

## Congenital Ichthiosis

(Case Report)

*Irma Manjavidze, Inga Mamuchishvili, Maka Rizvadze, Mzia Ugrechelidze, Karaman Pagava*

Department of Pediatrics, Tbilisi State Medical University

**I**chthiosis, known as a skin of aligator, is a diffuse keratosis. For the first time it was described in the manuscripts of ancient Chine and Egypt, 1-3 thousand year before Christ.

Inborn ichtiosis is the congenital anomaly of skin keratosis, developing during IV-V month of prenatal period. Pathogenesis is unknown.

In infants the inborn ichthiosis may be revealed in following different forms: vulgar ichthiosis, ichthiosis linked with chromosome, platform ichthiosis (erythrodermia similar to ichthiosis, nonbullar, congenital) and epydermolizing hyperkeratosis (bullar, congenital and erythrodermis similar to ichthiosis).

Our case report refers to platform ichthiosis described by L.Brocka in 1902. Transfers by autosomal-recessive type.

The disease is very rare, platform ichthiosis is the most serious form of this pathology. Rate of the disease is 1:300000. Reveals to as the congenital general lession of the skin.

Case report: in Tbilisi experimental delivery home of N1 clinical hospital, was born an infant (2nd May of 2001). It was the third pregnancy and third delivery of the woman. Both pregnancy and delivery completed physiologically. The duration of the first stage of the delivery was 7 hours and 15 minutes, II stage - 30 minutes fluid free period consisted of 6 hour and 45 minutes, fetal third was clean with a little amount.

Newborn was delivered alive, fullterm, couldn't be estimated by Apgar score, because of uglinen. Weight 3400 gr, length 52 cm.

Objectively: general lession of the skin, covered by thick, compact white - grey covery with deep red

coloured fissures excreting bloody secretion with strong decompositive smell. Ectopic protract eyelids, mild cranial bones, ectopic nose with hardly mentioned holles, deformed curricula, yawning position mouth so called fish mouth.

Deformed palms and foots syndactilia, protracted thorax, respiratory difficulties. Lung auscultation reveals poor respiratory sounds, heart tones were muffled systolic murmurs could be auscultated. Flatulented abdomen. Enlarged liver margins below the costal arch for 4 cm. The urination and meconium are not performed. Hyperemia of the ectopic pudendal labies: anal orifice suction reflex presented.

The newborn was consulted by the genetics: parents refused to conduct cytogenetical test.

Female newborn had the typical symptoms of ichthiosis.

Mother from the first pregnancy also delivered the baby with congenital ichthiosis, who died a few days later at the delivery home. As she said the baby had skin lessions only on the abdomen and joint areas.

From the second pregnancy she delivered practicaly healthy male baby.

The form of the ichthiosis can be determined by the specific dermatological and ophthalmogical tests. Taking family history and infant gender under the consideration we conclude that this is the case of autosomal-recessive transfere.

72 hours later newborn died.

Pathanatomical and morphological investigation has been conducted with following datas: right lung 8-5-2 cm. left lung 8-5-2,4 cm with deflated, red coloured posterior - inferior fegment.

Thymus 4-3,5-1 cm, heart 4,5-3,5-2 cm, oval orifice was opened, the wall of the left ventricle thickened a little. Liver 11-6,5-3,5 cm., spleen 5-3-1,5 cm, internal organs prethors, kidneys (right 5-3-2,5 cm, left 4,5-2,5-2 cm) with pink pyramides gall-bledder length - 4 cm, width 1 sm full of bills, pancreas 5-1 cm.

Big brain hemispheres - N. cerebellum of a little site.

Micromorphology: perivascular oedema of the brain. cerebellum without pathology. partial atelectasis of the lung, interstitial pneumonie, presence of the haemosiderine. Miofibrile dystrophia of the heart. Thimus stromal nidus fibrinoid necrosis. Perivascular cell infiltrarion of the liver. Delated pancreas ductus, with moderately presented connective tissue. Unmatured glomerules with mesenchimal cell proliferation and systemic dilatation of the tubules in the kidneys. Pulp hyperplasy of the spleen with clear plethoru. Intestines, uterus, ovaries without pathological changes, expressed hyperkeratosis, multiple ichthiosis of the skin multyryces.

Pathologanatomical diagnosis: Innate multyvices inborn ichthiosis, atrial septum defect, microstomia, hypoplasia of the auricula, aplasy of the nose, deformation of the extremities, joints, mild nidus displasia of the Kidneys: partial atelektasis, interstitial pneumonie of the lung, spleen pulp hyperplasia. Protein dystrophia of the liver and kidneys.

Basic cause of death are multiple congenital anomalies, some of them are uncompatible with life. On the basis of the clinical, pathanatomical and morphological

investigations the diagnosis of the platform ichthiosis has been confirmed (nonbullar inborn ichthiosiform erythrodermis). By the genetics the following recommendations have been given to mother: on the 10-12 week of the gestation in the case of the next pregnancies the byopsia of the chorion and the test on DMM should be certainly done to reveal the congenital abnormalitis, as the risk of the next fetus maldevelopment is 25%.



## References

1. Nizharadze G., Pavlenishvili I. Diseases of newborns. / Textbook-Publishing House Ganatleba, Tbilisi, 1990. p. 497-499.
2. Koznova S., Felikova N., Semenov E., Blenikova O. Hereditary syndroms and medical-Genetic Consultation, Publisher - "Practice" - Moscow, 1996. p. 182-186.
3. The Merck Manual. - Vol. 2. p. 624-625. 20th edition.
4. American journal of diseases of Children. November, 1998, vol. 147, N21. p. 36-38.

## **Врожденный ихтиоз**

*Инга Маджавидзе, Инга Мамучишвили, Мака Рижвадзе, Мзия Угрехелидзе,  
Караман Пагава*

Кафедра педиатрии и подростковой медицины  
Тбилисского государственного медицинского университета

### **Р Е З Ю М Е**

В экспериментальном родильном доме №1 клинической больницы родился новорожденный весом 3,400 г, ростом 52 см. Выражено было генерализованное поражение кожи, она была покрыта плотными беловато-серыми чешуйками, глубокими трещинками в местах сгибания и кровянистым выделением кровяного оксидата с резким запахом. Веки эктопированы, вывернуты, кости черепа мягкие, сетчатые, нос эктопирован, ушные раковины деформированы. Рот открыт - поза зевания. Ладони и стопы деформированы, грудная стенка выбухает, дыхание затруднено, дыхание проводится с трудом. Тоны сердца приглушены, во всех точках выслушивается шум. Живот вздут, плотный, печень выступает из под края реберной дуги на 4 см, селезенка на 2 см, мочеиспускания не было, меконий нет. Было проведено медико-генетическое консультирование DS. Пластинчатый ихтиоз (Ихтиозформная эритродермия, небулезная врожденная).