

## JUNCTIONAL EPIDERMOLYSIS BULLOSA (JEB)

### 1. Introduction

Junctional Epidermolysis bullosa is an umbrella term for all forms of EB, in which the blisters are formed precisely in the connective segment between the outer most skin layer/epidermis and corium/dermis. The name "Junctional" comes from the Latin word "iunctio", which means something like "connection". This joining segment is also referred to as the basement membrane.

In JEB it is a rule that more problems can be observed than just the blistering of the skin. The individual forms of JEB differ significantly depending on which component is affected by the genetic modification. Therefore, in cases of suspected JEB it is particularly important to know the exact diagnosis, if you want be prepared for the future course of the disease.



### Important points in a nutshell

- **Junction Epidermolysis bullosa is an umbrella term for all forms of JEB, where the blister formation occurs within the joining segment of the epidermis and dermis.**
- **JEB is caused by mutations in different genes and the various forms differ significantly from one another.**
- **In addition to the blistering of the skin, there are usually additional problems.**

## 2. Other rare forms of JUNCTIONAL EPIDERMOLYSIS BULLOSA

In addition to the better known types 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 lives here in Austria that is affected with this disease. 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 centre 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 try to accompany and arrange regular check-ups to observe very closely the affected child or adult.

In these cases, it is especially important to consult with a specialist centre for timely detection of problems by thinking ahead and learning from the other forms of EB you can certainly help accompany these families.

The following forms of JEB included in the current classification:

- **JEB with pyloric atresia (JEB-PA)**
- **JEB inversa**
- **JEB "late onset"**
- **LOC syndrome**

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-centre 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, except for the JEB "late onset", which as the name describes, has a late start (= late onset) of blistering. The blisters are caused by mechanical action on heavily stressed areas of the body, but can also occur in less stressed areas.

In the rare JEB forms, there are some that have serious additional problems and those that run a milder course. Since the forms clearly differ, it is particularly

important to have a correct diagnosis. Only when the exact diagnosis has been made you can be prepared for the future course of the disease.

The main characteristics of these rare forms:

**JEB with pyloric atresia:**

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 ahead is especially important so that the surgical care of the child is ensured. Apart from the Pylorus atresia this form has a difficult course and can be compared with the process in JEB generalized severe. Exceptions have been described.

The name **JEB inversa** means that the disease – as the term indicates - is directed inward (inverse).

It is not so much the skin manifestations in the foreground, but the problems with the mucous membranes of the internal organs. Nutritional difficulties and problems with the urinary tract are more pronounced for those affected by JEB inversa while the blistering of the skin is rather mild.

With **JEB "late onset"**, as the name describes it has a delayed start (= late onset) of the blistering. It may be, that the disease occurs only during the course of the first few years of life and then worsens with advancing age.

**LOC-Syndrome:** LOC is the abbreviation for "laryngo-onycho-cutaneous", which can be roughly translated into: "concerning the larynx, the nails and the skin."

This form has been described only in families originating from Pakistan. It seems to be a specific genetic change that apparently has only occurred in Pakistan. The LOC syndrome is a very serious form of JEB, it has in addition to the skin manifestations involvement of the respiratory mucosa and also complications to the eyes.

One can say little about the prognosis in the very rare forms of JEB. In cases with severe side effects life expectancy is very limited, yet with mild forms it can be normal.

Skin care and medical procedures do not differ in all these rare forms of JEB, than that of the better known forms of JEB. It must be ensured that these patients have the necessary skin and wound care materials in sufficient quantities and a medical contact available.