Epidermolysis bullosa congenita

Epidermolysis bullosa congenita (EBC), also known as Butterfly Wing Disease, is a rare congenital disease in which blister on the skin form spontaneously or by minor pressure or friction. Abou 30 000 people in Europe and about half a million worldwide suffer from this disease, with more than 360 pacients in Czech Republic. EB is caused by mutations in eleven genes and manifests itself mainly on the skin of patients. EB is basically divided into three forms - simplex, junctional and dystrophic - and more than 30 subtypes[1].

Shown here is five-year-old Iraqi boy Abdulrahman, who was diagnosed with congenital epidermolysis bullosa, a rare skin condition that produces painful sores and blisters and affects one in 5 million people. Corpsmen and doctors from MNF-W and the Iraqi army worked with medical specialists in the United States to diagnose the boy's condition and bring special medical products to Iraq to help ease the boy's suffering.

Types

- **simplex** – degeneration of the basal layer of the epidermis (clinical manifestation of a blister);
- **junctional** – blisters appear on histologically normal skin in the lamina lucida layer;
- **dystrophic** – blisters occur in the lamina dense [2].

Heredity

Autosomal dominant or autosomal recissive.

Patogenesis

The altered dermoepidermal junction results in loosening of this junction, resulting in blister formation.

1. Simplex form - mutations in genes encoding keratins 14 and 5.
2. Junctional form - mutation of at least 6 genes encoding laminin 5.
3. Dystrophic form - mutation of genes encoding collagen 7[2].

Clinical description

- Varied range of manifestations;
- skin, oral mucosa, GIT, respiratory, urogenital tract involvement;
- Subsequent scarring and contractures;
- patients avoid certain foods, leading to secondary malnutrition, anaemia, growth retardation and delayed wound healing[1].

Therapy

Care is aimed at prevention in order to prevent severe disability of patients. Interdisciplinary collaboration between dermatologist, histopathologist, paediatrician, haematologist, physiotherapist, geneticist, dentist, plastic surgeon, dietician, psychologist, orthopaedic surgeon, ophthalmologist is essential[1]

Specifics of dental treatment of patients with EBC

- Higher caries (reduced hygiene of the TMJ, inappropriate choice of diet composition),
- prohibition of the use of plasters,
- prohibition of wearing dentures,
- lubrication of instruments,
- prohibition of asymmetric tooth extraction (asymmetric pressure on the mucosa).

And all this, together with the frequent presence of microstomata, leads to a very difficult outpatient treatment.

Case report

Patient M.S. born in 2005 came to the clinic in June 2008. Because of her underlying diagnosis, her parents were recommended by the general dentist to be treated at a specialized clinic. On examination in the outpatient clinic, carious destruction of teeth 53, 54, 62, 64, 63, 74, 84 was found. No treatment was performed and due to the underlying diagnosis, she was indicated for restoration under general anaesthesia. In July 2008, 16 temporary teeth were extracted. After the procedure, there were numerous mucosal erosions and skin defects in the oral cavity. In June 2010, the patient was again referred by her general dentist for restoration in CA. On examination, the teeth 55, 65, 75, 85 were found to be destroyed. She was again indicated for restoration under general anesthesia. The carious teeth were extracted.

The patient is now dispensed at the clinic.

Links

External links

- FINE, Jo-David. *Inherited epidermolysis bullosa* [online]. Orphanet Journal of Rare Diseases, ©2010. [cit. 2022-17-12]. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2892432/pdf/1750-1172-5-12.pdf>.

References

1. BUČKOVÁ, Hana – BUČEK, Jan. Epidermolysis bullosa congenita – what's new. *Postgradual medicine.* 2004, vol. 3, p. 294, ISSN 1214-7664.
2. KUMAR, Vinay – ABBAS, Abul – FAUSTO, Nelson. *Robbins and Cotran pathologic basis of disease.* 7. edition. Elsevier Saunders, 2005. 1263 pp. ISBN 0-8089-2302-1.