**Epidermolysis Bullosa Simplex**

**What is epidermolysis bullosa simplex?**

Epidermolysis bullosa (EB) simplex is a rare inherited skin disorder which causes blistering.  In the most common form, blisters tend to be confined to the palms and soles, although occasionally they arise in the mouth, and are most troublesome during warm weather. The more severe form is called EB simplex Dowling-Meara and those affected have more widespread blistering which does not vary according to the time of year. EB simplex is different from the junctional and dystrophic forms of EB, and if you have EB simplex you will not go on to develop these other types of EB.

**What causes epidermolysis bullosa simplex?**

The top layer of skin (the epidermis) is composed of layers of cells. Each cell has an internal skeleton 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 another protein called plectin), which causes affected cells to be less resilient and to rupture when subjected to even minor physical stress. When the cells break, they separate from each other, fluid accumulates between them and a blister forms.

Weakness of the protein is caused by a minor abnormality (a mutation) in the gene responsible for producing that protein. A variety of such mutations have been identified in EB simplex, some of which are common amongst sufferers of the condition while others are only found in single families.

**Is epidermolysis bullosa simplex hereditary?**

Yes. It is inherited in a dominant fashion. This means that one parent of an affected person will also usually have the condition, although it is possible for EB simplex to appear for the first time in a person who has no other affected family member. Anyone who has EB simplex can pass the condition on to his or her children: there is a 50% chance that the child of an affected parent will inherit the blistering tendency. EB simplex affects men and women equally. It is not an infection, it is not contagious and it is not due to an allergy.

**What are the symptoms of epidermolysis bullosa simplex?**

The primary symptom of EB simplex is blistering, which is often painful and which can sometimes significantly affect daily activities of living and interfere with school and work.

Commonly, the blisters occur for the fist time during early childhood, affecting sites of friction such as under the elasticated areas of a nappy or, during the crawling stage, the hands and knees. Later, painful blisters tend to develop on the soles after walking only short distances. The formation of blisters is most troublesome during warm weather, and a few individuals may even be free of blisters during the winter. 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 drink will cause blistering within the mouth.

In the Dowling-Meara 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. In some, they may disappear for a short while during a feverish illness.

Thickened skin on the palms and soles, sometimes associated with an increased tendency to sweating at these sites, can be a feature of EB simplex.

Although abnormal blistering tends to be life long, it sometimes becomes a little less severe in adult life. Dowling-Meara EB simplex often improves dramatically after early childhood.

**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 large and numerous, but heal without leaving any scars. In the Dowling-Meara variant, blisters tend to occur in clusters on the body and limbs, often healing to leave residual pigmentation.

**How will epidermolysis bullosa simplex be diagnosed?**

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, your dermatologist may suggest taking a small sample of skin for more detailed microscopic examination. A blood test may be suggested to look for mutations of the genes likely to be involved.

**Can epidermolysis bullosa simplex be cured?**

In the past 20 years, there has been exciting and rapid progress in the understanding of EB simplex, but at the moment there is no cure. Several laboratories around the world are exploring strategies that they hope will lead to an effective treatment.

For information about available treatments please visit [this page](http://www.bad.org.uk/site/1153/default.aspx)on the website of the British Association of Dermatologists

