

EPIDERMOLYSIS BULLOSA SIMPLEX

1. Introduction

Epidermolysis bullosa simplex is an umbrella term for all forms of EB, where the blister formation develops within the outermost skin layer. The term "simplex" sometimes makes one tempted to believe that it is "simple", that it refers to uncomplicated and simple forms of EB. That's not quite right. It is true that some forms of EB compared with other forms of EB simplex seem less severe, it means a life can have limitations that can be different or perceived as stressful. There are also some forms of EB simplex, which are very rare, but have very serious effects on the lives and well being among the people with more severe forms of EB. It is therefore very important to know the exact diagnosis when EBS is suspected, if you want to adapt to the future course of the disease.



Important points in a nutshell

- **Epidermolysis bullosa simplex is the general term for all forms of EB, where blistering takes place within the outer skin/epidermis.**
- **EBS is caused by mutations in different genes.**
- **"Simplex" is not synonymous with "simple".**

2. EBS generalized severe

EBS generalized severe is caused by mutations in the genes of keratin 5 or 14. It is an autosomal dominant disorder, in most cases the disease is already known in the family. Instances of new cases by so-called spontaneous mutations occur, but for more detailed explanations, see the topic "Genetics".

EBS generalized severe is usually noticeable shortly after birth. The blisters appear all over the body, most commonly they are first seen on mechanically stressed parts of the body. Very typical for EBS generalized severe are rather small bubbles that appear in groups. Sometimes they form lines or are arranged in a circle. Typical is that the blisters are often found on the oral mucosa.

In hot summer weather almost all the affected people complain of increased formation of blisters. Warmth and sweating promote blister formation, hence it is at this time of year more often than usual that open sores and scabs form. Excessive sweating of the feet and mild cornification on the palm of the hand and soles of the feet may also occur and can be very uncomfortable.

Some of the affected people have observed changes in their skin color which are probably caused by pigmentation in areas where blisters occur. The pigmentation is harmless, yet the affected find it bothersome as the skin has a blotchy appearance.

Itching is a very common unpleasant side effect that is observed in EBS generalized severe and is a particularly unpleasant experience for the subject. Another peculiarity of EBS generalized severe is that despite the many blisters most sufferers prefer not to wear any bandages over the affected areas. They report, that under the dressings they perspire heavily and thus get more blisters, also at the edges of the bandages often creating new blisters. If the blisters are left uncovered they usually heal rapidly and easily. This is important to consider for wound care.

The prognosis for EBS generalized severe is generally quite good. For still unknown reasons, in some cases, the skin condition in EBS generalized severe improves during puberty. Unfortunately that is not always the case and one cannot predict how it will be in a particular case. Basically, as a general rule EBS generalized severe will accompany those affected for their entire lives.

The patients have a normal life expectancy, since in EBS generalized severe no additional problems usually occur that are caused by the disease.
