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# Alagille syndrome in the practice of a pediatrician

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Alagille syndrome (Q.44.7) is a multisystem disease with an autosomal dominant type of inheritance, due to have mutations in the gene JAGGED1, characterized by liver damage, pathology of the cardiovascular system, changes in the eyes, skeleton, kidneys, as well as characteristic craniofacial symptoms. It occurs with a frequency of 1: 70,000 newborns.

**Aim:** to introduce the clinical case of the patient of Regional Children's Clinical Hospital with Allagille syndrome.

**Materials and methods**: Patient E. (2 years old) was admitted with complaints of poor weight gain; psychomotor and physical retardation convulsive attacks with short-term impaired consciousness. A child from I pregnancy, proceeding against the background of placental insufficiency, ulcerative colitis.

Childbirth at gestational age 35 weeks, by cesarean section. Birth weight 1600g, height 45 cm. Apgar score 5-6 b., was on mechanical ventilation-7 days. She had congenital heart defect, which was operated on. Perinatal hypoxic-ischemic damage to the central nervous system. Conjugation jaundice. At 7 months the diagnosis was: congenital malformations of the central nervous system (external-internal hydrocephalus, thinning of the corpus callosum), myopathic syndrome, delayed psycho-speech and statokinetic development. Symptomatic generalized epilepsy, complex partial seizures.

The general condition is hard, muscle hypotension. The skin is pale with a marble pattern. Turgor and tissue elasticity are reduced. Thinning of the subcutaneous fat layer on the abdomen and extremities. The head is a hydrocephalic shape, a wide,

protruding forehead, deep-set eyes, a short nose with a thickening at the tip, a dysmorphic structure of the auricles, kyphoscoliotic posture. Heart sounds are muffled, rhythmic, systolic murmur at the apex. The abdomen is enlarged, the liver +3.5 -4.0 cm, the edge is tightly elastic, smooth. The spleen is +1.5 cm, below the costal arch, physiological settings are not broken.

**Results:** microhematurea (20-22 in sight), increased AsT (63,9), AlT(59), alkaline phosphatase (6500), LDG (615,8). A change in the JAG1-heterozygous variant with 3545A> T (GLU1182VAL) was revealed.

Shear fibroelastography: METAVIR F1-F2 scale - portal fibrosis with single septa (in 2 segments from 6 studies)

Final diagnosis: Alagil syndrome. Congenital CNS pathology: external-internal hydrocephalus, subcompensated delay in psychomotor development. Symptomatic epilepsy with tonic-clonic seizures. Nanism. Additional lobule of the spleen. Capillary hemangioma of the IV segment of the liver (fibrosis F1-2). Renal dysplasia (echoscopic). Isolated hematuria.

Specific therapy is not developed. The treatment is aimed at maintaining the functions of the affected organs and reducing the symptoms of the disease.

**Conclusion:** Diagnosis and treatment of Alagille syndrome requires a multidisciplinary approach, which allows improving disease prognosis.