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HEREDITARY ANGIONEUROTIC EDEMA: CASE FROM PRACTICE

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Abstract:

Hereditary and acquired angioedema are a complex problem in clinical allergology, their prevalence has not been studied enough. Hereditary angioedema is rare and accounts for no more than 2% of all cases of angioedema; in the general population, its hereditary nature is detected with a frequency of 1:10,000–1:150,000 people. In the Republic of Uzbekistan, this pathology has not been studied enough, statistical records are not kept. Considering the foregoing, our center set itself the goal of studying the prevalence of hereditary and acquired angioedema nosologies in the republic makes not more than 2% of all angioedema cases, in the total population hereditary angioedema is detected with the rate of 1:10000 (1:1500000 persons.

There are three forms of the disease: hereditary angioedema type I, hereditary angioedema type II and estrogen-dependent edema (type III). The first two forms are due to a genetically determined absolute or relative deficiency of the C1 inhibitor, in the third form its concentration and function are not changed.

Hereditary angioedema, unfortunately, today is not curable, despite the achievements of modern medicine, however, in the case of a full history taking, targeted diagnosis and timely treatment, it is possible to achieve significant positive dynamics in the course of the disease. In medical science, little experience has been accumulated in this nosology, which prompted us to publish a publication on such a rare clinical case.

Keywords: Angioedema, complement, C1-exterase inhibitor

Isolated angioedema is one of the most difficult problems in clinical allergology [1]. For doctors, the disease remains a nosology with a complex mechanism, with characteristic rapidly developing urgent conditions that are difficult to manage, and for patients it is a source of constant anxiety, a cause of disability, and sometimes, unfortunately, death [2]. Angioedema (AE) is the result of a local increase in the permeability of submucosal and subcutaneous capillaries and venules.

Hereditary angioedema (HAE) – autosomal-dominant disease, characterized by episodic angioneurotic edema of any area of the body. [3]. AE prevalence not studied enough. HAE is rare and accounts for no more than 2% of all AE cases, in the total HAE population are detected at a frequency of 1:10 000 – 1: 150 000 people [4]. There are three clinically identical types of HAE that can only be determined by examining the components of a blood complement [5]. There are three forms of the disease: HAE I type, HAE II type and estrogen-dependent swelling (HAE III type). The first two forms are due to the genetically determined absolute or relative deficit of the C1 inhibitor, in the third form its concentration and function are not changed [6]. Usually HAE debuts in the first 2 decades of life, more often in the puberty. In many patients, the disease remains unrecognized for decades; typical erroneous diagnoses are allergic or idiopathic AE and anaphylaxis [7]. Acute bouts of NAE occur spontaneously or under the influence of triggers: injury, stress, surgical interventions, ARVI and others. At the same time, 50% of cases of NAE are provoked by trauma and surgical interventions, and 30 – 40% – by stress [8]. The clinical picture of the NAE is characterized by the appearance of dense painless, not itching swelling, localizing in any part of the body: upper and lower extremities (arms, feet, hips), on the face (lips, eyelids), mouth cavity (tongue, soft sky), body, genitalia, as well as a slider layer of the upper respiratory tract and gastrointestinal tract. The isolated AE is characterized by a slow dynamic of symptoms:

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swelling slowly increases quite slowly within 12 – 36 hours and disappears within 2 – 5 days, abdominal symptoms disappear within 12 – 24 hours. The frequency of edema can vary from weeks to months and once a year [9]. The inefficiency of glucocorticosteroids and antihistamines when purchasing edema [10]. In his work, a doctor of any specialty should have knowledge on the topic of AE and NAE. In medical science, little experience has been gained in this nosology, which prompted us to give an example of such a rare clinical case. Patient J., 28 years old, entered the allergic department of the Republican Scientific and Specialized Allergological Center of the Ministry of Health of the Republic of Uzbekistan on March 2, 2023 with complaints about periodically appearing swelling of the lips, cheeks, chin and subjuncular areas; swelling is dense, pale, not accompanied by itching and urticaria. From the history it is known that for the first-time edema appeared on the face in 2021. (at the age of 26) after treatment by a dentist, since then, they have been steadily recurring with an average frequency of 1 – 2 times in 3 months. Swelling appear spontaneously, localize on the face and in the lip area, pass without a trace independently for 3-4 days, do not require medical attention, there is no effect from taking antihistamines. Triggers are injuries, cold, acute respiratory diseases, stresses, including exams and tests at school, taking bitter, colored drinks. Abdominal pain since 27 years old, 1 – 2 times a year, was regarded as intestinal colic, symptomatic therapy – without significant effect. Since 2022, abdominal pains are repeated monthly, do not decrease under the influence of drugs, pass independently for 2 – 3 days. Anamnesis of life: born in the Tashkent region, grew and developed according to age. He currently works as a welder. Of the transferred diseases: ARVI, lacunar angina, gastroesophageal-reflux disease, reflux-esophagitis of the II degree, chronic gastroduodenitis, enterobiosis, giardiasis. Do not smoke, does not use alcohol. Hemotransfusion was not carried out. Allergological history: when eating bitter products and colored carbonated drinks, swelling appeared in the face, lips, contact with other groups of potential exoallergenes is normal. Heredity: AE from close relatives denies. Upon admission to the clinic, the consciousness is clear, the condition is satisfactory. The physique is correct, according to the hypersthenic type. Height – 185 cm, body weight – 87 kg. BMI – 27.0 kg / m2. The food is elevated. Skin covers and visible mucous pale pink, no rashes. There are no edema and paste. Lymph nodes do not palpate. Muscle and bone system without features. Cardiovascular system: top of the V intercostal push. The boundaries of relative stupidity of the heart: right – 0.5 cm outside the right edge of the sternum in the IV intercostal region, left – 1 cm inside of the left LSKL in V intercrail, upper – at the lower edge of the III rib along the parastronal line on the left. The heart tones are amazingly clear, rhythmic, the rhythm is correct, bicular, there are no noise. MSS – 72 per minute, AD – 120/70 mm RT. Art. Breathing system: breathing through the nose is not difficult. The chest is normal-wall, symmetrical in shape. CSD – 17 per minute. Voting is carried out symmetrically above the entire surface of the lungs. When percussion, a clear pulmonary sound above the entire surface of the lungs. When auscultation, breathing is vesicular over the entire surface of the lungs, there are no wheezing. Digestive system: the tongue is wet, clean, mucous walls of the yoke of physiological color, the tonsils are unincreased, clean. The abdomen is in the correct shape, participates in the act of breathing. A typing sound is determined percutaneous above the surface of the abdominal cavity. The stomach is loose, painless. Liver – 10 × 9 × 9 cm in Kurlov. When palpating, the lower edge of the liver is sharpened, smooth, painless. The upper boundary of the spleen blunt – 9th rib along the front axillary line; the lower – 11th rib along the middle axillary line, the length – 8 cm, does not palpate along Sali. Urinary, endocrine and nervous systems without features.

A biochemical blood test revealed a slight increase in ALT transaminase to 51.2 ED / L, other indicators within normal limits. Immunoglobulin E total – 140.0 ME / ml (norm 0.0 – 100.0). No antibodies to helminths were detected, markers of viral hepatitis were not detected, and there were no antibodies to Helicobacter pylori either. According to the conclusion of an ultrasound examination of the abdominal organs from February 2023 – increase in the left lobe of the liver, seal of the walls of the gall bladder, diffuse changes in liver parenchyma (changes in the type of hepatitis) and pancreas. ECG rhythm is sinus, correct, with a frequency of 75 ud / min. X-ray – pulmonary pathology, mediastinum not detected. Thus, the patient was allocated leading clinical syndromes: local edema syndrome, pain abdominal syndrome. Based on complaints, data from the history, objective examination and the results of laboratory and instrumental studies, a clinical diagnosis is formulated: hereditary angioneurotic swelling with localization on the face, lips, subjuncular area, and abdominal organs. Further, the patient was taken to the dispensary of the clinic of the Republican Scientific and Specialized Allergological Center. Target – examination, study of the indicators of the complement system, verification of the diagnosis and selection of therapy. General and biochemical tests of blood, urine, co-program – without features. Viral diseases of the liver, syphilis, HIV (infection, clay invasions were excluded. Specific immunoglobulins to domestic, epidermal and pollen allergens are negative, sensitization has not been detected. Ultrasound examination of the thyroid gland – pathology not detected. Ultrasound examination of

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the abdominal organs – diffuse changes in the liver and pancreas.

A further patient examination algorithm – blood test of the C1 inhibitor, which confirms the diagnosis. The compliment, the C1 estease inhibitor (functional) – 7% (norm 70 – 130). Complement, C1 estease inhibitor (quantitative) – 0.045 g / l (norm – 0.21 – 0.43). The importance of examining close relatives of the patient should be emphasized, since persons with an unidentified NAE diagnosis in 35% of cases are at risk of death. Indications for long-term preventive therapy are: more than 1 severe attack per month or more than 1 abdominal attack per year or swelling of the face, neck, as well as frequent peripheral edema. This type of treatment is absolutely shown to our patient. The treatment of aminocapronic acid had no significant effect, facial swelling was recidivated. For continuous treatment, a drug from the group of attenuated androgenes – « Danazol », 200 mg, 1 capsule once a day is prescribed. Reception started immediately, with a positive effect, in 2 months, minor swelling of the lips was once observed, abdominal pains never bothered. The patient also needs to recommend the drug « Ikatibant » (« Firasir ») 30 mg, syringe-handle – blocker of bradikininin receptors of the 2nd type, for the purchase of acute situations and life-threatening edema. (in our republic this drug has not yet been registered by the Pharmacological Committee, but by the end of 2023 the RNSAC plans to obtain statistics on the nosologies of hereditary and acquired angioneurotic edema, will form the need for this drug and import into the republic).

The patient is given detailed recommendations for treatment in the future, as well as in the case of acute situations, in preparation for surgical interventions, before invasive research methods, tooth extraction.

Hereditary angioneurotic edema, unfortunately, is not treatable today despite the achievements of modern medicine, however, in the case of a complete collection of anamnesis, targeted diagnosis and timely treatment begun, it is possible to achieve significant positive dynamics during the disease.

After receiving the first results of the treatment, the data will be published by our center.

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