

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

2. EBS generalized intermediate

This group includes all types where blisters appear all over the body **and** have genetic mutations in the genes of keratin 5 or keratin 14. The mode of inheritance can be dominant or recessive. Further explanation can be found under the topic "Genetics".

Generalized EBS forms start at birth or in the first weeks of life with the formation of blisters induced from mechanical stress and heat.

An increase in keratinization (formation of "horny" calloused like skin) on the soles of the feet and excessive sweating may occur, also changes in the nails and occasional involvement of the oral and nasal mucosa are often part of the clinical picture.

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 that more often than usual, 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.

The prognosis in EBS generalized intermediate 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 EBS generalized intermediate 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 intermediate will accompany those affected for their entire lives.

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