testing confirms the diagnosis at an early stage and reduces morbidity and mortality due to the disease. WES is helpful to detect such cases.

Conclusion: Various genes such as DKC1, CTC1, RTEL1, TERF1, TINF2, TERC have been found to be responsible for DKC. RTEL1 is a DNA helicase necessary for telomere replication and stability. With the understanding of the molecular basis of the disease, patients with hematological findings at the time of diagnosis and those without skin findings were also identified. In our case, signs of bone marrow failure were observed primarily and no changes in nail dystrophy, leukoplakia and skin pigmentation and neurological findings were detected. In cases where the disease does not follow classical presentation, the use of genetic testing confirms the diagnosis at an early stage and reduces morbidity and mortality due to the disease. WES is helpful to detect such cases.

https://doi.org/10.1016/j.htct.2020.09.122

PP 60

Acquired aplastic anemia in childhood: single-center experience

M. Bilici[1,*], R. Tuna[1], S. Sahin[1], Z. Karakas[1], A. Unuvar[1], S. Anak[2], D. Tugcu[1], S. Karaman[1]

[1] *Istanbul Medical Faculty Pediatric Hematology-Oncology, İstanbul, Turkey*
[2] *Medipol University Hospital Pediatric Hematology-Oncology, İstanbul, Turkey*

Objective: Acquired aplastic anemia is a rare disease characterized by the irreversible loss of bone marrow function and threatens life when not treated. Bone marrow transplantation (BMT) or immunosuppressive agents (IST) are used in its treatment. In this article, we aimed to evaluate our patients with acquired aplastic anemia in epidemiological, etiological, and treatment outcomes.

Case report: Nine patients who were diagnosed with acquired aplastic anemia over ten years were evaluated.

Methodology: The patients admitted to the Istanbul Medical Faculty Pediatric Hematology-Oncology Outpatient Clinic between 2000 and 2010 were diagnosed with acquired aplastic anemia (those who underwent BMT, IST, or both) were evaluated retrospectively on patient files and computer records.

Results: Nine patients were diagnosed with acquired aplastic anemia over ten years. 4 of them were girls, and 5 were boys. The average age was 10 (1–17 years). There was a history of hepatitis in 3 cases and a history of metamizole use in 1 case. As a treatment, six patients were treated with IST, and five patients were treated with BMT. ATG 40 mg/kg/day 4 days, cyclosporin 10 mg/kg/day (6 months), methylprednisolone 2 mg/kg/day (2 months) and G-CSF 5 μg/kg (2 months) as immunosuppressive therapy. Response to immunosuppressive therapy was received at an average of 3 months. Two of them were fully responsive. One patient was lost due to septic shock before the IST response was evaluated. BMT was performed in 5 cases, three of them were unresponsive to IST. In the follow-up, two cases are in remission, and three are lost due to sepsis. When evaluating our 5 cases with dead, two of them were very severe aplastic anemia, the symptoms of sepsis were present in their first admission, and they died before the treatment started. Two of them died due to the complication of BMT in the very early period. One case was admitted with perforated appendicitis while in remission after BMT and died due to septic shock.

Conclusion: Two primary treatment modalities are used to treat patients with severe aplastic anemia; IST and BMT. The first option is BMT with the matched sibling donor. If there is no suitable sibling, IST is started first, and a fully compatible donor is searched immediately. If there is no response to IST, an allogenic BMT must be applied in the presence of a suitable donor. Our mortality rate is high compared to the literature because of severe disease presentation; most of them were late admission to the hospital due to low socioeconomic level.

https://doi.org/10.1016/j.htct.2020.09.123

HEMOSTASIS, THROMBOSIS, AND VASCULAR BIOLOGY

PP 61

The course of intracranial bleeding in the patient with immune thrombocytopenia

M. Babayev*, A. Ahmadova, K. Mehtiyeva, N. Babayeva

National Hematology and Transfusiology Center, Moscow, Russian Federation

Objective: The approach to the treatment of immune thrombocytopenia always remains relevant, despite the fact that the etiology and pathogenesis of the disease is quite clear, and it is clear that the development of the disease is based on the conflict between their own platelets and autoantibodies directed at them. The goal of treatment is to resist the creation of autoantibodies, protect your own platelets and lengthen their life. The proposed standards of treatment with steroids, reticuloendothelial system blockers, anti-lymphocytic antibodies, thrombopoietin, etc.did not find a clear place for a radical change in the course of the disease.

Case report: The article presents a case of a child suffering from chronic ITP who received various medical treatments with periodic remissions for 6 years. At the age of 10, the child had convulsions and neurological disorders due to acute respiratory infection and high temperature. In blood tests: PLT-10×10^9/L. CT scan of the brain showed the presence of intracranial bleeding. The prescribed “pulse therapy” with dexamethasone and platelet transfusions allowed for intracranial surgery (PLT – 234×10^9/L).). However, a few days later, due to the ineffectiveness of “pulse therapy”, and the risk of renewed bleeding, the patient was again transfused platelet mass and prescribed high-dose intravenous immunoglobulin (IVIG), which raised the platelet level to 210×10^9/L. Soon, this therapy was ineffective, and we had to re-transfuse the platelet mass and simultaneously prescribe thrombopoietin (Revoleyd). Against the background of this therapy, the platelet level was stabilized, and the resulting effect was long-lasting.