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PIGMENT INCONTINENCE SYNDROMEVLOCH-SULSBERGER

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Annotation: The author describes the problem and clinical case of Bloch-Sulzberger syndrome.

Key words: syndrome Blosn-Sulsberger, clinical, therapy.

Incontinentia pigmenti (IP, Bloch-Sulzberger syndrome or pigment incontinence) refers to genodermatosis, in which abnormalities of the skin and its appendages are combined with damage to other organs and systems of ectodermal origin (teeth, eyes, nervous system), due to a mutant dominant gene localized in X - chromosome; the gene is detailed for a male fetus. Cases of male disease are regarded as the result of spontaneous mutation [3, 4, 6, 8, 9, 11, 12].

The first description of this dermatosis in the literature was made in 1906 by A. Goggad. A. Goggad and Adamson for the first time demonstrated a patient with a clinical picture of pigment incontinence, and Zuchlenthner in 1925 for the first time described the skin manifestations of this dermatosis. The full clinical picture of the disease was described by Bloch and proposed the name of pigment incontinence, and in 1927 Bloch's assistant Sulsberger outlined in detail the clinic of Bloch's syndrome [3, 4]. A more detailed description and systematization of clinical observations was carried out by B. Bloch in 1926 and M. Sulzberger in 1927, and this syndrome got its name from their names.

According to Bloch and Sulzberger, the disease is congenital, with it the function of the pigment-forming cells of the basal layer is disrupted.epidermis and the pigment penetrates into the dermis [4].

According to some authors, the prevalence of the syndromeBloch-Sulzbergeris 0.2 per 100,000 population [13]. According to other data, the population frequency of pathology is estimated as 1:91,000; According to generalized data, more than 650 cases of the syndrome have been described in the literature. More than 90% of patients are female [14]. This is due to the fact that the classical type of IP inheritance among males, as a rule, leads to the development of a lethal outcome [4, 14].

The reason for the developmentIPaccording to some authors (Wadnifhski, 1955; Sprafke, 1963), it is an intrauterine allergization of the fetus, others (Yadasshon and Franceschetti, 1933) are a violation of a neurohumoral, vascular nature [4]. According to other authors (S.I. Dovzhansky and I.I. Pavlova,

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1974), the cause of the disease is trauma and infection of the mother during pregnancy. Other researchers (S.B. Belenky and T.M. Bolshakova (1960), Kyapige (1969)) suggest that maternal infections leading to perinatal damage to the fetus are the main cause of developmentIP[3]. There are also indications of chromosomal abnormalities in patients with pigment incontinence and their relatives (Uhzzat B. et al. 1978) [15]. Special variants of pigment incontinence are:

1. Reticulated pigmented dermatosis (Franceschetti-Yadasson syndrome).

2. Dermatosis reticular pigment Negeli (Naegeli).

3. There is also an abortive type of pigment incontinence - Asboe-Hanzen syndrome (syn. Pigmentary keratogenic bullous dermatitis, described by G. Asboe-Hanzen in 1953).

4. There is also an achromatic variant of the disease - Ito's hypomelanosis.

According to some researchers, in the clinical picture of the Bloch-Sulzberger syndrome, three stages are distinguished: bullous, papulo-verrucous, pigmented (Zgaise, 1963. Steinluht L.A., Zverkova F.A., 1976) [6].

Bullous stage - occurs already in the first hours of life, sometimes from the moment of birth in the form of edematous erythema with vesiculo-bullous, less often urticarial elements, which tend to be linear. The contents of the vesicles are usually transparent; when they open and dry, small erosions and crusts form. Rashes occur in attacks, spreading to new areas of the skin. Their most common localization is the limbs. Mucous membranes are not affected. In the general blood test, leukocytosis and eosinophilia are noted (Carney RG, 1976).

Papulo-verrucous stage - develops after about 2-3 months. and is characterized by the appearance of lenticular keratinizing papules, located predominantly linearly in the zone of former vesicles or randomly, often resembling a warty nevus. Verrucous skin changes persist for several months.

The pigment stage usually develops after 5-6 months. from the onset of the disease and is characterized by brownish-yellow patches of hyperpigmentation with lighter, irregularly shaped edges ("dirt splashes"). Sometimes their pattern is branched, linear (along the nerves) in the form of sinuous parallel symmetrical ribbons located mainly on the skin of the abdomen and less often on the limbs. In some cases, all stages of the disease can exist simultaneously, but more often papuloverrucous and pigmentary stages are observed. After puberty (2nd to 3rd decade), hyperpigmentation gradually subsides, and areas of hyperpigmentation may develop mild atrophy.

Skin manifestations develop mainly for the first time days or weeks of life, when spotted erythema appears on the body of the limbs, in places with peeling, with vesicle rashes, crusts, and later papules are detected. Some of the elements acquire a verrucous character. Vesicular and papular elements appear paroxysmal once or repeatedly. There is a tendency to a strip-like arrangement of the rash.

On histological examination:in the I (bullous) stage, spongiosis is detected with the formation of vesicles containing a large number of eosinophils. Papulo-verrucous stage II is characterized by acanthosis (up to the phenomena of pseudocarcinomatous hyperplasia), irregular papillomatosis and hyperkeratosis, dyskeratosis, which is more pronounced than in stage I. The pigment stage is characterized by a large accumulation of melanin in melanophages localized in the papillary dermis [3, 6, 7, 10, 11].

IP occurs almost exclusively in women and is often familial (Belostotskaya E.S. et al. 1983) [1]. The ratio of boys and girls is exactly 1:35. In some cases, they can occur in non-familial men [6, 7].

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Criteria (major and minor) for establishing the diagnosis of IP, used today in clinical practice, were proposed by Landy and Donnai in 1993 [12].*Big Criteria*: skin lesions that occur in several stages from infancy to adulthood (4 stages of skin damage).*Minor (or Optional) Criteria*: damage to teeth (hypo - and adontia, microdontia, abnormal shape of teeth), hair, nails, retina. Landyand Donnai also include CNS disorders as minor criteria.

Differential diagnosis of this syndrome should be carried out with the following phenotypically similar conditions:

1. Franceschetti-Yadasson syndrome;
2. Goltz-Gorlin syndrome;
3. bullous epidermolysis;
4. Urticaria pigmentosa etc.

The Franceschetti-Yadasson syndrome is also characterized by age spots, however, unlike the Bloch-Sulzberger syndrome, these changes are never preceded by an inflammatory phase and patients have impaired sweating, hyperkeratosis of the palms and feet. There are also no changes from other organs and systems.

The absence of phasing of skin changes, characteristic tumors of the central nervous system (medulloblastoma, astrocytoma) with corresponding neurological symptoms makes it possible to exclude the Goltz-Gorlin syndrome. Differential diagnosis of Bloch-Sulzberger syndrome and phosphate diabetes is not difficult, since phosphate diabetes is not characterized by skin changes, this disease has specific metabolic disorders and radiographic signs of damage to the skeletal system [2]. More difficulties arise with the differential diagnosis of Asbo-Hansen and Ito syndromes. Asboe-Hansen syndrome develops in the first days of life, papular, verrucous and cystic elements appear, located linearly on the limbs. Pigmentation develops only at the sites of rashes. In the achromatic variant of Ito's hypomelanosis, depigmented spots appear in early childhood in areas typical of IP. In the neurocutaneous form of Ito hypomelanosis, CNS disorders (convulsive syndrome, mental retardation) and bone anomalies are noted [4, 5].

Case Description.

Patient N.I. Born in 2002, he entered the second skin department of the AOKVD on June 10, 2019, as directed by a doctor of the military commissariat to clarify the diagnosis.

Complaints of the patient upon admission: damage to the skin of the trunk and extremities. He considers himself ill since early childhood, when for the first time (according to his mother) bubble elements appeared on the skin of the body. Parents contacted doctors at the place of residence and received treatment. After treatment, there was a significant improvement. After a certain time, papules and plaques began to appear in the skin, at times depigmented spots appeared in the lesions.

The patient went to dermatologists and received treatment, which made various diagnoses (toxicoderma, Dühring's dermatosis, vitiligo, pityriasis versicolor). However, there was no effect from the treatment.

Other family members did not suffer from this disease.

On June 10, 2019, the patient underwent a medical examination by the draft board and was sent to the Andijan Regional Department of Internal Affairs to clarify the diagnosis. The patient was consulted by the staff of the department and with a diagnosis of pigment incontinence (Bloch-Sulzberger syndrome) was hospitalized in the skin department.

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On examination: the skin pathological process is widespread and localized on the skin of the trunk and both upper lower extremities. In the lesions there are multiple hyperpigmented spots, yellowish-brown, in places papules and plaques. The location of the elements in the lesions resemble"Mud spatter" (Fig. 1, 2).

On the skin of both hands, elements in the form of spots with peeling onsurface, the foci are located in the form of stripes with depigmented areas.

When checking, the Balzer test and the symptoms of Besnier, Unna are negative.

The patient was consulted by doctors of related specialties - a general practitioner, a dentist, a neuropathologist, no pathological changes were observed, an oculist found iridocyclitis and cataracts.

The results of the laboratory examination. Complete blood count: Hb-102 g/l; er. - 4.1-1012/l; CPU-0.8; lei. - 9.6-10%, fell. -2%; seg. - 59%; eoz. - 6%; limf. - 29%; mon. -4%; ESR - 22 mm/hour. Urinalysis: clear, rel. sq.-1024; lei. - 5-6; er. 0-1 in sight; protein - abs. Blood sugar 4.4 mmol / l., Wassermann reaction - neg.

Based on the above data, the patient was clinically diagnosed with pigment incontinence (Bloch-Sulzberger syndrome).

The patient was prescribed symptomatic treatment without effect.

Thus, in this case, the Bloch-Sulzberger syndrome was diagnosed in a man, was not familial in nature, and a benign course was noted.

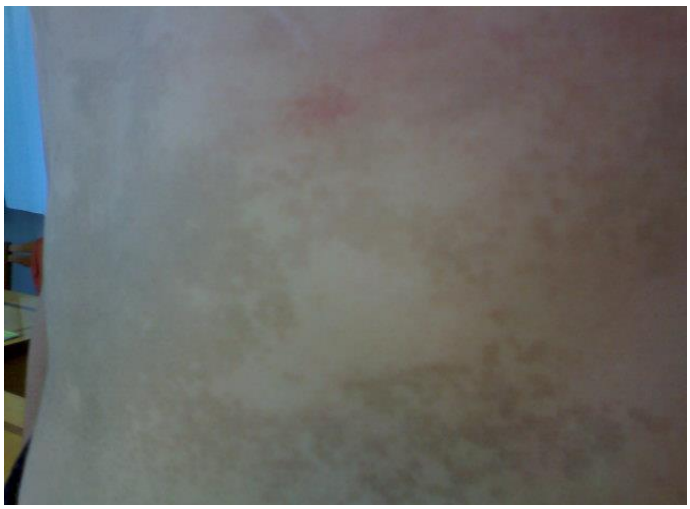

Fig. 1. Patient with Bloch-Sulzberger syndrome. Lesions located in the back.

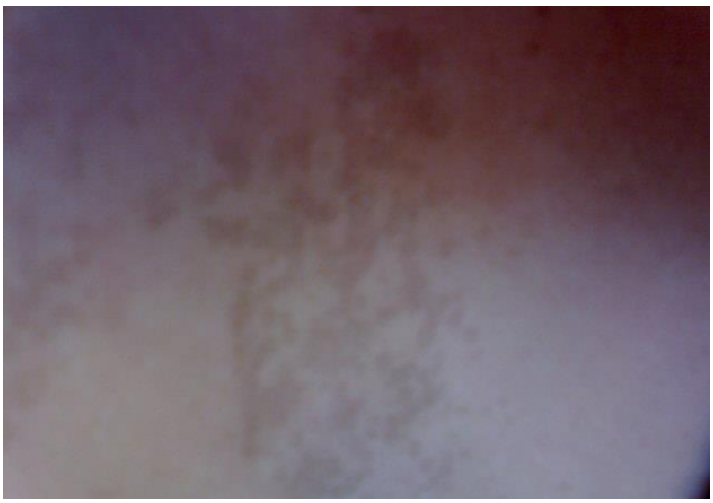

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Fig. 2. Patient with Bloch-Sulzberger syndrome. Lesions located in the abdomen.

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