paediatricians, physiotherapists, pain specialists, podiatrists and dietitians.

SELF-CARE (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. The 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.
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- If your condition is very disabling, you may be able to obtain further help with mobility by applying for Personal Independence Payment (PIP) or for a child under 16 Disability Living Allowance (see below for further information). Local authorities can sometimes provide or fund transport to school for children who are badly affected. 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 people with EB is available from DEBRA (UK).

DEBRA House
13 Wellington Business Park
Dukes Ride
Crowthorne
Berkshire, RG45 6LS
Tel: 01344 771961
Fax: 01344 762661
www.debra.org.uk

The NHS offers a commissioned EB service in the UK, please speak to your doctor so you get referred for assessment and management.

Personal Independence Payments (UK Government)

www.gov.uk/pip

www.gov.uk/dla-disability-living-allowance-benefit

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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