JUNCTIONAL EPIDERMOLYSIS BULLOSA

1. Introduction

Junctional Epidermolysis bullosa (JEB) 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. "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

- **Junctional Epidermolysis bullosa is an umbrella term for all forms of EB, 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. Junctional Epidermolysis bullosa generalized severe

If you are confronted with the diagnosis of junctional Epidermolysis bullosa generalized severe type, then you should ask first whether the diagnosis is really 100% sure. The diagnosis is very serious, so with this form it is of great importance to be certain. When the diagnosis becomes assured, then you must be prepared for the hard time that lies ahead, especially if you are the parent of a child with this condition. It is difficult to find helpful words to make you more familiar with this situation, but it should at least be attempted. However we would like to bring to your attention that it is imperative that you discuss anything you read here once again with a specialist for EB and / or the doctor you trust.

The life expectancy of children with junctional Epidermolysis bullosa generalized severe type is unfortunately very limited. It ranges from a few weeks to a few years of life, with the affected child not usually reaching the age of 2 years. In some individual cases, the course cannot be accurately predicted. There are reports from other countries that a few children have lived longer. How long the life will be for any individual case may never be predicted, the only certainty is that it is very limited. The task at this time is for the medical professionals and parents to provide as much help for the child as possible. Despite feelings that naturally occur and can be overwhelming, you need to be aware that your child needs all the love and care you can give them. While your time together is limited, you will experience this period intensively. Try to live one day at a time and to accept every day as it comes. Enjoy the good days, and try to be there for your child as best as you can, also on difficult days

We would now like to take this opportunity to give you some medical information, so that you can adapt to the problems that might occur due to the disease. Given the severity of the disease the facts seem very dry, but to be well prepared you need in our opinion expert information. This is the basis to make correct and quick decisions in terms of the affected child and the family concerned.

JEB generalized severe is caused by mutations in the genes for laminin. It is an inherited autosomal recessive disease. For more detailed explanations see the chapter "Genetics".

The blistering in JEB generalized severe type begins at birth or within the first few days of life. The blistering can be very mild during the first months of life, then it increases significantly. While the blisters are healing, superficial scars and a slight atrophy of the skin can remain.

As with all other forms of EB by hot weather in the summer or even warm clothing, increased blistering can occur.

The mucous membrane of the mouth and nose, the entire intestinal tract and unfortunately the airway are also affected by the disease. This can lead to difficulties in the diet. Babies with JEB generalized severe can often be breastfeed at least for a while. The involvement of the lining of the airway is initially recognized by a hoarse-sounding crying, which can later evolve into a respiratory infection.

Blisters can also occur in the area around the eyes.

Special characteristics of JEB generalized severe are poorly healed wounds and excessive formation of scar tissue on the fingertips, in or around the mouth and also in the buttock region. The general condition can also be worsened by fluid and protein loss. Isolated infections of the skin and the respiratory tract can occur and often become more frequent. Over time, additional problems often develop, such as anaemia, nutritional problems and growth retardation.

A child with JEB generalized severe must always be closely observed in order to respond quickly and appropriately with infections or when other complications occur. You have to realize that in spite of the many ways of treating this condition, JEB generalized severe has no cure at present.

Over time the complications can be severe and it is not always possible to truly eliminate them. The inclusion of a palliative care team is therefore recommended from the beginning. These teams will act according to the principle: "Even though you really cannot do anything, there is much you can do."