RED BLOOD CELL DISORDERS

PP 27

ROLE OF SERUM HEPCIDIN AND RETICULOCYTE HEMOGLOBIN CONCENTRATION IN EVALUATION OF ANEMIA IN ULCERATIVE COLITIS PATIENTS

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Objective: One of the most common extra-intestinal signs of ulcerative colitis (UC) disease is anemia, which has a considerable influence on patients' quality of life. **AIM:** The aim was to evaluate the role of serum hepcidin and reticulocyte hemoglobin concentration (CHr) in the study of anemia in UC patients. **Methodology:** We recruited 80 UC patients and 30 healthy individuals of matched age and sex as controls. Subjects were subdivided into three groups – Group I: 50 anemic UC patients, Group II: 30 nonanemic UC patients, and Group III: 30 healthy controls. **Results:** CHr showed a statistically highly significant decline in Group I than Groups II and III. Serum hepcidin showed a significant difference between Groups I, II, and III. Also, a significant negative correlation between CHr, serum hepcidin and severity of UC and a significant positive correlation between CHr and hemoglobin level, MCV, serum ferritin, and transferrin S. While serum hepcidin had a significant positive correlation with hemoglobin level, MCV, serum ferritin, transferrin S., and CHr. **Conclusion:** CHr had an excellent performance in prediction of iron-restricted anemia and was the test of best performance in prediction of iron-deficiency anemia ± ACD. Serum hepcidin had an excellent performance in prediction of ACD.

https://doi.org/10.1016/j.htct.2022.09.1262

IMMUNODEFICIENCIES / NEUTROPHIL DISEASES

PP 28

GRISCELLI SYNDROME TYPE 2 –CLINICAL APPROACH AND CASE REPORT

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Objective: Griscelli syndrome type 2 is rare authosomal ressesive disorder caused by a defect in the RAB27A gene, which affects a melanosome-anchoring complex in melanocytes, affecting release of cytolytic granules from T and NK cells. Children with GS type 2 develop an uncontrolled T-lymphocyte and macrophage activation syndrome known as hemophagocytic syndrome (HS) or hemophagocytic lymphohistiocytosis (HLH). We describe a 3 years old girl patient classic features of Griscelli syndrome type 2 **Case report:** A 3-year-old girl was admitted to hematology with complaints of LAP, hepatosplenomegaly and pancytopenia (WBC- 3080/μl, Hb-7.6 g/dl, Neutr.-350/μl, PLT - 165000/μl). The patient's condition was below the percentile, her skin was bronze, her hair was silver-grey. HLH criteria were met (triglycerides 458 mg/mL, ferritin 3445 ng/mL, fibrinogen 180 mg/dl). Morphology of the bone marrow was hypocellular, signs of dyserythropoiesis (stage I) and megakaryocytes were reduced **Methodology:** According to the clinical and laboratory data (hepatosplenomegaly, increased ferritin, hypertriglyceridemia, pancytopenia, hyperthermia resistant to antimicrobial therapy, silver-gray hair, pigment balls of hair seen light microscope) and the death of another undiagnosed child in the family, suggested likely primary HLH and GS. As a result of genetic analysis (homozygous mutation c.514_ 518del-CAAGC(p.GLN172Asnfs*,rs767481076)1 in the RAB27A gene), the diagnosis of GS type 2 was confirmed. **Results:** The patient was treated according to the HLH 2004 protocol. CSA levels were measured once a week. IVIG support was given based on IgG levels. HSCT was planned from patient's healthy HLA-matched sibling, but HSCT was delayed because the brother was infant. After 45 weeks of maintenance therapy, etoposide was discontinued, dose of dexamethasone was reduced to 5 mg/kg, but CSA was continued at the same dose. Control studies are carried out once a week. As far as possible HSCT is planning **Conclusion:** The prognosis of patients with Griscelli syndrome is poor. It is usually rapidly fatal within 1-4 years without aggressive treatment and bone marrow transplantation at onset of an accelerated phase. HSCT is more successful when implemented early course of the disease. Palliative care includes treatment and prophylaxis care infections and immunosupressor therapy in accelerated phases. Some patients have died after transplantation, but others have had lasting remissions

https://doi.org/10.1016/j.htct.2022.09.1263

LEUKEMIA

PP 29

THE COURSE OF TOXIC HEPATITIS IN LEUKEMIC PATIENTS AT THE STAGE OF SUPPORT THERAPY.

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Objective: One of the main tasks in the treatment of acute leukemia is to prevent the development of complications of chemotherapy, as well as the timely choice of the correct tactics for the treatment of complications. Because forced breaks associated with complications negatively affect the end result

of leukemia treatment. Practice shows that one of the organs affected by chemotherapy is the liver, and its damage directly depends on the toxicity and duration of chemotherapy. Our task was to conduct research work in this area, and to study toxic liver lesions in patients with leukemia. Before that, we carried out similar work at the stages of induction and consolidation of treatment of acute leukemia in children. And this period of research work is devoted to the supportive stage of therapy in patients. Objective: To study the frequency of toxic liver damage in children with acute leukemia during support therapy, to choose treatment tactics according to the severity of toxic hepatitis. **Methodology:** The study group included 51 children with primary acute lymphoblastic leukemia who completed the induction stage with complete remission and retained this result for the entire period of consolidation. The age of the patients ranged from 2.5 years to 15 years. Of these, there were 28 boys and 23 girls. The patients were from Baku and the regions of the republic. Treatment of acute lymphoblastic leukemia was carried out according to two branches of the Moscow – Berlin – 2015 program: B and T İmRG. The protocols of maintenance therapy of these branches do not differ, and both begin with the 31st week of the general program, end on the 104th. Each protocol consists of 8 stages of a combination of chemotherapy drugs Metotreksat + 6-Mercaptopurine, which last for 6 weeks and alternate with two-week courses of reinduction - Deksametazon + Vinkristin. Before the start of maintenance therapy, all patients with leukemia confirmed the preservation of the previously achieved remission, and the functional and organic state of the liver. With positive results, the continuation of leukemia treatment began. And when the symptoms of toxic hepatitis were detected, the severity was determined. According to this indicator, 3 forms of flow were issued: light, medium-heavy and heavy forms. The tactics of conducting therapy of each form were chosen by us. **Results:** Of 51 patients, 42 had toxic hepatitis (82%). It was mild in 12 patients (23.5%), moderate in 26 children (50.9%), and severe in 4 children (7.8%). In the mild form of hepatitis, patients were prescribed intravenous administration of Riboksin + Aevit (orally) for 10-14 days, or alternatively, per os Ursobil + Aevit. This combination made it possible to restore all clinical and laboratory parameters in patients within 14-21 days, and at the same time, without interrupting chemotherapy. Moderate and severe forms of hepatitis occurred mainly during the period of reinduction (54.7%). The administration of intravenous adeomethionine (Heptral) in the form of monotherapy for 8-12 days allowed continuous reinduction courses. Following him, the administration of an oral combination of Ursobil + Lipoic acid + Aevit for 14-21 days allowed to preserve the long-term effect. In severe hepatitis, chemotherapy was suspended, and patients were prescribed intravenous adeomethionine (Heptral) in combination with oral Ursobil + Aevit for 10-14 days, and along with this detoxification therapy was carried out in parallel. Such treatment gave an improvement in clinical and laboratory parameters. Subsequently, intensive therapy was suspended, and chemotherapy was started accompanied by Ursobil + Aevit + Lipoic acid for the next two weeks. This choice of therapy allowed us to preserve the restored indicators for a long time. There were no deaths or severe complications from toxic hepatitis in any case.

https://doi.org/10.1016/j.htct.2022.09.1264

HEMOGLOBINOPATHIES (SICKLE CELL DISEASE, THALASSEMIA ETC...)

PP 30

RARE UNSTABLE ALPHA GLOBIN VARIANT HB TAYBE (HBA1:C.118_120DELACC) WITH HBA2 POLY A MUTATIONS, CAUSES TO HEMOLYTIC ANEMIA IN TWO CASES FROM AZERBAIJAN

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Objective: Sequence variants are usually silent and rarer in α-globin, some may lead to an unstable protein with hemolytic or thalassemic phenotype. The most common *HBA* variant is ConstantSpring which is leading to an unstable elongated protein chain. Others may be due to ins/del in the α-globin, one of them Hb-Taybe caused deletion of Thr residue at codon 40 of the *HBA1*. We report, for the first time, 2 cases with hemolytic anemia due to the presence of Hb-Taybe in *trans* with *HBA2* poly-A mutations. **Methodology:** Patients were managed in the Thalassemia Unit of the National Hematology Center. Detailed pedigrees are drawn, medical recordings are reviewed and peripheral blood samples are collected. Sanger sequencing was perfomed with *in house* designed primers (*HBA1*, NM_000558.5 and *HBA2*, NM_000517.6) on genome analyser (ABI3500). Deletion and duplication analysis is performed by MLPA. **Results:** P-1: A 4-yr-old male who was diagnosed at the age of 1 yr with congenital hemolytic anemia. He presented with jaundice, the blood film showed moderate hypochromia and anisopoikilocytosis. He was transfusion dependent. P-2: A 45-yr-old female who was diagnosed at the age of 5 yr with congenital hemolytic anemia. She underwent splenectomy at the age of 32 yr due to moderate anemia. After splenectomy she was not transfusion dependent. Relevant clinical and laboratory data is presented at table. **Conclusion:** The clinical presentation is variable from mild hemolytic anemia to regular transfusion requirement. One patient had to have splenectomy in order to ameliorate the transfusion requirement. This study supports the requirement of α-globin gene analyses, and a careful evaluation of cases with hemolytic anemia, particularly in populations where thalassemias are endemic, in order to avoid missing any of the rare globin variants and to offer accurate genetic counseling.