News From The First Regional Symposium On Hereditary Epidermolysis Bullosa (Mediterranean – Central and Eastern Europe)

University Department of Dermatology and Venereology, Zagreb University Hospital Center and School of Medicine, Zagreb, Croatia, with Referral Center of the Ministry of Health and Social Welfare of the Republic of Croatia for Hereditary Bullous Epidermolysis and Debra Croatia, organized The First Regional Symposium on Hereditary Epidermolysis Bullosa (Mediterranean – Central and Eastern Europe) under the auspices of the Ministry of Health and Social Welfare of the Republic of Croatia, Academy of Medical Sciences of Croatia and Croatian Dermatovenereological Society of the Croatian Medical Association. The Symposium took place in Zagreb, April 3-4, 2009.

Epidermolysis bullosa (EB) is a group of rare, inherited skin diseases characterized by recurring painful blisters and open sores, often in response to minor trauma, as the result of the unusually fragile nature of the skin. Some severe forms may involve the eyes, tongue and esophagus, and some may produce scarring and disabling musculoskeletal deformities.

Hereditary epidermolysis bullosa (HEB) is one of 400 monogenetic genodermatoses that have a complex multigenic background and are influenced by environmental factors such as UV light, which may lead to skin tumor growth. The prevalence of HEB in Europe is >30,000. Confirmation of HEB diagnosis depends on the clinical picture, histopathology, immunohistochemistry, electron microscopy, cell culture, mutation (DNA) analysis, and antigen mapping. Socioeconomic aspects of EB refer to local therapy (external preparation and dressing material) and need for frequent consultations and treatments.

This Symposium dealt with all topical aspects of the disease. The Symposium was attended by 80 participants from 14 countries (Austria, Belgium, Bulgaria, Bosnia and Herzegovina, Croatia, France, Germany, Italy, Macedonia, Mexico, Montenegro, Romania, Slovenia, and United Kingdom). There were seven invited speakers from Austria, France, Germany, Great Britain and Mexico. During the Symposium, emphasis was put on the-state-of-the-art on EB, along with historical aspects that were presented by Head Doctor Štefanija Puretić, a doyen on EB from Zagreb, Croatia. Mutations in the genes for structural proteins in keratinocytes and dermoepidermal junction were presented by C. Has from Germany, and immunofluorescence in the diagnosis of EB by G. Pohla-Gubo, Austria.

EB has been classified into the following types: EB simplex (keratin genes); junctional EB (non-Herlitz and Herlitz type – laminin 322), and dystrophic EB (type VII collagen). The new classification of EB was presented by J. W. Bauer from Austria.

Diagnosis, prevention and therapy of EB depend on mutation in the genesis, therapeutic response, coding for components of the skin, mucous membranes, hair and nails. EB has been associated with a number of complications, e.g., growth retardation, pseudosyndactyly, contractures, squamous cell carcinomas, skin infections, impaired wound healing, enamel defects, caries, microstomia, gastrointestinal tract stenoses, and EB nevi.

J. W. Bauer from Austria presented successful novel management of EB, which relies on the center of competence, support group, university department, family physician, family and patient education for wound skin care, for example, wearing cotton gloves, non-adhesive tape (binding, padded layers of dressing) and placing ointment in the

Acta Dermatovenerol Croat
2009;17(2):147-148

eyes; cotton wool underneath blood pressure cuff, change of dressing and splints after hand surgery; installation of eating device, learning stretching exercises of the fingers, range of motion, etc.

Our Symposium emphasized historical data on 30-year experience in the diagnosis and management of EB at University Department of Dermatology and Venereology, Zagreb University Hospital Center from Zagreb, Croatia. The experience of DEBRA Croatia in the treatment and educational programs was presented in an instructive way; the experience in plastic and reconstructive surgery in dystrophic HEB patients was presented by A. Kljenak from Zagreb Children's Hospital in Zagreb; and on esophageal dilatation by S. Višnjić from Croatia. The management of genitourinary problems and correction of anemia and bone mineralization was successfully presented by J. Mellerio from the United Kingdom. A. Hovnanian from France talked about new gene therapy and novel options for EB therapy. An overview of medical problems and complications in EB was given by S. Murat-Sušić and coworkers from Referral Center. Splint therapy and procedures, and anesthesiology aspects of EB were also discussed. Three cases of squamous cell carcinoma were reported by K. Husar from University Department of Dermatology and Venereology, Zagreb University Hospital Center. At present, prevention, i.e. good skin care and wound care when blisters arise, remain the cornerstone in the management of squamous cell carcinoma and melanoma. Despite all efforts, degeneration to malignancy is inevitable. For now, close surveillance and early treatment offer best hope.

It was a successful Symposium. The next, 2nd Regional Symposium on EB in Croatia, to be held in two years, will hopefully offer reports on successful results in the treatment of HEB patients.

Professor Jasna Lipozenčić, MD, PhD

Your childern should be in the air. Protect their skin with Nivea cream; year 1929.
(from the collection of Mr. Zlatko Puntijar)