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A RARE CASE OF THE CUTANEOUS FORM OF MASTOCYTOSIS IN A SAKHA CHILD

The article is devoted to a rare disease which is mastocytosis in a child. In Russia, this disease is registered with a frequency of 0.12–1 case per 1000. At the moment, the etiology and the pathogenesis of the disease are not fully understood. Mastocytosis is a rare disease with a favorable prognosis in children. Due to the small occurrence, diagnosis and treatment are often difficult. All children diagnosed with mastocytosis should be registered at a dispensary due to the possibility of transition to a systemic option at an older age. The article presents a clinical case of mastocytosis in a Sakha child.

Keywords: mastocytosis, rare disease, dispensary registration, allergy, urticaria, consultation, observation.

Introduction. Mastocytosis is a heterogeneous group of rare diseases of myeloproliferative nature in which there is excessive accumulation and proliferation of mast cells in tissues and organs. In Russia, the incidence is 0.12-1 case per 1,000 patients [5,6]. Despite the heterogeneity of the clinical picture, the leading link in the pathogenesis of mastocytosis is occupied by molecular and genetic mechanisms [4]. According to the opinions of a number of authors, mastocytosis in children is a temporary manifestation of mast cell hyperresponsiveness, which debuts at an early age, proceeds as skin forms and spontaneously regresses when the child reaches pubertal age [1,2,3,5]. Despite the available specific criteria and recommendations, the diagnosis of mastocytosis in Russia is fraught with certain difficulties. Considering the peculiarities of the course of the disease, namely its tendency to spontaneous self-resolution in children, patients with mastocytosis require consultative care first, and then medication [5,6]. Timely consultation of the patient about the disease avoids delaying treatment, as well as worsening the course of the disease, which has a positive impact on the quality of life of patients.

The clinical example: The boy named I., of Sakha origin, was born as the first child in a family in one of the Arctic regions of Yakutia. The weight at birth was 3200, the height was 50 cm. The pregnancy proceeded smoothly. The childbirth was on time and independent. The Apgar score was 8/9. Neonatal jaundice up to 1 month was registered during his neonatal period. He grew and developed during the first year according to his age. He was breastfed until the age of 1 year. Hereditary history was not aggravated. Mother had atopic dermatitis.

At the age of 9 months, the child had a rash on the face, back and chest: elements of 0.5-0.7 cm in diameter, irregularly shaped, scarlet-red color, prone to merging, rising above the skin surface. The rashes periodically reddened and swelled when rubbing, bathing in warm water. The pediatrician considered these rashes to be hemangiomas.

In August 2022, on the recommendation of the district pediatrician, the child was referred for examination to the Pediatric Center of the Republican Hospital No. 1-NCM.

The child was examined by an allergologist-immunologist. His condition was estimated as satisfactory. His well-being was not impaired. On examination of the face, chest and back, there were multiple brownish-colored itchy spots or papules of oval or circular shape, merging with each other in some places. Regional lymph nodes were not palpated. There was no fever. Pharynx was calm. Peripheral lymph nodes were not enlarged. Nasal breathing was not obstructed, no discharge. In the lungs breathing was conducted in all parts, vesicular, no rales were audible. Respiratory rate (BFR) was up to 22-24 per minute. Heart tones were clear, rhythmic, no murmurs were audible. Heart rate was 118-122 beats per minute. The abdomen was soft and painless. The liver and spleen were not enlarged. Stool and diuresis were not disturbed.

A preliminary diagnosis was made: Mastocytosis. Cutaneous form.

Paraclinically: in the general blood count: hemoglobin (HGB) - 122 g/l (RI: 120-160 g/l); red blood cells (RBC) - 4,5x10^{12}/l (RI: 4.1-5.2x 10^{12}/l); platelets (PLT) - 250x 10^{9}/l (RI: 150 - 450x 10^{9}/l); white blood cells (WBC) - 4.8x 10^{9}/l (RI: 4.5 - 13x 10^{9}/l); lymphocytes (LYMF) - 37% (RI 38-72%); monocytes - 1% (RI:2-10%); stab neutrophils - 2% (RI: 1-5%); segmented neutrophils - 48% (RI: 43-60%); eosinophils - 12% (RI: 0-5%); determining the ESR by the Panchenkov method -10 mm/h (RI: 1-15 mm/h). According to the general blood analysis, eosinophils increased.

Biochemical blood count: total protein 60g/l (56-75 g/l); albumin, 40 g/l (37-55 g/l); alanine aminotransferase (ALT), 43.43 U/L (less than 40 U/L); aspartate aminotransferase (AST), 36.4 U/L (less than 40 U/L); blood glucose, - 4 mmol/L (3.3-6.1 mmol/L); total bilirubin, - 5.2 mol/L (3.4-17.1 μmol/L), creatinine, - 38.5 μmol/L (35-110 μmol/L), urea, - 3.5 mmol/L (4.3-7.3 mmol/L). Immunogram results: immunoglobulin A - 2.3 g/L (RI: 0.21-2.82g/L); immunoglobulin M - 0.68 mg/mL (RI: 0.47-2.40 mg/mL); immunoglobulin G - 12.7 mg/mL (RI: 4.83-12.26 mg/mL); immunoglobulin E - 150 U/mL (RI: 0-60 U/mL); CD3+ - 65.00%(RI: 62.0-69.0%); CD4+ - 35. 00% (RI: 28.1-65.0%); CD8+ - 29.00% (RI: 26.0-68.0%). Conclusion: There was a sharp increase in the content of immunoglobulin E.

The level of total immunoglobulin E was 340 IU/ml.

Antibodies to giardia and Helicobacter pylori were not detected.

Allergoscreen: milk - 1.5 IU/ml.

Abdominal ultrasound examination: slight increase in the size of the spleen (56mmx21mm), which was in line with age standards.

Determination of the level of total tryptase in blood serum in the laboratory "Hemotest" ImmunoCAP - 20 ng/ml. Conclusion: the level of total tryptase was elevated, which may indicate cutaneous mastocytosis.

Dermatologist consultation: there were numerous yellow-pink, urtic, itchy

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spots and papules on the skin of the trunk and extremities. On the skin, when rubbing the elements, their reddening and blistering were noted, which indicated a positive Darier-Unna symptom.

The diagnosis was made on the basis of major criteria: characteristic clinical picture of the rash and positive Darier-Unne's symptom, as well as on the basis of laboratory tests: increase of total tryptase (20 ng/ml) in blood serum and data of instrumental methods of examination: increase of spleen. No manifestations of systemic mastocytosis were revealed. Clinical diagnosis: Mastocytosis, cutaneous form. Pigmented urticaria.

The child was prescribed antihistamines (zirtek drops 5 drops 2 times a day for 30 days), external treatment (emollients and root protectors - emolium cream, atopik, locobase ripea).

Recommendations: Dispensary observation by a pediatrician, allergologist-immunologist and dermatologist. Hypoallergenic diet: exclude cottage cheese, cheese, beef. Medical therapy: antihistamines for up to 1 month. The treatment with emollients and root protectors.

Conclusion: Cutaneous mastocytosis is a rare disease with a relatively favorable prognosis in children. Due to low incidence, diagnosis is often difficult. Therefore, we present a clinical case of a child with the cutaneous form of mastocytosis. All children with mastocytosis should be registered by a pediatrician, an allergist and a dermatologist because of the possibility of transition to the systemic form at an older age.

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PARKINSON DISEASE AND ISCHEMIC STROKE (CLINICAL CASE)

Parkinson's disease (PD) – is a chronic neurodegenerative disease that ranks 2nd in prevalence in the world and has a steadily progressive course. It is clinically manifested by motor disorders in the form of hypokinesia, muscle rigidity and/or rest tremor. In addition, patients have non-motor symptoms, some of which may occur long before the development of typical motor manifestations. Cerebrovascular diseases are in the first place in terms of mortality and disability. A number of studies have revealed that PD reduces vascular risk factors due to low activity of the sympathetic part of the autonomic nervous system, disorders of the hypothalamic-pituitary-adrenal axis, as well as due to treatment with dopaminergic drugs. Contradictory results are expressed by a number of other authors, according to which PD is associated, on the contrary, with an increase in the risk of stroke. This article presents a clinical case of a patient with an established diagnosis of PD who has developed an ischemic stroke. The possible mechanisms of the combination of two diseases, the influence of the neurodegenerative process on the recovery processes and the timing of hospitalization are discussed.

Keywords: Parkinson disease, ischemic stroke, non-motor symptoms, cognitive impairment.

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Introduction. Parkinson's disease (PD) – is a chronic neurodegenerative disease associated with the loss of dopaminergic neurons in the striatum with the accumulation of Levi bodies [1, 2, 5]. Several factors play a role in the development of PD, including genetic predisposition and environmental factors [1, 2]. PD ranks 2nd in prevalence among neurodegenerative diseases after Alzheimer's disease, reaching 1% in the group of people over 60 years old and up to 4% in people over 75 years old [10]. It is clinically manifested by motor disorders (hypokinesia, muscle rigidity, rest tremor) and a wide range of non-motor symptoms (affective, cognitive, sensory, vegetative, and others) [6, 8].

PD can occur in combination with other neurodegenerative and/or vascular diseases of the brain, including acute disorders of cerebral circulation (ONMC) [4, 9]. Stroke is the most important medical and social problem of the elderly, is in the first place in terms of prevalence, mortality and disability [3, 11]. The combination of PD and cerebrovascular diseases can vary from 8.6% to 12% [4]. There is a lot of contradictory data about the relationship between BP and ONMC. It has been established that PD reduces the activity of the sym-