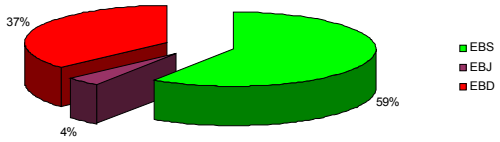
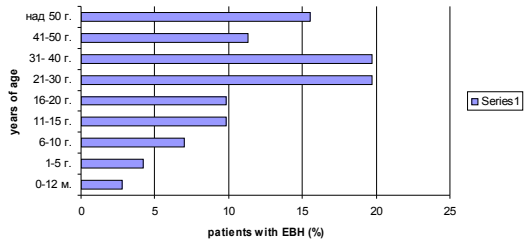
## ASSOCIATION EPIDERMOLYSIS BULLOSA - BULGARIA (A. E. B. B.)

Till now **the epidemiological study on EBH in Bulgaria for 37 years period** (**1970-2007)** over the distribution of EB in the country have discovered nearly **110 patients**.

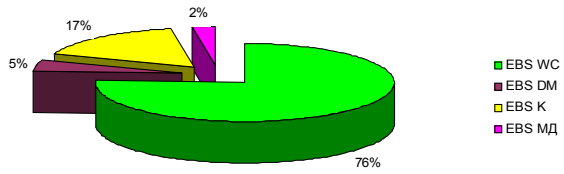



The upper pie-chart shows **the distribution of EB patients by major types.**
The highest percentage is that of EBS, followed by EBD.
The disorder of all patients begins at birth or up to three months later.
**The patients aged** from 20 to 29 are the majority.




**The prevalence** of all types of EBH in Bulgaria (general population 7,973,671) up to 31 December 2007 is 8.6 cases/per million inhabitants; of EBS – 5,3/mill; of EBJ –0,1/mill; EBD – 3,1/mill.
**The analysis of the mode of inheritance** of EBH cases showed 35 patients (59%) with AD mode of inheritance and recurrence risk of 50 %, 9 patients (15%) with AR mode of inheritance and recurrence risk 25%, and 15 patients (26%) were classified as sporadic cases without clear pedigree criteria for a single gene (Mendelian) mode of inheritance.
The application of the genealogical method was mandatory in the assessment of the recurrence risk of the appropriate family members.
The diagnosis in our patients has been confirmed histologically and 40 0f them have been investigated by electron microscopy.

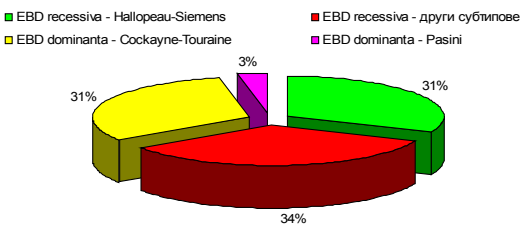
**We diagnosed 48 patients as suffering from EBS.**




The distribution of these patients by subtypes indicates that EBS Weber-Cockaine (EBS WC) has the highest percentage, followed by EBS Koebner (EBS K).
The mildest form of EBS is Weber-Cockaine, which is presented by blisters and vesiculation on the palms and soles predominantly. We observed AD mode of inheritance in all affected families.
Generalized skin affection was present in the EBS Koebner and EBS Dowling-Meara (EBS DM) patients. We diagnosed 1 patient with EBS with Progressive muscular dystrophy (EBS MD).
The ultra-structural examination in EBS demonstrated cytolysis of the basal keratinocytes. The erosions healed without scars in all EBS patients.

**We diagnosed 5 patients affected by EBJ/Herlitz – 4, non Herlitz -1.**
All the 4 patients with EBJ Herlitz (letalis) died before they were one years of age. The EB Junctionalis non Herlitz patient had a mild course of the disease.

**The EBD group consisted of 32 patients.**
Almost 1/3 of them have dominant dystrophic EB subtype Cockaine-Touraine and another 1/3 have recessive DEB subtype Hallopeau-Siemens.



**The clinical features of EBD subtype Cockaine-Touraine** are presented by generalized skin affection with haemorrhagic bullas, atrophic scars and nail dystrophy.
**The patients with recessive subtype EBD Hallopeau-Siemens** are the most seriously affected. With the progress of age EBD Hallopeau-Siemens patients develop severe muscle-skeletal deformations. But the most severe complication of this subtype DEB is squamoscell carcinoma, which is the main cause of death in early age: 35 – 40.
**The ultrastructural examination in all DEB patients** showed cleavage under LD.

74 % of the patients with EBD demonstrate severe mucous membrane affection. 70 % of the them had severe muscle-skeletal deformations. There is a 29 years old young man with EB dystrophica, total atresia of the oesophagus and a nutritional gastrostoma.
The most severe forms of the disease lead to early death, because of unfavourable conditions of bringing up, lack of medicaments and insufficient training of medical staff.

**Parents take care** of their affected by EB children. 30 of the sufferers are heavy invalids while the others, earn their own means of living. All of them wish to correspond with friends from other countries. All patients and especially these with EB dystrophica need psychological and financial assistance. The arrangement of orthopaedic, dentologic, dermatologic and psychiatric services for them is essential. We are working for more precise diagnosis of the disease and for better possibilities of treatment and rehabilitation.

**Bulgarian research and education center of EB is the Department of Dermatology and Venerology - Medical University, Pleven.**
**The Association Epidermolysis Bullosa Bulgaria** was registred on 19th of February 1995 in Pleven's Region Court. It is a voluntary, charity organization with ideal object. It unites the patients, their relatives and friends as well as people from all social spheres, with the only purpose to make patients life better. **The A.E.B.B.** is a member of a network of organizations for EB in the world – DEBRA INTERNATIONAL since 1992.

**The main tasks of the Association Epidermolysis Bullosa in Bulgaria are:**

1. To create a National Register and information Bank for the patients with EB in Bulgaria;
2. To support daily necessities of the patients with EB (medicines, dressings and food);
3. To prepare information materials for physicians, nurses, patients with EB and their relatives;
4. To establish personal contacts between the patients and consolidate the international links with the organizations of EBH individuals in Europe;
5. To create favorable conditions for raising, education, professional orientation of the patients with EB.

**A.B. E. B. has to organize information activity:**

- to favour informal communications among patients, and medical specialists
- to publish bulletins about association's activity with participation of the members
- to prepare information concerning different problems related with the disease.
- to inform public about the problems of the EB patients
- to stimulate and organize charity to collect funds
- to help the patients with EB to feel adequate members of society, to facilitate their life, to improve their physical and emotional status by joining them in to different and proper activities.

A.E.B.B. have a website: **http://www.debrabg.com** in Bulgarian language.
We have organized four workshops in 2000, 2004, 2005, 2006.
In 2005 we established contact with **INFORMATION CENTRE RARE DISEASES AND ORPHAN DRUGS** in Plovdiv, Bulgaria.
In 2007 we become a member of **ALIANCE OF THE PEOPLE WITH RARE DISEASE** in Bulgaria.
**At present /2008/ we collaborate with Prof. Lenna Bruckner-Tuderman and D-r Cristina Has from the EB Center in Freiburg, Germany**, where we do the immunofluorescence analysis of the skin samples and DNA mutagen analysis of the affected EB families.