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 connection zone between the outer most skin layer/epidermis and corium/dermis. "Junctional" comes from the Latin word "iunctio", which means something like "connection". This connection zone is also referred to as basement membrane.
In JEB it is as a rule that more problems can be observed besides the blistering on 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

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

2. JUNCTIONAL EPIDERMOLYSIS BULLOSA generalized intermediate

JEB generalized intermediate is caused by mutations in the genes for laminin or collagen 17. It is an inherited autosomal recessive disease, for more detailed explanations see the chapter ”Genetic“.

The blistering in JEB begins at birth or within the first few days. The blistering can be very mild during the first months of life and then it increases significantly. Due to the healing of the blisters, superficial scars and a slight atrophy (= atrophy) of the skin can remain.

The effect of the sometimes severe blistering can appear very threatening in the first weeks of life, a baby with JEB generalized intermediate must therefore be well observed so you can react quickly in cases of infection or other complications. Other problems may also occur later in the course of the disease, i.e. anemia, nutritional problems and growth retardation.

Changes and/or loss of the nails on the fingers and toes often occurs during the later course of this form of EB. Unfortunately with this form of EB the structure of the dental substance is disturbed, often causing pronounced tooth enamel defects. For this reason, the teeth should be given special attention.

A very common problem is severe, sometimes complete loss of hair. In the forms in which collagen 17 is involved the hair loss is frequently described as a problem. Especially (but not only) for female sufferers it is a great psychological burden. No means have been found to prevent or stop it from happening; however the use of a good wig can help.

Occasional involvement of the oral and nasal mucosa and rarely the eyes may occur. Also possible is a mucosal involvement of the urinary tract, such as the bladder and urethra. Fortunately this is rare, but for those who are affected it can be very uncomfortable.

At some of the affected skin discoloration is also observed, presumably due to pigmentation in the areas where blisters were. These pigmentary changes are harmless, yet can be seen as bothersome by the affected person as the skin has a

blotchy effect. Periodic inspections, especially of the darker pigmentation are necessary to be sure that there are no malignant changes.

In the hot weather during summer almost all affected complain about increased blistering. Heat and sweating promote blister formation so it is at this time more often than usual that open sores and scabs form. Excessive sweating of the feet and cornification on the palms of the hands and soles of the feet may also occur.

The prognosis and life expectancy in JEB generalized intermediate despite all the problems is generally quite good, even though the disease is for life. But the scenario is different for each case ranging from very mild to severe, which can often be overshadowed by many additional problems. In some individual cases it cannot be accurately predicted, good medical monitoring is therefore necessary.