Epidermolysis bullosa – a clinical study

Overview. The study included case histories of 41 patients with epidermolysis bullosa, who were treated in Hospital of Dermatology and Communicable Diseases during 2000-2014.

Results. Patients' age has oscillated from 2 till 52 years old (M/F-28/13; Rural/Urban – 25/16). Nosological spectrum ranged as follows: simple epidermolysis bullosa (EBS) Koebner–11 cases; EBS Weber-Cockayne – 9 cases; EBS associated with pigmented lesions – 1; jonctional epidermolysis bullosa – (EBJ) Herlitz – 1; dystrophic dominant form of EB (EBDD) Cockayne-Touraine – 12; EBDD Passini – 3; recessive form of EBD (EBDR) Hallopeau – Siemens – 4. Thus, EBS has been established in 51.2% of cases, EBJ – 2.4% of cases and EBD – in 46.34% of cases. Nail involvement was observed in 48.8 cases of EB. Hyperhidrosis, palmoplantar keratoderma, alopecia and ichthyosiform lesions were marked in 12.2% of cases. About half of patients has manifested a plenty number of associated diseases (anemia, pneumonia, hepatitis, pancreatitis, pyelonephritis). Dental anomalies and mucous membranes involvement were reported in 1/3 of patients, one in every 10 patients presented a mental retardation. In 20% of cases pyococcal complications (pyodermas) was described, in addition patients with EBDR Hallopeau-Siemens have developed keloid scars, acrosclerodactylitis, muscular contractions, mutilations, esophageal stenosis. A 2 month old infant with EBJ-H has died. More benign evolution was marked in patients with dominant forms of EBD (63.4%). All patients have presented a new bulla during obvious therapy. Epithelization of erosions occurred within 5-14 days. Topical treatment with "Mepilex Lite", "Mepitel" reduced twice the time of lesion epithelization (3-7days).

Conclusions. Epidermolysis bullosa profile certifies prevalence of this disease among males, as well as, increased number of dominant forms, association with nail and mucous membrane involvement, dental anomalies, anemia, microbial infections, in severe cases – acroscelro-dactylitis, mutilations, esophageal stenosis. "Mepilex Lite" and "Mepitel" dressings have reduced the time of lesion epithelization.

Key words: epidermolysis bullosa, clinical features, treatment difficulties.

Pemfigus benign familial Hailey-Hailey – prezentare de caz

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Generalități. Boala Hailey-Hailey (pemfigus benign familial) este o genodermatoză (mutații în gena ATP2C1) rară, cu transmitere autosomal-dominantă, caracterizată printr-o fragilitate crescută a pielii și structură anormală a desmozomilor. La instalarea diagnosticului, pe lângă tabloul clinic specific și antecedentele familiale, o importanță semnificativă o are examenul histopatologic: bula situată suprabazal, papilomatoza – papile dermice cu aspect „în degete de mănușă", care proemină în interiorul bulei, pe alocuri cu asociere de hiperkeratoză.

Prezentare de caz. Prezentăm cazul unei paciente de 43 de ani, din mediul rural, observată în secția femei a Spitalului Dermatologie și Maladii Comunicabile, cu diagnosticul *Pemfigus* benign familial Hailey-Hailey, durata maladiei fiind de aproximativ 7 ani. Pacienta s-a tratat ambulator, de mai multe ori, pentru intertrigo și lichen plan, cu efect curativ neînsemnat. Cazuri asemănătoare s-au constatat la bunică, tată, soră și frate. Pentru prima dată, erupția s-a observat pe gât. Datele obiective relevă prezența plăcilor eritemato-veziculo-buloase, zemuinde, circinate, cu tendință spre confluare, macule hipercromice, localizate în pliurile axilare, submamare și pe gât, asociate de prurit. Semnul Nikolsky a fost negativ. Diagnosticul s-a stabilit în baza următoarelor date: tablou clinic, antecedente familiale, examen histopatologic. S-a observat o evoluție favorabilă în tratamentul indicat, care a inclus: corticoizi pe cale generală (doze moderate de prednison cu reducere treptată), dermatocorticoizi, loțiuni antiseptice.

Concluzii. Cazul este adus în discuție pentru raritatea acestuia și particularitățile clinico-evolutive specifice. Maladia a evoluat timp de mai mulți ani cu aspect de intertrigo și lichen plan, ceea ce a creat dificultăți de diagnostic pozitiv și diferențial.

Cuvinte-cheie: *pemfigus* Hailey-Hailey, genodermatoză rară, prezentare de caz.

Familial benign pemphigus Hailey-Hailey: a case presentation

Overview. Hailey-Hailey disease (familial benign pemphigus) is rare autosomal-dominant genodermatoses (induced by a mutation in ATP2C1 gene), which is chracterized by an increased skin fragility and abnormal structure of the desmosomes.

A case report. A 43 year old female patient, originated from a rural region,was treated in Hospital of Dermatology and Communicable Diseases for familial benign pemphigus,the disease lasted for approximately 7 years. Initially,the patient was treated for intertrigo and lichen planus, but without any significant effect. Similar clinical manifestations have been seen in the patient's relatives: grandmother, father, sister and brother. For the first time, skin lesions occurred on the patients' neck. Clinical inspection revelead presence of erythematous macules covered with vesicle, bulla and erosions with oozing, which had a tendency to confluent together, itching hyperpigemented macules were seen in axila, submamary folds, as well as on neck. Nicolsky sign was negative. Diagnosis was made on the basis of clinical signs, heredetary anamnesis and histopathological report. Treament, which included systemic steroids, moderate dosage of prednisolone, topical steroids and antiseptic lotions, has shown a positive result.

Conclusion. This case was presented for discussion due to its rarity.

Key words: Hailey-Hailey disease, rare genodermatosis, case report.