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A CLINICAL CASE OF PARTIAL RED CELL APLASIA IN A 2-YEAR-OLD CHILD

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Currently, no more than 300 patients with pure red cell aplasia have been described. A rare case of pure red cell aplasia in a Sakha child is presented in the article.

Keywords: anemia, erythrocytes, red cell aplasia, treatment, sequencing, Sakha, children, Yakutia.

Introduction. Pure red cell aplasia (PRCA) is a rare form of congenital hematopoiesis resulting from apoptosis of erythroid precursors in the bone marrow due to a defect in ribosome biosynthesis [1-4]. PRCA was first described by Kaznelson in 1922. Subsequently, a number of cases of this disease were described, with thymoma tumor of the thymus gland (thymoma) identified in a significant proportion of patients [2].

Currently, most of the genetically deciphered cases of PRCA are the result of haplotypic deficiency of genes encoding small or large ribosome subunit proteins; single cases of PRCA resulting from mutations of GATA1, FLVCR1, and TFR2 have also been identified. Congenital forms of PRCA with a debut at 2 years of age have been described. The course of the disease is chronic, and in some cases remission is achieved [1-3]. This article presents an interesting case of a Sakha patient with pure red cell aplasia (PRCA).

Clinical example. Child, name I., Sakha, 2 years old. Anamnesis of life - child from the 1st pregnancy, which proceeded in the first half with toxemia, in the 2nd half proceeded without features according to the mother's speech. Delivery 1, in term, 41 weeks, natural. Weight at birth is 3020 grams, height is 51 cm. Was breastfed on the first day. Apgar score 8/9b. Discharged home from the nursery on the fifth day. Breast-feeding up to the age of 1 month. Psychomotor development up to the age of 1 year of age corresponded: holds his head after 2 months, rolls over by 4 months, sits after 6 months, crawls after 7 months, walks after 10 months.

Mother's diseases are anemia, allergic diseases. Heredity, according to the mother, was not aggravated.

Preventive vaccinations according to the national vaccination calendar.

Past illnesses: frequent acute respiratory infections, COVID 19, bronchitis, bronchial asthma.

Allergic history: citrus, dust.

There were no operations carried out.

Past medical history: since January 2021, the child has been bothered by frequent acute respiratory infections, with a prolonged cough, pallor, lethargy and weakness. In February 2021, the child was referred to the central regional hospital for treatment. Complaints on admission: frequent acute respiratory infections and prolonged cough, pale skin, weakness, lethargy, cough. Discharged in March 2021 with improvement.

On 29.09.21, with complaints of prolonged cough, pale skin, weakness, lethargy, cough, the child was referred to the pulmonology department of RH #1-NCM.

According to the results of the examination: general analysis and biochemical analysis of blood contained no features. Tests for infections were taken: CMV DNA was positive. Blood PCR for CMV was negative, herpes simplex virus 1.2 was negative. Urine PCR for CMV was positive. Clinical diagnosis: Obstructive bronchitis. Lingering course. Combined etiology. CMV infection with bronchopulmonary involvement.

The child was treated with Viferon 150,000 IU 2 times a day, azithromycin 10 mg/kg/day (120mg) once a day for 5 days, hofitol 0.5 ml 3 times a day, linex 1 capsule 3 times a day, ursodeoxycholic acid 50 mg 2 times a day, valganciclovir 16 mg/kg 2 times a day. On 14.10.22 the child was discharged in satisfactory condition.

From 31.01.22 to 26.02.22, the child had a coronavirus infection. Since 17.02.22 his condition worsened: weakness, lethargy, pronounced pallor. On February 17, 2022, the child was hospitalized in the infectious diseases department of RH#1-NCM.

Paraclinically: General blood test of 16.02.22.: white blood cells (WBC) $11x10^9$/l (RI: 7.5 - 11x10 ^{9}/l), red blood cells (RBC) - 1.72 $x10^{12}$/l (RI: 4-4.9$x10^{12}$/l), hemoglobin (HGB) - 44 g/l (RI: 110-5=132x10 g/l), stabular neutrophils - 6% (RI: 1-5%), segmented neutrophils - 21% (RI: 35-55%), lymphocytes - 69% (RI: 35-55%), monocytes - 4% (RI:4-6%), eosinophils - 4%(RI: 0-5%), reticulocytes 0. 2% (RI: 0.4- 1.3%), COE 37 mm/hour (RI: 1-15 mm/hour). Conclusion: Hypochromic anemia of the 2nd-3rd degree.

Biochemical blood test of 2/16/22: Alanine aminotransferase (ALT) 43.43 U/L (RI:00-40.00 U/L), aspartate aminotransferase (AST) 39.4 U/L (RI: 00-40.0 U/L), blood glucose 4 mmol/L (RI: 3.3-5.60mmol/L), total protein 59.5 g/L (RI: 51.00-73. 00 g/l), albumin 46.5 g/l (RI: 35.0-50.0 g/l), creatinine 25.5 µmol/l (RI: 35.00-110. 00mmol/l), urea 6.8 mmol/l (RI: 3.3-5.8 mmol/l), serum iron 62.9 g/l (RI: 9.0- 30.4 g/l), total bilirubin 4 µmol/l (RI: 3.4- 17.10 µmol/l). Conclusion: elevated serum iron level.

On February 17, 2022 the child was urgently transferred to the oncohematological department of PC RH#1-NCM.

Myelogram from 18.02.22: granulocytic growth with delayed myelocyte stage with absence of erythroid growth indicators. The morphological picture is typical for pure red cell aplasia.

The patient was consulted with the Federal State Budgetary Institution "Research Institute of the Russian Academy of Medical Sciences named after Dmitri Rogachev", and an examination to clarify

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the diagnosis was recommended. Molecular genetic studies confirmed the presence of mutations in ribosomal genes (RPS19, RPS10, RPS24, RPS26, RPL5, RPL11, RPL35a, RPS7, RPS17), indicating pure red cell aplasia.

L-lycine was prescribed, hemotransfusion #1 - 30 ml.

On discharge - general blood test dated 30.12. 2021 - white blood cells (WBC) - 4.4*10^9/l (RI: 7.5 - 11x10^9/l), red blood cells (RBC) - 4.3x10^{12}/l (RI: 4-4.9x10^{12}/l), hemoglobin(HGB) - 120.2 g/l (RI: 110-132x10 g/l), stab neutrophils - 3% (RI: 1-5%), segmented neutrophils - 27% (RI: 35 - 55%), lymphocytes - 55%(RI: 35-55%), monocytes - 5% (RI:4-6%), reticulocytes 0.4 (RI: 0.4 - 1.3%), COE 3 mm/hr(RI: 1-15 mm/h). Conclusion: There was an improvement in the laboratory indexes.

Clinical diagnosis: Pure red cell aplasia. Concomitant diagnosis: Atopic dermatitis. Limited form. Subacute course. Congenital heart defect. Grade 2 mitral valve insufficiency. Left supplementary coronary artery. E66.0 Grade 1 paratrophy. Hypoplasia of the right kidney. Physiological phimosis.

The sick child was discharged from the hospital with recommendations: Prednisolone at a dose of 1.5 tablets per day, divided into 3 doses. Re-admission to the hospital in 1 month.

In the period from 23.05.22 to 7.06.22 he was twice hospitalized to the oncohematology department of PC RH №1-NCM on emergency indications due to worsening of his condition and changes in blood tests.

Examination on admission: Objective examination on admission: Height - 85 cm, weight - 16 kg, temperature - 36.1, heart rate - 24 per minute, heart rate up to 96 per minute. Condition was moderate in relation to the underlying disease. His well-being was not impaired. His appetite was not disturbed. Sleep was calm. Clear consciousness. The build was correct. Subcutaneous fatty tissue; distributed evenly. The pharynx was not hyperemic. The mucous membranes of the mouth and pharynx were clear, pale in color. Nasal breathing was free. The bones and joints system had no features. Peripheral lymphatic system: lymph nodes were not enlarged. The thorax was regular in shape. On percussion of the chest - clear pulmonary sound. On auscultation of the chest, vesicular breathing was heard, no rales. The heart tones were clear and rhythmic. The abdomen was soft and painless. The liver and spleen were not enlarged. Urination was free and painless. There were no peripheral edemas.

Paraclinically: general blood test of 14.06.22 - White blood cells (WBC) - 4.23x10^9/l (RI: 7.5 - 11x10^9/l), red blood cells (RBC) - 2.3*10^{12}/l (RI: 4-4.91012/l), hemoglobin(HGB)- 60.0 g/l (RI: 110-132x10 g/l), stab neutrophils - 1% (RI6 1-5%), segmented neutrophils - 22% (RI: 35-55%), lymphocytes - 54%(RI 35-55%), monocytes - 9% (RI:1-5%), reticulocytes 4 (RI: 4-6%), COE 3 mm/hr (RI: 1-15 mm/h). Conclusion Decreased hemoglobin, erythrocytes.

The child was clinically diagnosed with: Primary disease: Pure red cell aplasia.

Treatment was prescribed: Table #15, L-leucine 1000 mg/sq.m. - 540mg. Three times a day for 3 months, replacement therapy by transfusion of washed red blood cells.

Against the background of the therapy the condition and blood parameters improved. At present the child is receiving treatment at the place of residence, periodically comes to the oncohematology department of RH №1-NCM for replacement therapy with washed red blood cells.

Conclusion: Pure red cell aplasia (PRCA) is a rare syndrome characterized by a reduced number of erythroid cell precursors in the bone marrow. In pure red cell aplasia, a careful examination of the patient is necessary to exclude a neoplastic process, differential diagnosis with sideroblastic anemia and acute leukemia, transient erythroblastopenia, congenital hypoplastic anemia and Pierson syndrome. NGS sequencing and telomere length determination are used in the diagnosis. The main treatment for pure red cell aplasia is red cell transfusion and administration of L-leucine and glucocorticosteroids.

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