

DYSTROPHIC EPIDERMOLYSIS BULLOSA (DEB)

1. Introduction

Dystrophic Epidermolysis bullosa is an umbrella term for all forms of EB, where the blisters are formed in relation to the dermis layer of the skin. "Dystrophic" originates from the ancient Greek syllable "dys" and means "bad" and "tropein" for "feed" and "grow". Since the people suffering with this form are struggling with difficult food problems more than others, this gave the name to these forms. In all forms of DEB it is always the same block or always the same protein molecule of genetic changes involved, namely the collagen 7. These changes can then lead to the collagen 7 to be completely missing, or is greatly reduced or somewhat limited in its function. There are different degrees of severity of DEB depending on the changes in collagen 7. It is important to know the exact diagnosis when DEB is suspected, if you want to adjust to the further course of the disease. In DEB in addition to the blistering of the skin there are usually additional problems that we discuss in more detail with the different forms.



Important points in a nutshell

- **Dystrophic Epidermolysis bullosa is an umbrella term for all forms of EB, where the blister formation occurs in the bottom layer of the skin, the dermis.**
- **DEB is caused by different changes to a particular gene, the gene for collagen 7.**
- **The modes of transmission are different, there are recessive and dominant inherited dystrophic forms of EB.**
- **The individual's forms differ significantly, depending on whether collagen 7 is completely absent, reduced and/or limited in its function.**
- **In addition to the blistering on the skin, there are often additional problems (involvement of the mucous membranes and changes to the hands among other things).**

2. DOMINANT DYSTROPHIC EPIDERMOLYSIS BULLOSA (DDEB)

All forms of dominant dystrophic EB are caused by mutations in the gene for collagen 7. As the name implies, these forms are inherited as autosomal dominant. For more detailed explanations see the chapter „Genetics“.

Depending on how pronounced the changes are to the affected skin block the collagen 7 we distinguish the following forms:

- **DDEB generalized**
- **DDEB acral**
- **DDEB pretibial**
- **DDEB pruriginosa**
- **DDEB only nail changes**
- **DDEB-BDN (BDN = dermolysis bullous of the newborn)**

In these forms of EB blistering normally begins at birth. In the generalized form of DDEB the blisters can occur all over the body, but in the other forms they occur only in individual parts of the body. With healing of the blisters, sometimes scarring occurs and a slight atrophy of the skin can be seen in areas of intense blistering.

The first weeks of life may run a different course. In some children only a few blisters emerge in others the sometimes severe blistering can be very threatening. A baby with a generalized form of dominant dystrophic EB must therefore be well observed, in order to respond in time for infections or other problems.

Other problems may also occur in the course of time, for example- anemia, nutritional problems or growth retardation. Overall they are much milder than in the recessive forms of dystrophic EB.

During the further course changes to these forms of EB often result in and/or loss of the nails of the fingers and toes. Preventive measures have been tried, yet so far without success.

Some sufferers are observed to also have skin discoloration, probably due to pigmentation in areas where blisters have occurred. These pigmentary changes are harmless in themselves, but by those affected are often felt as disturbing as the

skin has blotchy areas. Regular inspections, especially of darker pigmentation, are necessary to make sure that no malignant changes (= skin cancer or a precursor of skin cancer) are hidden behind them.

Involvement of the oral mucosa may occur. The resulting complicated oral hygiene promotes the development of tooth caries. Therefore, the teeth and the dental hygiene must be given special attention in these forms.

In hot weather during the summer almost all affected complain of increased blistering. Heat and sweating also promote the formation of blisters, so it is more common than usual at this time to develop open sores and scabs.

The prognosis and life expectancy is quite good in the dominant dystrophic forms of EB despite all the problems, even if the disease persists for life. The course is variable and ranges from very mild gradients up to some severe cases, which are accompanied by additional problems. Individual cases cannot be accurately predicted, therefore good medical monitoring is in every case necessary during their whole life.

Now a few comments on the individual subtypes of DDEB with each characteristics:

DDEB generalized:

Generalized means that blisters can occur all over the body and are not limited to individual areas. Blisters occur more frequently in mechanically highly stressed parts of the body: on the knee or on the elbows and hands. Additional problems, such as those described above, are more likely than in the other forms, but may be absent.

DDEB acral:

The blisters are formed the most on the extremities. The outermost extremities of the body are referred to as "acral", for example the hands and feet, as well as nose, chin and ears. Other problems rarely occur in this form.

DDEB pretibial:

The focus of blistering is on the front of the lower leg, other parts of the body hardly incur blisters. Nail changes are frequently seen yet more problems are rarely observed.

DDEB pruriginosa:

The main feature in this form is the particularly strong itching, which has given this form the name "pruriginosa" meaning "itchy". The blistering is often not so pronounced, it may be generalized or limited to specific parts of the body. Other problems are rare but can occur.

DDEB only nail changes:

As the name suggests in this form, only nail changes occur and usually without other symptoms.

DDEB-BDN:

"BDN" means "dermolysis bullous of the newborn", which can be roughly translated as "bullous retinal detachment of the newborn." In this special form the newborn suffers from development of blisters over a large area after birth, the blisters will then heal relatively rapid. This may initially look quite dramatic, the babies should be observed and treated just like all other newborns with EB.

One must be aware that there are also other diseases of the newborn, which initially look similar to EB, but they require completely different treatment. There should be no confusion. After a few weeks in DDEB - BDN no more blisters are formed, or only very rarely since this is a temporary condition. This form of EB is very rare and it is necessary to wait until the diagnosis is in fact established.