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What is EB?

What is Epidermolysis Bullosa?

Epidermolysis Bullosa, known as EB, is the name of a rare group of genetic skin disorders causing blistering and shearing of the skin from even the gentlest friction and sometimes spontaneously.

There are three major types of EB; Simplex, Dystrophic and Junctional. These vary from relatively mild to incapacitating, crippling, and sometimes fatal disorders. Within these there are over twenty different sub types of EB, each with their own characteristic symptoms.

All forms of EB are genetic in origin and the genes responsible for most of the sub-types of the condition are now known. Some still await identification.

The genetic defects result in the skin cells collapsing or skin layers not adhering properly to each other, causing areas of structural weakness. This fragile skin is particularly vulnerable to damage from friction, causing the continual blistering that is the characteristic feature of the condition.

What does the name mean?

The skin is a composite structure. The outer layer is called the 'epidermis'; the inner one the 'dermis'. 'Bullosa' is simply the name for a blister, whilst 'dystrophic' means mutilating. 'Lysis' means breakdown. Hence epidermolysis means breakdown of the epidermis. This is one group; however, there are other types which are less mutilating.

What causes the disorder?

The understanding of the causes of EB has grown greatly over recent years. All forms of EB are genetic in origin and the genes responsible for several different sub-types of the condition are now known. Others still await identification. The genetic defects result in the skin layers not adhering properly to each other, causing areas of structural weakness. This fragile skin is particularly vulnerable to damage from mild friction, causing the blisters which are the characteristic feature of the condition. Researchers are very optimistic about the possibility of developing treatments as knowledge of EB increases. D.E.B.R.A. is committed to ensuring that this optimism becomes a reality.

What are the symptoms of Epidermolysis Bullosa?

In the simple types of the disease, blisters are confined to the skin and very occasionally affect the mouth. These are often numerous but heal without permanently damaging the skin. Nevertheless, it causes discomfort and pain throughout the sufferer's life, and a great curtailment of normal social activities, including schooling.

In addition, the real or imagined reaction of the public to the appearance of the skin may lead to withdrawal from social activities, and to feelings of isolation, depression and defensive shyness.

The more serious types of the disease occur at a different, deeper layer of the skin levels; therefore, the damage is more serious. The blisters continue to enlarge until opened, destroying the outer skin as they do so. They heal with scarring, faintly resembling burns, and in time the appearance of all the skin becomes changed.

The scarring may cause severe deformity in parts of the body, most notably the hands. Here it eventually leads to fusion of the fingers and thumb, rendering the hand useless. Plastic surgery can be performed, but it is both difficult and painful. Despite advances in the making of suitable hand splints, the effect is not permanent and the child may require several operations on the hands and, to a lesser extent, on the toes.

The most extreme type of the disease can be fatal. Infants can be born with areas of the skin completely missing and the blisters do not heal.

Does the disorder affect any other parts of the body?

Yes, it commonly affects the mouth, as well as other linings of the body.

When the mouth is affected there is considerable pain and discomfort when eating, even in the milder form. This is similar to the pain and discomfort caused by mouth ulcers. In severe forms there are grave problems with eating, and the scarring left after the blisters have healed may cause the sufferer to have difficulty opening their mouth, making eating and speaking even more of an effort.

A similar effect can occur in the gullet, leaving a sufferer unable to eat properly for several days whilst the blisters heal. Many children need to have all their food liquidized. It is possible for the gullet to become so narrowed by frequent blistering that the food can no longer pass down it. An operation is then necessary to re-open it, and this may have to be repeated at intervals over the years. This is the most distressing aspect of the disease; problems of undernourishment may occur and further aggravate the condition.

Other linings of the body may be affected, and also conjunctiva of the eye, causing temporary blindness. The scars remaining after the healing of each blister may in time cause permanent loss of sight.

Does the disorder impair mental health?

Not at all. Despite the considerable disfigurement the disease can cause, the mental health of all the sufferers is normal. Indeed the above-average intelligence of many of the sufferers means that they find their physical limitations very hard to come to terms with. They are more than ever aware of the restrictions placed on them and many of their hopes and ambitions cannot be fulfilled.

Is it painful?

The degree of pain varies considerably according to the severity of the individual affliction. Most of us know how painful a single blister on the heel can be – can any one of us imagine what it must be like to have blisters every day of our lives caused simply by the normal friction of clothes? Obviously, this places major restrictions on daily life and activities.

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