

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

2. Localized EBS (formerly called Weber-Cockayne EBS)

Localized EBS 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".

The localized EBS is usually termed a mild subtype. This may be true in comparison with other forms of EB, but for those affected with this form of EB it also means a life with limitations. Compared with healthy skin, these limitations are very clear and burdensome.

Sometimes EBS is already noticeable shortly after birth, but it may also be that the first blisters only appear within the first months of life. In rare cases, the blistering begins later in childhood or adolescence.

The blisters are caused by mechanical action in heavily stressed areas of the body. The blisters therefore occur mainly on the palms of the hands and soles of the feet, occurring rarely on other parts of the body. Basically blisters can also occur all over the body with EBS, the blister formation is usually associated with stronger mechanical action such as falls and accidents. A belt that is too tight fitting can also be a possible cause.

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 the 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.

The prognosis in localized EBS forms is generally quite good. As soon as the patients and their families have learned to deal with this disease, they rarely need medical treatment. 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.

For still unknown reasons, in some cases the skin condition in localized EBS 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 common rule, localized EBS will accompany those affected for their entire lives.

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