A CASE OF LOWE SYNDROME IN A 4-YEAR-OLD GIRL

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Relevance. Lowe's syndrome is a rare multisystem disorder characterized by congenital cataracts, glaucoma, mental retardation, seizures, postnatal growth retardation, and renal tubular dysfunction with chronic renal failure. The disease is inherited in an X-linked pattern, the prevalence is 1/500,000, and men are often affected. In children, clinical symptoms are characterized by renal, neurological, and ocular abnormalities. Lowe's oculo-cerebro-renal syndrome occurs as a result of mutations in the OCRL gene (Xq25), leading to the accumulation of phosphatidylinositol (4,5)-bisphosphate, impaired membrane transport and impaired remodeling of the actin cytoskeleton. In the kidney, disruption of endosomal transport reduces protein reabsorption in the proximal tubule. In the eye, abnormal actin remodeling leads to disorganization of the embryonic lens epithelium and abnormal development of the trabecular meshwork, which regulates the outflow of aqueous humor from the eye.

The purpose of the study is to study the features of the clinical course of Lowe's syndrome in children.

Material and methods. A 4-year-old girl was admitted to the nephrology department of the children's clinic in Andijan. The patient underwent objective, laboratory, instrumental and genealogical research methods**.**

Research results

Complaints from parents about a decrease in daily urine output, swelling in the face and abdomen, moodiness, decreased appetite, sometimes constipation,

strabismus, mental and physical development retardation**.** ***Anamnesis vitae:*** a girl was born from the fifth pregnancy, from a closely related marriage (paternal godchildren). The mother's pregnancy proceeded against the background of severe toxicosis, TORCH infection (cytomegalovirus) and severe deficiency anemia. She took drugs with teratogenic effects (acyclovir, drotaverine). The birth was complicated (chronic fetal hypoxia and asphyxia of the newborn). ***Anamnesis morbi:*** the initial symptoms of Lowe's syndrome in the patient were observed at an early age and were treated with a diagnosis of Congenital nephrotic syndrome, without renal dysfunction. Related: Protein-energy deficiency I-II degree. Congenital strabismus. Deficiency anemia of moderate severity. From an early age, the patient often suffers from intercurrent illnesses, such as acute respiratory viral infections, laryngitis, bronchitis, tonsillitis, for which reason she is registered at the dispensary as a “frequently ill child”. ***Objectively:*** the girl’s general condition at the time of examination was severe due to symptoms of general intoxication. He doesn’t make contact well, doesn’t speak, doesn’t answer questions. The girl is significantly behind her peers in mental and physical development. There is swelling on the face and abdomen. Stigmas of dysembryogenesis were discovered: strabismus, low forehead, dental diastema, anomaly of the jaw bones and auricles, short neck, short frenulum. Peripheral lymph nodes (submandibular) are enlarged in the size of a pea, painless, mobile**.** Osteoarticular system: chest deformity in the form of “shoemaker’s chest”, “X” shaped deformity of the legs. Muscular system: generalized muscle hypotonia, decreased and absent deep tendon reflexes. Auscultation: puerile breathing in the lungs, no wheezing, heart sounds are muffled and rhythmic. Pulse - 115 beats per minute. Blood pressure 80/50. The abdomen is soft, enlarged due to ascites, the liver is enlarged + 3 cm, painless, the spleen is not enlarged. There are no changes in the kidney area, Pasternatsky's symptom is negative on both sides. Nocturia was detected (400 ml during the day, 200 ml at night), relative density from 1003 to 1009. Stool is irregular, constipation is noted. ***Lab tests;*** general blood test: hemoglobin - 68 g/l, erythrocyte 2.1x1012, leukocyte - 9.5x109, ESR - 18 mm/hour. General urine analysis: quantity - 50 ml, specific gravity - 1006, protein in urine -

2.89 g/l, squamous epithelium - 6-7, renal epithelium - 5-7, leukocytes - 3-6, altered erythrocytes - 3-5, unchanged red blood cells - 0-2, hyaline casts - 3-5. Biochemical blood test: hypoproteinemia (total protein - 37 g/l, hypoalbuminemia (16%), urea - 26.5 mmol/l, creatinine - 360 μmol/l, hypercholesterolemia (12 mmol/l), calcium - 1.4 μmol/l Ultrasound of the kidneys - diffuse changes in the renal parenchyma like glomerulonephritis. Hypoplasia and pyeloectasia of both kidneys. *Consultation with an ophthalmologist:* congenital strabismus and retinopathy. *Consultation with a neurologist:* second degree mental and psychomotor development retardation. *Consultation with a traumatologist-orthopedist:* rachitic bone deformity. *Genealogical research:* X-linked, autosomal recessive type of inheritance. Based on the results of clinical and laboratory studies, the following **clinical diagnosis** was established: main: Tubulopathy. Lowe's syndrome (oculo-cerebro-renal syndrome); associated: Nephrotic syndrome. Severe deficiency anemia. Chronic tonsillitis is a toxic-allergic form. ChBD. Recommended: conservative, symptomatic treatment. Kidney transplantation.

Conclusion. In the clinical course of Lowe's syndrome in children, closely related marriage, the teratogenic effect of various drugs during the first trimester of pregnancy in the mother, TORCH infections, anemia, hereditary diseases in parents that require clinical and geneological examination before and during pregnancy in the mother play an important role. and in the child after birth.