Bloch-Sulzberger syndrome — a rare X-linked dermatosis

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Objective. Bloch-Sulzberger syndrome is a rare genetically determined dermatosis, manifested by skin changes in combination with pathology of the eyes, skin appendages, teeth, central nervous and musculoskeletal systems. The disease manifests in the neonatal period and has four stages: vesicular, verruciform, hyperpigmented and hypopigmented. It occurs mainly in girls, since inheritance has an X-linked dominant character.

Case report. A 7-year-old girl, appealed to the department in satisfactory condition, with complaints of mental deficiency, reduced memory, behavior disorders, multiple hyperpigmented spots on the limbs and body. The child after birth had multiple vesicles on an erythematous background on the body, localized linearly, mainly on the flexor surface of the limbs, as well as on the skin of the chest and back. After birth, he was diagnosed with progressive erythema and vesicular rash. In connection with skin manifestations, she was examined for intrauterine infections — pathology wasn't detected. The patient had no family history of skin diseases. Subsequently, the appearance of hyperkeratosis in the form of plaques, warts and lichenoid growths along the Blashko lines was noted in the affected areas of the body. The girl was under the supervision of a pediatrician. During the examination in the Department revealed: mental retardation, hyperpigmented spots of gray-brown color with light edges, on the extremities of the rash in the form of "mud splashes", on the trunk — in the form of a "spiral", "ring" or "marble cake", with the location of pigmentation along the lines of Blashko.

Results. The course of Bloch-Sulzberger syndrome is suspected in a child with clinical manifestations of mental retardation, stages of the skin process with a debut in the newborn period. To confirm the diagnosis, a molecular genetic examination was performed: mutations of the IKBKG gene, which is located on the X-chromosome, were detected.

Conclusion. Typical skin manifestations allow you to clinically suspect the presence of Bloch-Sulzberger syndrome in newborns, to conduct timely molecular genetic diagnostics to make the correct diagnosis. In view, the importance of differential diagnosis in the neonatal period and the rare disease, a multidisciplinary team of neonatologists, pediatricians and neurologists to this disease is necessary.