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CLINICAL CASE

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ALPORT SYNDROME IN A 16 YEARS OLD CHILD

ABSTRACT

This article focuses on a rare disease -- Alport syndrome in children. Alport syndrome has a poor prognosis as it leads to the development of chronic renal failure. In addition to the renal symptoms in children with Alport syndrome is marked hearing loss; various eye disorders; delayed physical development; congenital anomalies (deformed ears, high palate, joined or extra fingers – no more 7 signs);rarely – Lam. The article presents a clinical observation of Alport syndrome in a child 16 years of age.

Keywords: hearing loss, hereditary disease, renal insufficiency, congenital anomalies, and poor prognosis.

Hereditary nephritis (the more famous the name, Alport syndrome) is a quite rare pathology. According to official data, in Russia per 100,000 newborn babies have 17 with this malformation. In Europe 1% of all patients with chronic renal failure (CRF) is the people with hereditary nephritis. The main and only reason why children are born with Alportsyndrome, is a genetic mutation. Damaged one of the extrarenal symptoms of congenital nephritis occur later: hearing loss (first child ceases to distinguish high-pitched sounds, then normal speech); various eye disorders;delayed physical development; congenital anomalies (deformed ears, high palate, joined or extra fingers – no more 7 signs);rarely – Lam (overgrowth of smooth muscle fibers) of the esophagus, trachea, bronchi [1].

The aim: to show the features of the Alport syndrome in a child 16 years of age.

Observations: the Patient, 16 years, was admitted in the Nephrology Department of the National medical center with complaints of recurrent dizziness, weakness, fatigue, hearing loss.

From the disease anamnesis: ill since birth. Is under medical supervision of a nephrologist since 2006, the audiologist since 2008 with a diagnosis of Hearing loss 1 degree. Pre-school age observed recurrent gross hematuria, with the years, increasing proteinuria. In 2007, he was treated with prednisolone for 6 months, efficacy was not. Since the end of 2008, a decrease of hearing and was diagnosed with Bilateral sensorineural hearing loss of 1 degree. In 2009, spring was examined in the Scientific center of children's health (our center) in Moscow, where he was diagnosed with Hereditary nephropathy (Alport syndrome). The last hospitalization was in November 2016 – the patient was taken urgently, in connection with significant deviation in the urine. Constantly gets Eralfon scheme, omeprazole, folic acid. Planning comes to control examination and treatment in Nephrology BUT PDTS «RB No. 1НЦМ».

From the anamnesis of life: Child from first pregnancy occurring in the 1st half with toxicosis in the second half of the AD. Delivery at 38 weeks, labor operational.The Apgar score 8/8. Body weight at birth 2880, height 50 cm, Cried at once, a loud cry. Rash, diaper rash was not. Neonatal jaundice appeared on day 3, stayed 6 days, moderately severe. To the breast applied on the first day, sucked actively. Umbilical remnant fell on the fifth day. Discharged home on the seventh day.Natural breastfeeding to 12 months.Input solid foods from 6 months.Psychomotor development at the age up to 1 year. The disease: SARS, acute respiratory infections, chickenpox, pneumonia, and recurrent bronchitis. Preventive all age. BCG in the maternity hospital 23.08.2000.Mantoux from 14.11.08 -7 mm, 22.10.10 – negative. Medical withdrawal in 2010. The heredity is not burdened, the mother of 34 years, has a chronic disease – pyelonephritis. Allergic anamnesis burdened. Food Allergy to citrus.

Objective status at survey: assessment of the condition of the child moderate, harmonious physical development. BMI =17.1, which is the norm. Skin and visible mucous membranes pale, dry. The hair is dry. On the left hand in the upper third of the forearm in the wrist and elbow there is aarteriovenous fistula for hemodialysis. Child low power.Subcutaneous fat layer mild. Peripheral lymph nodes were not expressed. In the lungs vesicular breathing, wheezing no. The abdomen is soft, painless. The liver and spleen are not enlarged.

The patient survey.

Survey results: ultrasound of the kidneys from 26.01.2017 Expressed diffuse changes of renal parenchyma with a decrease in age sizes. Seal of the renal sinuses.

General analysis of blood from 23.01.2017 HGB – hemoglobin-116r/L.

Biochemical analysis of blood from 25.01.17 g: serum Creatinine blood 377,2 μmol/l, iron levels, blood - 7.1 μmol/l, ferritin serum was 8.2 μg/l; the phosphorus level the blood to 1.5 μmol/L.

Blood for parathyroid hormone from 25.01.17 g – 195 PG/ml

KOS blood (from 25.01.17 g): ctHb – 177 g/l mmol/L. The General analysis of urine from 25.01.17 g : protein - 1.67 g/l; leukocytes 3-5 in p/Zr; erythrocytes changed entirely in the field of view; erythrocytes unchanged entirely in the field of view.Consultation consultation from 27.01.17 g: Diagnosis of Dry rhinitis.

Helik – test 27.01.17 g: Hp(++).

Conclusion: the result is positive

Fegds from 27.01.17 g: Conclusion: Duodenogastric reflux. Catarrhal distal esophagitis.Stagnant gastroduodenopathies. Chest x-ray in direct projection from 27.01.17 g: Conclusion: In the lungs without focal and infiltrative changes. ECG from 27.01.17 g: Conclusion: sinus rhythm with heart rate of 57 beats per minute, bradycardia. The vertical position of the EOS.

Consultations of specialists: Gastroenterologist from 27.01.17 g: Diagnosis: Primary: K21.0 Gastroesophageal reflux disease with esophagitis of the lower third of the esophagus. Collateral: K29 common chronic superficial gastroduodenitis, active stage associated with H. pylori.

The survey was delivered clinical diagnosis:

Primary: Hereditary nephritis. Alport Syndrome.

Related: Bilateral sensorineural hearing loss, 1 table K21.0 Gastroesophageal reflux disease with esophagitis of the lower third of the esophagus.K29 common chronic superficial gastroduodenitis, active stage associated with H. pylori.Dry rhinitis

Complication: Chronic renal failure, terminal stage.

During the hospital stay the treatment: Mode of ward, Table 7G, end-stage renal failure with hemodialysis, and extra food. Hemodialysis for 4 hours a day. Omeprazole 20 mg-1 capsule 2 times a day.Eralfon 2000 IU, 3 times a week. Alfacalcidol 0.5 μg - 2 tablets 1 time a day. Lineks 1 capsule 3 times a day. Control of blood pressure 2 times a day.

During the hospital stay, the dynamics of the patient's condition are positive, the efficacy of the treatment is stable.

Recommendations: the Child is sent to the Department of transplantation at the RCCH Moscow, for carrying out kidney transplantation (06.02.17 g). To continue training and treatment in hospital. Strictly follow a diet. The control of the KLA, OAM, blood pressure. Correction treatment when indicated.

The plan: re-sugar curve, re-consultation of a cardiologist and a gastroenterologist.

CONCLUSIONS

1. In the absence of specific treatment, the main goal becomes slowing development of kidney disease. Children are prohibited physical activity, assigned complete and balanced nutrition.

2. The use of hormones and cytotoxic drugs does not lead to significant improvement. The main treatment remains transplantation (transplantation) kidneys.

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