EPIDERMOLYSIS BULLOSA

1. Introduction

The human skin consists of several layers. The epidermis is the outer most layer which is the “outer skin”. "Bullosa" means "bubble-shaped" and "Lysis" solution. Therefore Epidermolysis bullosa (EB) is literally called "bubble-like detachment of the epidermis”.

Epidermolysis Bullosa is not the name for a single skin disease, but a large group of clinically and genetically different diseases. Their common feature is the formation of blisters. Blisters form from low mechanical stress on the skin and/or mucous membranes. There are a number of sub-types of Epidermolysis bullosa, that differ due to the skin layer in which the blisters are formed and what exactly the genetic cause is. These types come with some additional features and problems.

Important points in a nutshell

- **In all types of EB the skin forms blisters after a low mechanical stress.**
- **Due to genetic changes in each individual type of EB a building block for the anchoring of the skin layers with each other is missing or is not fully established.**
- **The various forms of EB can be distinguished by:**
  > the skin layer in which the blisters are formed
  > the missing protein molecule
  > the exact location of the genetic modification
  > the severity
  > additional problems also caused by the lack of the molecules.
- **Even with similar-sounding name the forms of EB can be completely different!**

2. General Information

Human skin is a complex functional system, which is composed of many layers, components and interim substances. So the skin can fulfill all its functions there must be the correct amount of building blocks present and they all need to be in an accurately defined amount and well coordinated with each other. When a building block is missing or there is not enough of them, or has a different structure as it should have, then the functioning of the whole system is compromised. Depending on whether it is a particularly important or not so significant building substance, either more or less serious problems can occur.

EB is due to genetic changes (or named "mutations") of certain protein molecules, which are responsible for the necessary cohesion of the individual layers of the skin are no longer or not fully developed by the body cells. This results in individual layers of the skin that can no longer be held together well. With mechanical action, such as by rubbing, these skin layers then dissolve and the anchorage is loosened somewhat from each other. This enables the interstitial fluid to penetrate and spread out to intermediate spaces and continues to expand, thus more interstitial fluid still penetrates - and on the skin, a liquid-filled blister becomes visible. This results in blisters and sores all over the body, in some cases, not only "outside" on the skin, but also on the mucous membranes in the mouth and the digestive tract or in the eyes.

What's more, some these components are not only in the skin, but also have functions in other areas of the body. If the plan for this component is now changed, other functions can also be impaired, so that in some forms of EB there may be other concerns in addition to the typical skin manifestations, for which we will discuss in details under the relevant form.

The different forms and degrees of severity of EB are thus determined by the respective existing genetic variation. The precise function and nature of the lack of or inadequate function determines how serious the consequences will be.

The skin essentially consists of three different layers:

- Outer skin or epidermis
- Dermis or corium
- Subcutaneous or hypodermis

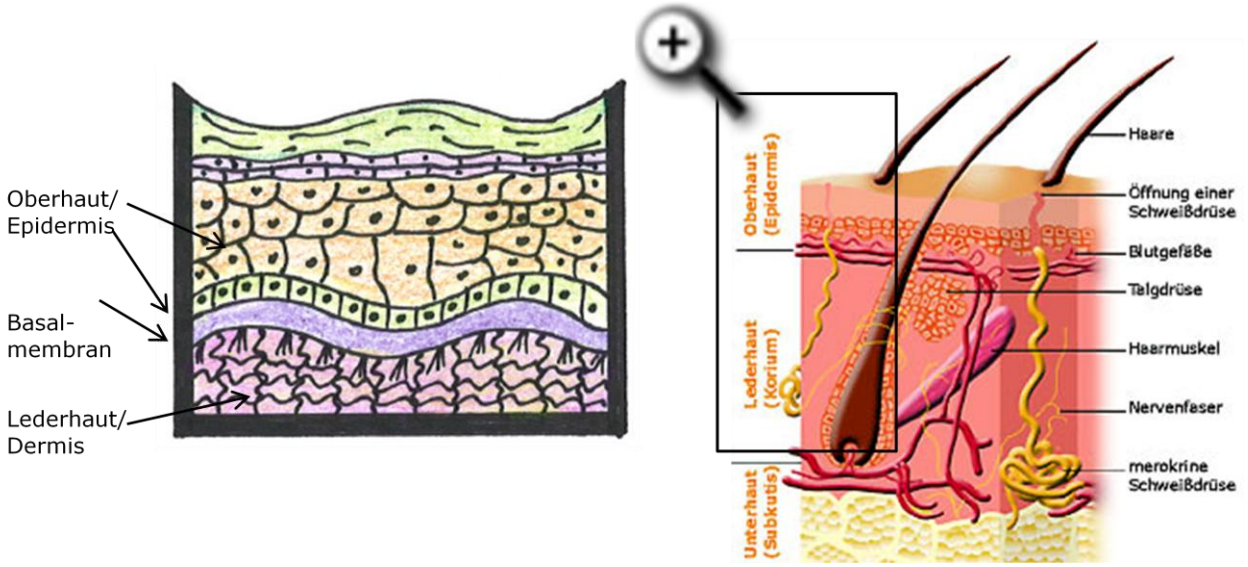

Illustration 1: A. Waldhoer, EB House Austria Illustration 2: http://www.qualimedic.de/haut.html

You can distinguish the many different types of EB, depending on whether the blister is formed in a more superficial layer of the skin or in a deeper layer, if the blisters are all over the body, or only in high stress areas of the body (such as hands and feet) or if the missing component has an important function other than for the skin. For many years we have divided EB into three broad groups depending on the layer in which the blisters are formed:

The three groups are:

I) EB simplex (blister formation within the outer skin/epidermis)
II) Junctional EB (cracking between outer skin - dermis and epidermis/ dermis/corium)
III) Dystrophicans/dystrophic EB (cleft formation within the dermis corium)

Each of these three large groups is then divided into sub-types, a total of more than 25 types are described. There is still a disease that belongs to none of the three groups, but is now recognized as a form of EB.

Therefore there is now a fourth main group:

IV) The Kindler Syndrome.

In 2014 EB experts joined together from all over the world and tried to newly arrange and to partly rename all known EB-forms, an overview of this new reorganization can be found in “Inherited epidermolysis bullosa: Updated recommendations on diagnosis and classification” - an update.
It is likely that by the improved possibilities of genetic analysis and diagnostics that in the next few years yet unknown forms of EB will be added.

If you do not yet know with which type you are dealing with, you should please ask which type is most likely in question before you read our information on a specific type. Do not be tempted to think that similar sounding diagnoses are also similar in their course! Above all, the common abbreviations often lead to confusion and mistakes, a "junctional EB generalized severe" for example, has a completely different course as a "junctional EB generalized intermediate".

If you or your loved one should suffer from a very rare, new or not yet clearly defined form of EB, then please contact an EB Consulting Center for more detailed information. In spite of progress in the diagnosis and despite all efforts, there is still a not negligible percentage of those affected, which apparently is a form of EB, but so far no genetic findings to support this. This uncertainty is very uncomfortable especially for the patients and their doctors, but unfortunately a fact which we must live with. We hope that in the future, with further research and development more and more uncertain cases can be solved!