KINDLER-SYNDROME

1. Introduction

Due to the basis of certain characteristics the Kindler syndrome (KS) does not fit into one of the three main types of Epidermolysis bullosa, so it is made into a separate disease in addition to the other forms of EB, the fourth group it is solely for KS. The condition got this name from Theresa Kindler, who first described the disease. It differs from the other forms of EB in that the blisters must not be formed within a certain layer of the skin, but in different layers. The blisters are formed also by mechanical action. Moreover there are other features, such as pigmentation and photosensitivity.

Important points in a nutshell

- **Blisters can develop in different skin layers.**
- **Its key characteristics are blistering of the hands and feet, pigment changes to the skin and marked photosensitivity.**
- **The mucous membranes are often involved.**

2. KINDLER-SYNDROME

The cause of the Kindler syndrome was found in changes in the Kindlin gene. It is an inherited autosomal recessive gene, for more detailed explanations see the chapter „Genetics".

The blistering starts right after birth, especially involving the extremities. Acral is what the extreme ends of the body are called, for example the hands and feet, as well as nose, chin and ears. At first the formation of blisters is in the foreground, it then decreases over the years.

Gradually a distinct poikiloderma develops, which roughly translates to "coloured skin." It is caused by pigment changes and it makes the skin blotchy. With age dry skin with mild scaly patches and hair loss may occur. The skin on the hands and feet typically tend to atrophy and the skin appears thinner.

In addition, the patient suffers from sensitivity to light, which can vary between very strong to very mild.

Frequently there is also an involvement of the mucous membranes. This means for those concerned that in the area of the mouth, esophagus and also the area in and around the anus problems may occur. In the mouth severe gingival inflammation can occur. Constrictions (stenosis =) may form in the esophagus and in the intestinal tract. This can result in difficulty swallowing and difficulty in bowel movements. In some of the affected inflammation of the intestinal mucosa which has been described, they may require treatment in first few months of life.

In Kindler syndrome adhesions of the soft skin areas, such as in the fingers and sometimes in the areas of external sexual characteristics can occur. Whether these adhesions occur and how severe they are, is different in every case and cannot be predicted.

The prognosis is good in spite of all these problems; the life expectancy is generally not affected. In adulthood regular check-ups with skin checks should be carried out in addition to the necessary treatments, as there is an increased risk for developing skin cancer. Early identification plays a special role; precursors of skin cancer can be detected and can be quickly removed. Special attention should be given to the hands and face area.