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DIAGNOSTICS OF HEREDITARY SPHEROCYTOSIS

Introduction

Hereditary spherocytosis also known as Congenital Spherocytosis (CS) is a hereditary hemolytic anemia that presents with spherically shaped red blood cells (RBCs) due to abnormal proteins of its membrane. RBCs cannot maintain their original shape which is biconcave in nature because of mutations in genes of the membrane proteins that help in its structural stability. Abnormal structure of the membrane proteins namely spectrin, ankyrin, band 3, and band 4.2, plays a huge role in develpment of CS. To explain further, the mutations of the listed genes encoding membrane proteins is considered the primary etiology. Pathogenesis: Abnormal proteins of erythrocytes' membranes lead to a defect in connection between the cytoskeleton of erythrocyte with its lipid bilayer which is on the surface. This defect can lead to a loss of RBC membrane stability and deformability with progressive membrane lipid bilayer loss in the microvasculature. Destruction of the membrane can lead to spherocytosis, a drop in mean corpuscular volume (MCV), an increase in mean corpuscular hemoglobin concentration (MCHC), and increased osmotic fragility of RBCs. CS clinical presentations depend on its severity and mutations. To determine the pathomorphology of CS, blood smears are viewed under a microscope which shows small sphere-shaped cells that are pale at the periphery and dense. Diagnostic methods typically include a complete blood count, reticulocyte count, and peripheral blood smear, which reveals spherocytes. To confirm the diagnosis, a binding test called EMA (eosin-5-maleimide) and an osmotic fragility test are done [1]. Epidemiologically, Hereditary spherocytosis is the most common genetic hemolytic disease found amongst those of Northern Europe and North America region. It is rarely seen among others. It is observed in 1 of 2000 individuals, with a significant number of asymptomatic cases being undiagnosed [2]. Complications of CS are namely: stones formation in gallbladder, enlarged spleen and risk of infection due to splenectomy [3].

Morbidity associated with CS can be significant, particularly due to complications like anemia and splenic dysfunction, while mortality is insignificantly low in treated cases [5]. Males and females are equally predisposed, but some studies suggest a slight male predominance [4].

The data analysis will discuss further on the gender predominance. Hereditary spherocytosis is more often seen in children below five years. Management of this condition includes use of folic acid supplement, supportive therapy and in severe conditions, a splenectomy is done to decrease the risk of hemolysis and anemia [6]. The general prognosis for patients suffering from hereditary spherocytosis is reasonably good but sometimes it varies depending on the disease severity.

Goal

To give a morphological characteristic of anemia in hereditary spherocytosis.

Material and methods of research

The medical histories and outpatient cards of 16 patients aged 1 month to 18 years with an established diagnosis of NS were analyzed. The study was conducted at the state institution "Republican Scientific and Practical Center for Radiation Medicine and Human Ecology" The diagnosis of HA was established based on the anamnesis data and laboratory parameters indicating signs of hemolysis: reticulocytosis(due to compensation for destroyed erythrocytes) in the peripheral blood, increased levels of free bilirubin and total lactate dehydrogenase (LDH), decreased haptoglobin in the biochemical blood test due to hemoglobin binding to it. At the first stage of diagnosis, we determined the severity of anemia, the regenerative capacity of the erythrocyte sprout, as well as the morphologicalcharacteristics of anemia based on the erythrocyte indices: mean corpuscular volume (MCV), mean corpuscular hemoglobin content (MCH), and mean corpuscular hemoglobin concentration (MCHC).

Statistical processing of digital data was carried out using the StatSoft Statistica 10.0 application software package. Analysis of variation series was carried out using a nonparametric method, presenting data as a median (Me), 25% and 75% quartiles.

The results of the research and their discussion

An age-specific analysis of the primary diagnosis of NS was conducted. Most often, the diagnosis of NS was established at the age of 1 to 5 years – 62.5% (n = 10). The average age of patients was 4 years and boys were predominated with a ratio of 3:1.

The peripheral blood analysis in the evaluation of congenital spherocytosis is as follows: the hemoglobin levels had a media value of 93.2g/l (88,3; 112). ranging from 75g/l (severe anemia) to 125g/l (mild anemia). However, there were 2 findings of severe anemia in 2 boys (75.5 and 78 g/l –12.5%), 5 with mild anemia (31.25%) and 9 cases with moderate anemia (56.25%).

The leukocyte median value was $8.6x10^9$/l ranging from $5.6x10^9$/l to $21.76x10^9$/l (leukocytosis). I recognized 3 cases of highly increased leukocytes in 3 boys (14.7, 18.9 and 21.7 –18.75%) and the rest within normal range (81.25%).

Erythrocyte median value was $3.64x10^{12}$/l ranging from $2.42x10^{12}$/l to $5.77x10^{12}$/l /l; 8 (50%) have low erythrocyte count showing possible anemia and the other 8 (50%) had normal range

Analysis of the red blood cells indices shows: Median mean corpuscular volume(MCV) was 75.4(72,9; 81,7) fl, ranging from 60–125 fl (norm is 80-94fl) thus 14 patients had microcytic (87.5%) and 2 patients have normocytic (12.5%). Another significant index to check is mean corpuscular hemoglobin(MCH) with the median of 25.9 (34,9; 37,5)pg; ranging from 21.7 to 32.3pg(norm is 27–33 pictograms); in this analysis 10(62.5%) out of these cases had below the normal range signifying microcytic anemia and the other 6(37.5%) were normocytic The MCHC had a median value of 354g/dL ranging from 278 to 387g/dL(norm is 320 to 360g/dL); 6(37.5%) out of 16 cases had a high MCHC showing hyperchromic anemia, 2(12.5%) showed hypochromic anemia while 8(50%) showed normochromic anemia. The hematocrit values

(ranging from 19.8% to 41.7% with a median value of 26.1%) vary in gender with the norm of females (37%–47%) and males (42%–52%); the 4 girls had low hematocrit values ranging from 23.2% to 30.7% and the 12 boys all had low hematocrit values ranging from 19.8% to 41.7%. Hemolytic anemias are classified as hyperregenerative, as evidenced by a high level of reticulocytes. In our observation, the median Ret (%) was – 7.8 (5.4; 12) %. ESR level had a median value 7mm/hr ranging from 2–13mm/hr which are all within normal range.

Conclusion

The obtained results allowed us to draw the following conclusions. Hereditary spherocytosis was most often diagnosed at the age of up to 5 years (62.5%). Boys were three times more likely to get sick. At the initial diagnosis of NS, anemia of mild severity was observed and was characterized as normocytic, normochromic and hyperregenerative. Congenital spherocytosis is a significant hereditary condition with distinct pathological features. Early diagnosis can lead to favorable outcomes and improved quality of life. Continued research is essential to better understand the genetic basis and long-term implications of this condition.

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