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Clinical Case of Congenital Ichthyosis

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Abstract

In this clinical observation is given an example of lamellar ichthyosis A distinctive symptom of this type of ichthyosis is the birth of a child in a collodion film resembling plastic. A few weeks after delivery, this film gradually breaks down and peels off, and the child shows one of two types of ichthyosis: lamellar or ichthyosiform erythroderma. This process is dangerous because it also increases the risk of infection with other infections, dehydration, instability of body temperature. In this regard, this clinical case once again alarms practitioners and refreshes theoretical and practical knowledge concerning "Lamellar ichthyosis". It is impossible to completely recover from ichthyosis, but it is possible to significantly improve the quality of life.
Keywords: ichthyosis, lamellar, congenital, colloidal, closely related marriage, hyperkeratosis

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Клинический случай врожденного ихтиоза

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Резюме

В данном клиническом наблюдении приведен пример ламеллярного ихтиоза. Отличительный симптом этого вида ихтиоза – рождение ребенка в коллодийной пленке, напоминающей пластик. Спустя несколько недель после родов эта пленка постепенно разрушается и отслаивается, а у ребенка проявляется один из двух типов ихтиоза: ламеллярный или ихтиозиформная эритродермия. Этот процесс опасен тем, что также увеличивает риск заражения другими инфекциями, обезвоживания, нестабильности температуры тела. В связи с этим этот клинический случай еще раз насторожит

практических врачей и освежит теоретические и практические знания, касающиеся ламмелярного ихтиоза. Полностью вылечиться от ихтиоза невозможно, но можно значительно улучшить качество жизни.
Ключевые слова: ихтиоз, ламеллярный, врожденный, коллоидный, близкородственный брак, гиперкератоз

■ INTRODUCTION

Ichthyosis (syn.: diffuse keratoma, sauriasis) is a hereditary disease characterized by a diffuse keratinization disorder of the hyperkeratosis type and manifested by the formation of scales on the skin resembling fish scales. It is inherited both by dominant and recessive type [6, 11; pp. 6–7]. There are four main forms:

a) vulgar ichthyosis;
b) ichthyosis linked to the X-chromosome;
c) autosomal recessive congenital ichthyosis;
d) congenital bullous ichthyosiform erythroderma [12, 15; p. 7].

Ichthyosis vulgaris is the mildest form of ichthyosis and the clinical picture includes 3 signs: peeling, follicular hyperkeratosis and increased folding (hyperlinearity) of the skin of the palms and soles [13; p. 7]. Hyperkeratotic scales are located mainly on the extensor surfaces of the limbs and are most pronounced on the anterior and lateral surfaces of the shins. The skin on these areas is dry, the keratinized scales are large, thick and dark and they are difficult to exfoliate. Follicular hyperkeratosis (hyperkeratosis of the mouths of hair follicles) is best seen on the extensor surfaces of the shoulders and hips. The dryness of the skin significantly decreases in summer, especially when staying in a warm and humid climate and at sea, and it increases in winter [5, 7; p. 6].

Lamellar ichthyosis is inherited by autosomal recessive type. This form was described by E. Seligman (1841) as "epidermal desquamation of newborns". The term "lamellar ichthyosis" was introduced by L. Gross and L. Torok (1895). According to F.J. Bling, Rook (1968), this form of congenital ichthyosis occurs with a frequency of one case per 300,000 births. The disease occurs among different ethnic groups of the population, more often this pathology is found in families with closely related marriages [18, 19; p. 7]. The disease manifests itself at the birth of a child with a picture of the so-called "collodion" fetus. This term was first used when describing congenital ichthyosis X. Halepo, R. Watelet (1892). However, the collodion fetus may be the initial manifestation of not only lamellar ichthyosis, but also other forms: sex-linked ichthyosis; epidermolytic ichthyosis; Sjogren-Larsson syndrome [17, 20, 21; p. 7].

The pathogenesis of lamellar ichthyosis is dominated by insufficiency of the enzyme transglutaminase and proliferative hyperkeratosis (intensive synthesis of keratin as a result of a sharp increase in the functional activity of epidermal cells) [2, 14, 23; pp. 6–7].

It usually exists from birth, there are no bullas, a newborn is usually covered with a "collodion film", which within 1–2 days turns into large scales on sharply reddened skin [8; p 6.]. In severe cases, the skin is covered with a thick white horny shell with deep red cracks, from which a bloody fluid is released. There is palmar-plantar hyperkeratosis with cracks, increased growth of hair and nails (with deformation), but at the same time there is a decrease in sweat and fat [1, 16; pp. 6–7]. Ectropion. It is often associated

with endocrine and neurological disorders, ocular pathology, hearing loss, and various developmental abnormalities. The disease often has a fatal outcome. In contrast to the dominant form of congenital ichthyosiform erythroderma, histological signs in this form are nonspecific. There is moderate hyperkeratosis with focal parakeratosis [14; p. 7]. The granular layer is often thickened. Acanthosis and chronic inflammatory infiltrate in the dermis. Autoradiographic examination of the skin showed an increase in the proliferative activity of epidermal cells. The number of keratinosomes and the content of intercellular cement is increased, which enhances the adhesion of horn cells [5, 9; p. 6].

Differential diagnosis of lamellar ichthyosis should first of all be carried out with other ichthyosiform genodermatoses accompanied by erythroderma: nebulose congenital erythroderma, desquamative erythroderma Leiner – Moussou, erythrodermic forms of psoriasis, Netherton syndrome, etc. [3, 17, 22; pp. 6–7].

Recessive X-linked ichthyosis affects only men and is more severe than with vulgar ichthyosis. The disease begins at 2–6 weeks of life, the skin lesion is localized in the folds, on the back of the neck, in the elbow and knee bends, on the trunk, and the face, palms and soles are free of the lesion. The rashes are represented by large dark brown dense scales, which makes the skin look dirty, as if it has not been washed for a long time. The skin condition of patients does not improve with age [4, 5, 24; pp. 6, 8].

■ CLINICAL OBSERVATION

Below is a clinical case of congenital ichthyosis under our supervision. Patient T.A. entered to the Department of pediatric dermatology of the TashPMI clinic on 01.03.23. Upon admission, complaints were made of pronounced about dryness, a feeling of tightening of the skin, peeling with large scales, itching of moderate intensity. During pregnancy, prenatal diagnostics was carried out – ultrasound of the fetus in screening, were not detected. deviations. Repeatedly received outpatient treatment at the place of residence. The treatment gave a brief temporary effect. He also received inpatient treatment in the dermatological department of the TashPMI clinic. Delivery on time, natural, at 38 weeks. Birth weight 3200 g, height 52 cm, head circumference 36 cm, chest circumference 35 cm. Apgar score 6/8 points.

Father and mother are somatically healthy. The dermatological history of both parents is not burdened. When collecting a family history, it was not possible to establish the presence of ichthyosiform manifestations in the patient's relatives in four generations.

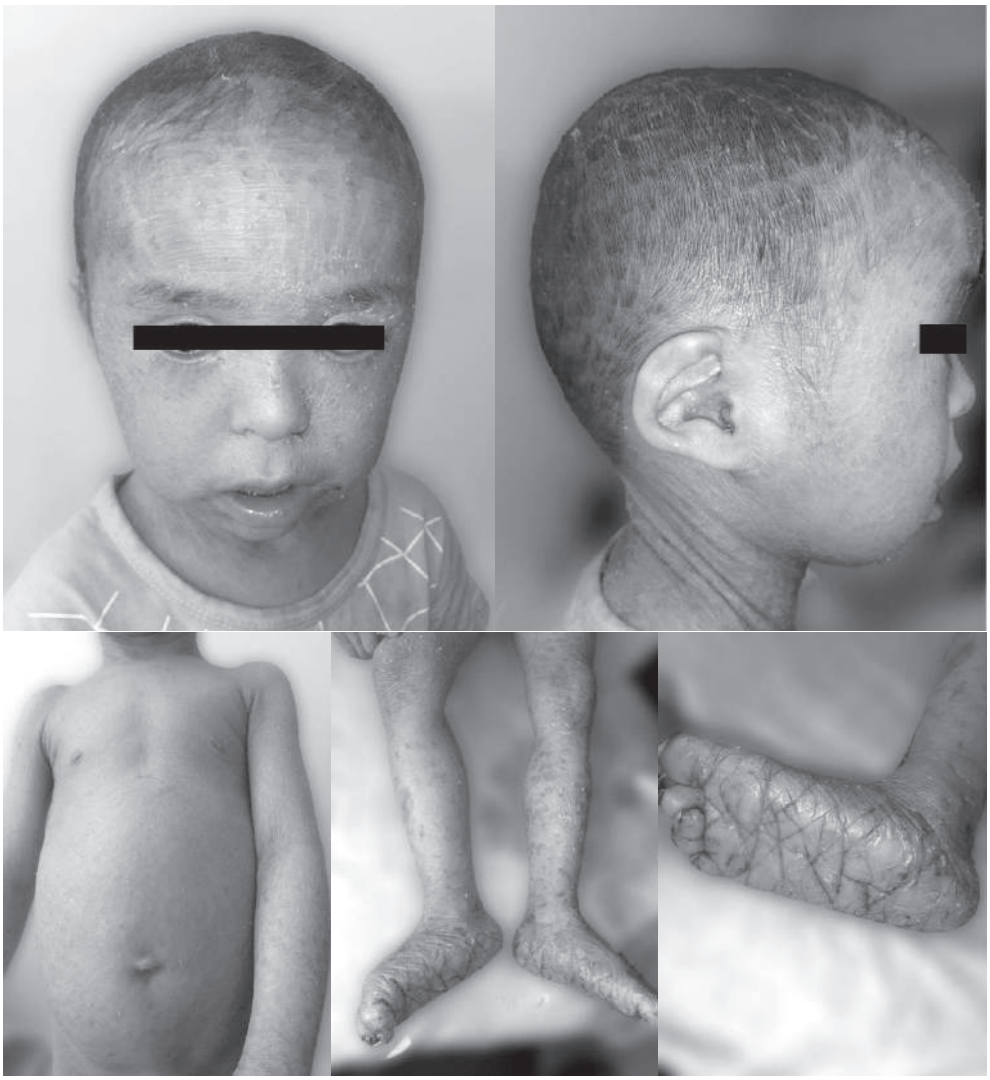
On objective examination, the condition is satisfactory, the consciousness is clear, the position is active. Body temperature 36.6 °C. Visible mucous membranes are clean. The lymph nodes are not enlarged. Musculoskeletal system without pathologies.

Breathing is vesicular. The frequency of respiratory movements is 17 per minute. The heart tones are clear, the rhythm is correct. Arterial pressure is 100/60 mm Hg. The abdomen is of the correct shape, symmetrical, participates in breathing, soft, painless with palpation. The liver is palpated along the edge of the right costal arch. Bubble symptoms are negative. Pasternatsky's symptom is negative. There is no soreness during palpation of ureteral points. Healthy bladder and bowel habits.

Local status: the pathological skin process is widespread, symmetrical, subacute-inflammatory. Against the background of hyperemic, moderately infiltrated, dry skin, large-plate peeling is noted by dense yellow-brown scales with a diameter of 1–1.5 cm, arranged in vertical rows, tightly attached in the center and slightly rising along the

periphery. In the area of the knee, ankle and elbow joints, the skin is thickened, lichenized, with dense hyperkeratotic layers. The skin in the folds of the neck had a linear crack. In the armpits, the skin is slightly macerated. The skin feels dry, tight. On the scalp, the sebaceous crusts are dense and hard. Subjectively: moderate itching.

The auricles are not deformed, pressed against the head. There was an accumulation of horny masses in the nasal and auditory passages. On the scalp, hair growth was preserved. The nail plates of the hands and feet are not changed. The skin of the face is yellow-brown, dry, lichenized, with radial orientation of wrinkles around the mouth, the outer corners of the eyes are raised, pronounced ectropion is noted (fig.).



Patient's appearance

A number of laboratory tests were carried out:

General blood test: Hb – 88g/l, Er – $3.1*10^{12}$/l, Colo index – 0.8, leuc – $6.9*10^{9}$/l, ESR – 7 mm/h.

Blood biochemistry: ASLO+, CRP–, RF+++, Ca 1.9 mmol/l.

General urinalysis: leukocyturia (leukemia 6–7–8).

General. Stool analysis: without pathologies.

Ultrasound of the liver: Diffuse liver changes.

Ophthalmologist: Inversion of the lower eyelids. Bacterial conjunctivitis.

Otorhinolaryngologist: no acute ENT pathology was detected.

Neurologist: no neurological pathologies have been identified.

Based on clinical manifestations and anamnesis data, the main(clinical) diagnosis was made: Congenital (lamellar) ichthyosis, moderate-severe course.

Concomitant diagnosis: hypochromic anemia of the I degree.

Treatment was carried out in the clinic:

1. Aevit – 1 cap 2 times a day.
2. Calcium gluconate 10% – 3.0 ml + saline 0.9% – 30.0 ml 1 time a day.
3. Neuromultivitis – ½ tab 2 times a day.
4. Supralgan 2% – 0.4 ml 1 time a day in /m in the evening.
5. Folic acid – ½ tab 2 times a day.

Locally:

1. Dexeryl cream + Aevit oil 2 times.
2. Salicylic ointment 2% – 2 times a day.
3. Mupiroban ointment – 2 times a day at the site of erosions.

He was discharged from the hospital with clinical improvement, the therapy helped to reduce hyperemia of the skin, the intensity of itching and peeling of the skin, softening of hard scales, epithelization of painful cracks.

■ CONCLUSION

The interest of this clinical case is the relatively rare occurrence of lamellar ichthyosis in the practice of a dermatovenerologist, a combination of skin symptoms with pathology of the bone system and the organ of vision. Despite the fact that in this patient, when collecting anamnesis, it was not possible to establish the presence of ichthyosiform manifestations in relatives, dermatovenerologists should pay attention to timely medical and genetic counseling of patients, their relatives and married couples in case of detection of hereditary dermatoses in the anamnesis.

This contingent of patients is subject to dispensary observation and complex treatment in order to maintain an acceptable level of quality of life.

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