

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 easy 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 affected. 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 upper skin/epidermis.**
- **EBS is caused by mutations in different genes.**
- **"Simplex" is not synonymous with "simple".**

2. Very rare forms of EBS (EBS other types)

In addition to the better known forms of EBS, which each have a separate chapter, there are other types within this rare disease group that are even more rare. For example this means only one or two or maybe no one that is affected with this disease lives in your country. Therefore, little can be said or written about the general course of the disease. In such cases it is especially important to get in touch with a specialist center as they will try to learn all that is known about this disease through international contacts and literature.

Sometimes this is not enough. You can help the most by accompanying and arranging regular checkups and closely observing the affected child or adult. By early detection of problems, thinking ahead and by learning from other types of EB, we can also help these families.

The following types of EBS are in this group according to the current classifications:

- [Basal EBS-types:](#)
 - **EBS with muscular dystrophy (EBS-MD)**
 - **EBS with mottled pigmentation (EBS-MP)**
 - **EBS migratory circinate (EBS-migr)**
 - **EBS Ogna (EBS-Ogna)**
 - **EBS with pyloric atresia (EBS-PA)**

- [Suprabasale EBS-forms:](#)
 - **Lethal acantholytic EBS**
 - **EBS with plaktophilin-1- deficiency**
 - **Superficial EBS (EBSS)**

If you or your loved one has been diagnosed with one of these types of EB, then we strongly recommend that you contact an EB-center near to where you live, as we only discuss very general information. In most rare forms of EBS blistering starts at birth or in the first weeks of life. The blisters are caused due to mechanical action on heavily stressed areas of the body, but can also occur in less stressed areas.

In the form known as basal EBS there are some types with serious additional problems and ones that have a milder course.

Two examples of EBS with additional serious problems that we want to look at in more detail:

EBS with muscular dystrophy (EBS-MD):

In this type, a building block for the skin is affected that also plays a major role in the musculature. Over the years this can lead to an increasing muscle weakness that over time can force the problems with the blistering on the skin to become less of a focus. In this situation it is recommended to consult a neurological child-care center from the start. How pronounced this weakness will ultimately be, can hardly be predicted for each individual case.

EBS with pyloric atresia (EBS-PA):

In this case the transition from the stomach to the intestine (the "pyloric sphincter" or "pylorus") is closed. A problem like this is usually already detected before birth. The children need to be operated on soon after birth, therefore planning before the birth is especially important so that the surgical care of the child is ensured.

EBS suprabasale:

For these forms of EBS only case reports are available. At this point we therefore do not want to go into further details, yet not hide the fact that these cases are apparently very dramatic and from our perspective the term „simple“ is actually very misleading.

Skin care and medical procedures differ in all of these forms of EBS, not like the better known forms of EBS.

However, it must be ensured that these patients have necessary skin and wound care materials in sufficient quantity and if they should have any problems a medical contact is available.

With the very rare EBS forms little can be predicted about the general prognosis. In any case, these rare forms of EBS accompany those affected their whole lives.