CLINICAL AND LABORATORY PROFILE OF CHILDREN WITH THALASSEMIA IN A NON-ENDEMIC REGION

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Background

Iron deficiency anemia (IDA) is the most common form of anemia encountered by pediatricians in outpatient practice [1,3]. Microcytosis of red blood cells is a typical but not pathognomonic finding in IDA; differential diagnosis of IDA with the second most common microcytic anemia – thalassemia – is difficult, especially for asymptomatic forms in heterozygous carriers [2,4,5]. In this regard, the study of clinical and laboratory criteria for the differential diagnosis of these diseases is relevant.

Objective

To determine the clinical and laboratory profile of children with thalassemia in a non-endemic region using the model of the Oryol region.

Materials and methods

In the database of the only specialized department of pediatric oncology and hematology Kruglaya Scientific-clinical multidisciplinary center for medical care for mothers and children in the Oryol region, for the period 2015-2022 a search was conducted for newly identified cases of thalassemia in children. For a retrospective analysis, medical records of patients with thalassemia witch diagnosis was verified by hemoglobin electrophoresis were used. Complaints, medical history, age, nationality, clinical and laboratory manifestations of the disease were subject to registration. Due to the rarity of the pathology, only descriptive statistics methods were used; quantitative values are presented in the format Me (A; B), where Me is the median, A and B are the 1st and 3rd quartiles, respectively.

Research results

Over 8 years in the Oryol region, thalassemia was diagnosed in 8 patients (five boys and three girls) aged from 2 to 17 years with a median of 5.5 years (quartile 1; 3 = 4.5; 8). All children were born from marriages where at least one of the parents or second-degree relatives was a native of Azerbaijan (62.5%, n=5), Tajikistan (25.0%, n=2) or Uzbekistan (12.5%, n=2) n=1), while only half of the patients indicated anemia in relatives (50.0%, n=4).

Complaints characteristic of anemic syndrome (weakness, pallor) were reported by only 1 patient/parent (12.5%); the remaining cases were detected by chance during a routine blood count assessment. The age of onset was 1.2 months (1.1; 3), clinical and laboratory manifestations included anemic syndrome

(12.5%, n=1), splenomegaly on ultrasound (37.5%, n=3), decreased hemoglobin (Hb) level, microcytosis and hypochromia of erythrocytes (100%, n=8). Manifestations of jaundice, symptoms of hemolytic crises, deformation of skeletal bones, and cholelithiasis were not recorded in any of the children. Mild anemia was diagnosed in 87.5% of patients (n=7), moderate anemia – in 12.5% (n=1); median Hb level was 105.0 g/l (102.8; 108.5), RBC 5.9 x 109/l (5.3; 6.2), MCV 57.5 fL (56.3; 58. 8), MCH 18.5 pg (17.0; 19.3). All children had normal serum ferritin (SF) levels. Based on the results of hemoglobin electrophoresis, the diagnosis of β-thalassemia was established in 87.5% of patients (n=7), one patient was a cocarrier of α- and β-thalassemia. Prior to initial referral to a hematologist, all patients in the pediatric area had received iron medications without success.

Conclusion

Cases of thalassemia in a non-endemic region are sporadic and asymptomatic; typical clinical and laboratory manifestations of hemolysis do not occur practically; the disease occurs under the guise of mild IDA. Peculiarities of the genealogical history, as well as normal or elevated levels of SF in children with microcytic anemia are an indication for referring children to a hematologist.

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