

organ failure. Case 5 is a 9-year-old male with ALL had a bloodstream and port catheter infection after the first induction therapy. *Herbaspirillum huttiense* was detected in the blood culture taken from the port catheter. The patient was successfully treated with meropenem without port removal. Case 6 is a 10-year-old girl with ALL had a bloodstream and port catheter infection during the second induction therapy. *Ralstonia pickettii* was detected in the blood culture taken from the port catheter. The catheter was removed and the patient was successfully treated with piperacillin-tazobactam. Case 7 is a 7 month old male with Juvenile myelomonocytic leukemia had a bloodstream and port catheter infection in the neutropenic period. The patient was constantly inserting the port catheter into her mouth. *Staphylococcus salivarius* was detected in the blood culture taken from the port catheter. Then, 5 day after, *Rothia mucilaginosa* was detected in the peripheral blood culture. The patient was successfully treated with meropenem without port removal. Case 8 is a 9-year-old girl with ALL had a infective endocarditis and sepsis during the induction therapy. *Magnusiomyces capitatus* was detected in the peripheral blood culture. The patient was treated with fluconazole and amphotericin-B, but she died of multi-organ failure.

Conclusion: Many different microorganisms can cause infections in immune-compromised children as a result of primary disease or chemotherapy. Though empiric antibiotic therapy should be initiated early, the treatment should be revised according to the antibiogram and catheter should be removed as needed.

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PP 58

Idiopathic hypereosinophilic syndrome associated pulmonary hypertension in a child

Y. Akcabelen*, T. Bayhan, G. Cinel, I. Ece, N. Ozbek

Ministry of Health Ankara City Hospital, Ankara, Turkey

Objective: Hypereosinophilic syndrome (HES) is defined by showing eosinophilic infiltration in any tissue or organ and increased eosinophils in peripheral blood. Other pathologies that cause eosinophil increase must be excluded. Pulmonary eosinophilic infiltration may have different symptoms and signs, but clinical presentation as PHT has not been shown in children.

Case report: A 6-month-old girl presented with dyspnea and hypoxia. A blood cell count and a morphological evaluation of a peripheral blood smear and confirmed hypereosinophilia (white blood cells 40,600/ μ L, eosinophils 18,900/ μ L, hemoglobin 10.3 g/dL, and platelets 425,000/ μ L). There was not any cellular morphological abnormalities in bone marrow aspiration examination. Pneumonia and parasites, allergic diseases, clonal abnormalities, cancer and vasculitis that might have caused HES were excluded. Echocardiogram showed 38 mmHg for pulmonary arterial pressure (PAP), suggesting pulmonary hypertension (PHT). After exclusion of other causes such as vasculitis, connective tissue

diseases, bronchopulmonary dysplasia, congenital heart diseases, lung diseases, and chronic thromboembolic PHT. The patient was diagnosed with pulmonary arterial hypertension associated with idiopathic HES. Methylprednisolone treatment was started at 2 mg/kg/day. PHT and HES were both improved in the evaluation one month later.

Conclusion: Eosinophilic infiltration causes thickening and remodeling of the pulmonary artery intima and media, thereby causing pulmonary hypertension. Thus, PHT can be seen as HES clinical presentation. With corticosteroid therapy, HES and PHT clinical findings can be controlled.

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PP 59

A rare variant of dyskeratosis congenita: RTEL1 defect

C. Coskun^{1,*}, S. Unal², N. Akarsu²

¹ Hacettepe University, Department of Pediatric Hematology, Ankara, Turkey

² Hacettepe University, Research Center for Fanconi Anemia and Other Inherited Bone Marrow Failure Syndromes, Ankara, Turkey

Objective: Dyskeratosis congenita (DC) is a rare hereditary disorder characterized by bone marrow failure, malignancy predisposition and skin findings. As the disease progresses, patients may develop pulmonary fibrosis, esophageal stenosis, urethral stenosis and liver cirrhosis. Herein, we present a patient who was referred with a diagnosis Diamond Blackfan anemia and was diagnosed to have dyskeratosis congenita on whole exome sequencing (WES).

Case report: A 18 month-old girl who was initially transfused at the age of three-months old and was on mostly transfusion programme, was referred to our center for molecular work-up with a diagnosis of DBA. There was second degree consanguinity between parents. On physical examination, body weight: 8.7 kg (5th percentile) height: 44 cm (<3rd percentile) was measured. Cubitus valgus was seen with camptodactyly. Liver and spleen were not palpable. Complete blood count showed hemoglobin (Hb) 7.9 g/dL, mean corpuscular volume (MCV) 104.1 fl, white blood count 6.9×10^9 /L, absolute neutrophil count 1.3×10^9 /L, platelet count 682×10^9 /L, reticulocytes 2% and peripheral smear showed hypochromia and macrocytosis in erythrocytes. Biochemical parameters, globin electrophoresis, vitamin B12 and folic acid levels were normal. Parvovirus B19 was negative. ADA2 enzyme level was determined as 24 U/L (5–20 U/L). Steroid was started at the age of 18 month-old with a clinical suspicion of DBA. She became transfusion independent after steroid initiation. WES analysis for DBA for the patient revealed RTEL1 gene mutation (c.1368G>T p.1trp456Cys). This mutation was found compatible with DC and no other mutations in DBA related genes were detected, including CNV analyses for large deletions. Steroid was ceased gradually and she did not require further transfusions after complete cessation.

Results: In dyskeratosis congenita 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.

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.

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PP 60

Acquired aplastic anemia in childhood: single-center experience

M. Bilici^{1,*}, R. Tuna¹, S. Sahin¹, Z. Karakas¹, A. Unuvar¹, S. Anak², D. Tugcu¹, S. Karaman¹

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.

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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- $10 \times 10^9/L$. 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 – $234 \times 10^9/L$). 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 $210 \times 10^9/L$. 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.