

RISK FACTORS FOR LOW SYNDROME IN CHILDREN

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Introduction. Hereditary diseases of the urinary system in children often develop gradually, and only when the child lags behind in physical development and the first signs of renal failure are detected in an advanced stage, a pediatrician or nephrologist is consulted. It should be noted here that although there are various phenotypic stigmas of dysembryogenesis, also in the internal organs, including stigmas of the kidneys or small developmental anomalies, doctors do not always pay serious attention to this, which leads to late diagnosis of the disease.

The purpose of the study is to study the risk factors for the development of Low's syndrome in children.

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

Results. *Complaints* according to the parents about the lag in mental and physical development, strabismus, decrease in daily urine, swelling on the face, abdomen, lethargy, capriciousness, loss of appetite, constipation.

Anamnesis vitae: the girl was born from IV pregnancy, from a closely related marriage (on the mother's side). The mother's pregnancy proceeded against the background of severe toxicosis, herpetic infection and moderate deficiency anemia. Childbirth proceeded with complications (placental abruption, fetal asphyxia).

Anamnesis morbi: the initial symptoms of the disease in the patient were observed at the age of 1 year and were treated with a diagnosis of Congenital nephrotic syndrome, without impaired renal function. Companion: Protein-energy malnutrition II-degree. Strabismus. Anemia deficient II-degree. The treatment carried out was ineffective. Since childhood, the patient often suffers from

infections of the upper respiratory tract (4-5 times a year), such as SARS, bronchitis, tonsillitis, rhinitis, which is why she is registered as a "Frequently ill child" in the dispensary. *Objectively*: the general condition of the girl at the time of examination is severe due to symptoms of general intoxication. The girl is significantly behind in mental and physical development from her peers. He makes contact poorly, does not speak, does not answer questions. Edema is noted on the face and abdomen. *Stigmas of disembryogenesis* were found: strabismus, anomalies in the development of teeth and jaws, low hair growth, short neck, low forehead, low and protruding ears, short frenulum. Peripheral lymph nodes (submandibular, cervical) are enlarged in size, painless, mobile. Osteo-articular system: "O" shaped deformity of the legs, deformity of the chest in the form of "chicken breast". Muscular system: hypotonia, absence of deep tendon reflexes. *Auscultatory*: breathing in the lungs is vesicular, heart sounds are muffled, rhythmic. Pulse - 110 beats per minute. Blood pressure 90/60. Breathing in the lungs is puerile, no wheezing. The abdomen is soft, enlarged due to ascites, the liver is enlarged + 2 cm, painless, the spleen is not enlarged. There were no changes in the kidney area, nocturia was noted, that is, the amount of urine during the day was 500 ml (during the day 200 ml, 300 ml per night), relative density from 1004 to 1010. Pasternatsky's symptom is negative. Chair irregular, constipation. *Lab tests*; complete blood count: hemoglobin - 65 g/l, erythrocyte 2.0×10^{12} , leukocyte - 9.2×10^9 , ESR-15 mm/hour. Urinalysis: quantity-50ml, specific gravity-1007, protein in urine-2.8g/l, squamous epithelium-6-8, renal epithelium-5-6, leukocytes-3-4, altered erythrocytes-3-4, unchanged erythrocytes-0-3, hyaline cylinders-3-4. *Biochemical blood test*: hypoproteinemia (total protein-38 g/l, hypoalbuminemia (15%), urea-23.3 mmol/l, creatinine-320 $\mu\text{mol/l}$, hypercholesterolemia (10 mmol/l), calcium-1.2 $\mu\text{mol/l}$.

Ultrasound of the *kidneys* is a diffuse change in the renal parenchyma of the type of glomerulonephritis. Hypoplasia of both kidneys, left-sided pyelectasis.

Oculist consultation: strabismus and retinopathy.

Consultation of a neurologist: lag in mental and psychomotor development.

Consultation traumatologist-orthopedist: bone deformity.

Genetic study: X-linked, autosomal recessive inheritance pattern.

Based on the results of clinical and laboratory studies, the following clinical diagnosis was established: main: Tubulopathy. Lowe's syndrome (oculo-cerebro-

renal syndrome); concomitant: Nephrotic syndrome. Anemia deficient severe. Chronic tonsillitis is a toxic-allergic form. Frequently ill child.

Recommended: conservative, symptomatic treatment. Kidney transplant.

Conclusion. An important role in the development of tubulopathy, including Lowe's syndrome, is played by hereditary diseases in parents, exposure to adverse environmental circumstances, infections, consanguineous marriages and the teratogenic effect of various drugs during the first trimester of pregnancy in the mother, which require genetic testing during pregnancy in the mother and after the birth of a child.