

of a 13.5 years-old female born to Turkish consanguineous parents with a novel missense mutation occurring outside the DNAJ domain of the DNAJC21 gene. Whole-exome and Sanger sequencing confirmation revealed a homozygous missense mutation in DNAJC21 gene c.463T>C, p.W155R which was considered as pathogenic in in silico analyses. Initially, this patient's vague and atypical symptoms led to uncertainty of the underlying diagnosis. Upon confirmation of the genetic mutation, a number of functional studies such as diepoxibutane test, proliferation test from peripheral blood mononuclear cells, and cytokinesis-block micronucleus cytome assay performed with the patient cells confirmed the likely diagnosis of an SDS-like syndrome attributable to DNAJC21 dysfunction. Through the analysis of this rare case, we illuminate the pleiotropic features of this unique bone marrow failure syndrome and emphasize the paramount role of genomic testing to discriminate a range of closely related bone marrow failure disorders.

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STEM CELL TRANSPLANTATION

PP 77

THE ROLE OF THERAPEUTIC DRUG MONITORING OF INTRAVENOUS BUSULFAN FOR PREVENTION OF SINUSOIDAL OBSTRUCTION SYNDROME IN CHILDREN

Dilek GURLEK GOKCEBAY¹,
Özlem Arman Bilir¹, Seda Şahin²,
İkbal Ok Bozkaya¹, Namık Yaşar Özbek¹

¹ University of Health Sciences Ankara City Hospital
Department of Pediatric Hematology Oncology

² University of Health Sciences Ankara City Hospital
Department of Pediatrics

Objective: Busulfan is a widely used alkylating drug for conditioning of hematopoietic stem cell transplantation (HSCT). Higher exposure of Bu is associated with toxicity and (sinusoidal obstruction syndrome) SOS, whereas lower exposure is associated with graft failure or relapse risk. Therapeutic drug monitoring (TDM) has been recommended to overcome these issues. We aimed in this study to compare HSCT outcomes in children with and without TDM of Bu. **Methodology:** This retrospective study conducted at our Transplantation Unit between 2012 and 2021. Patients aged 0-18 y underwent HSCT who received Bu-based conditioning and completed post-transplant +100 days included in the study. Data were collected including demographic information, primary diagnoses, conditioning regimen, graft-related data, dose of Bu, time to neutrophil and platelet engraftment, presence of SOS, acute or chronic GvHD, and clinical outcomes. SPSS 18.0 was used for statistical analysis. **Results:** 172 patients (59 girls, 113 boys) with a median age of 4.70 years (IQR 2.41-10.01) were enrolled in the study. TDM of Bu was performed in 126 patients. 32 patients (19%) developed moderate or severe SOS. Incidence of SOS was significantly higher in the group without TDM. A multivariable analysis showed that presence of acute GVHD and 2 or more alkylating agents in conditioning

regimen were associated with SOS. HSCT related outcomes, relapse, OS and EFS did not differ between two groups. **Conclusion:** To improve treatment outcomes of Bu, TDM and dose adjustment, following the first dose, has highly recommended regardless of the dosing guideline was used. We also demonstrated the incidence of SOS decreased in patients with TDM, but other HSCT related outcomes were not influenced. Optimal cumulative Bu exposure can balance between efficacy and toxicity of HSCT in children.

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PP 78

A CASE OF POLYCYTHEMIA DIAGNOSED AS HEMOGLOBIN ANDREW-MINNEAPOLIS

Mustafa Bilici¹, Serap Karaman¹,
Aysegül Unuvar¹, Deniz Tugcu¹,
Gülsah Tanyildiz¹, Ayca Dilruba Aslanger²,
Oya Uyguner², Rumeysa Tuna Deveci¹,
Sifa Sahin¹, Zeynep Karakas¹

¹ Department of Pediatric Hematology and
Oncology, Istanbul University, Istanbul, Turkey

² Department of Genetics, Istanbul University,
Istanbul, Turkey

Objective: Polycythemia is a rare condition in which an increase in erythrocyte mass is observed. It can be primary or secondary. Primary polycythemia occurs as a result of congenital or acquired mutations that regulate erythroid development. Although secondary polycythemia is mostly seen secondary to hypoxia due to cardiac/pulmonary reasons, it also develops as a result of congenital mutations. Globin gene mutations that increase the affinity of hemoglobin for oxygen are one of these rare causes. **Materials and Methods:** We present a male case who was referred to us for polycythemia. **Results:** A 15-year-old male patient with no known disease was referred to us after his school screening revealed high hemoglobin (18 g/dL). In complete blood count, other series were normal (wbc $5.8 \times 10^3/\mu\text{L}$ neu $3.3 \times 10^3/\mu\text{L}$ plt $174 \times 10^3/\mu\text{L}$), bilirubins and liver functions were within normal limits. On physical examination, conjunctiva and hands were plethoric, there was no hepatosplenomegaly, intermittent headaches were present, and neurological examination was normal. The patient was examined for the etiology of polycythemia. Hyperchromic erythrocytes were found in peripheral smear, no signs of hemolysis were observed. EPO level (8 mIU/ml) was in the normal range and JAK2 (V617F) mutation was negative. The patient's cardiac and pulmonary functions were within normal limits. Hemoglobin electrophoresis was sent from the patient. HbA was determined as 59.2, HbA2 2.8, Variant Hb 38. c.435G>T mutation was detected in the HBB genetic analysis, and this was considered to be compatible with Hemoglobin Andrew-Minneapolis. It was learned that the patient's mother and her cousins had similar findings, and some of them had undergone phlebotomy. Phlebotomy was planned in the presence of the patient's hemoglobin value > 18 g/dL and clinical findings. Phlebotomy was performed 3 times, aspirin was not started because there was no

history of thromboembolism. In our 1-year follow-up, the hemoglobin value was 17-17.5 g/dL. **Conclusion:** More than a hundred globin gene mutations associated with erythrocytosis have been described. Hemoglobin Andrew-Minneapolis mutation is one of them. Hemoglobin's affinity for oxygen has increased and EPO level is normal/increased. Due to the low number of cases, treatment recommendations were prepared based on polycythemia vera guidelines. Patients should be closely monitored in terms of hyperviscosity and thromboembolism, aspirin prophylaxis and phlebotomy are recommended according to symptoms. While investigating the etiology of polycythemia, hemoglobin electrophoresis is necessary, although it is very rare.

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LYMPHOMAS

PP 79

THE SMALLEST PRIMARY BONE LYMPHOMA

Fatma Burçin KURTİPEK, Volkan KÖSE, Seda ŞAHİN, Derya ÖZYÖRÜK, Neriman SARI, Sonay İNCESÖY ÖZDEMİR, Arzu YAZAL ERDEM, Meriç KAYMAK, İnci İLHAN ERGÜRHAN

Ankara City Hospital

Case report: Primary lymphoma of bone (PLB) is a rare malignant condition with lymphocytic infiltration of the bone; it accounts for 2–3% of all primary bone tumours in adults and children. Here we report a little girl with isolated PLB of B cell lineage focussing on diagnosis, evaluation and treatment strategy. Our case can help to get acquaintance with PBL, it should be taken into consideration as a different diagnosis for osteolytic lesions of bone.

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PP 80

MRI FINDINGS OF BONE MARROW AT THE BEGINNING OF LEUKEMIA

Seda Şahin, Neriman Sarı, Tülin Demirkan

Ankara Şehir Hastanesi

Case report: Pediatric ALL/lymphoma (LBL) is a clonal hematopoietic stem cell disorder which's highly aggressive. There is an overlap between ALL and LBL which shouldn't cause delay in the diagnosis of each other. We'll describe a patient who presented with leukemia symptoms such as fever, bone pain, who didn't have obvious atypical cells in his peripheral smear, BM aspiration and involvement in scintigraphy but had diffuse bone marrow (BM) involvement in the lower extremities in his MRI. BM biopsy showed ALL/LBL.

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PP 81

THREE CASES WITH BURKITT LYMPHOMA PRESENTING WITH CHOLESTASIS

Fatma Tuba YILDIRIM, Derya ÖZYÖRÜK, Arzu YAZAL ERDEM, Selma ÇAKMAKCI, Neriman SARI, Sonay İNCESÖY, İnci İLHAN

Ankara City Hospital

Case report: Cholestasis secondary to neoplasm is rare in children. It is also rare in Burkitt lymphoma and may be cause to treatment delay. We report 3 cases diagnosed with Burkitt lymphoma with cholestasis. All patients had jaundice and high direct bilirubin levels. They were given LMB chemotherapy protocol. After COP chemotherapy, cholestasis disappeared rapidly in all patients. In conclusion, cholestasis at initial resolves rapidly with chemotherapy despite high liver function tests in Burkitt lymphoma.

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BRAIN TUMOURS

PP 82

HIGH GRADE GLIOMA OF CENTRAL NERVOUS SYSTEM: SINGLE CENTER TREATMENT EXPERIENCE

Nur OLGUN¹, Deniz KIZMAZOĞLU¹, Batuhan OZDOĞAR², Dilek INCE¹, Emre CECEN¹, Ceren KIZMAZOĞLU³, Şefika AKYOL¹, Ayşe DEMİRAL⁴, Rıza CETİNGÖZ⁴, Ayşe ONAL¹, Handan UCAR⁵, Erdener OZER⁶

¹ Dokuz Eylül University Institute of Oncology, Pediatric Oncology

² Dokuz Eylül University Faculty of Medicine, Department of Pediatrics

³ Dokuz Eylül University Faculty of Medicine, Department of Neurosurgery

⁴ Dokuz Eylül University Faculty of Medicine, Department of Radiation Oncology

⁵ Dokuz Eylül University Faculty of Medicine, Department of Radiodiagnostic

⁶ Dokuz Eylül University Faculty of Medicine, Department of Pathology

Objective: To evaluate characteristics and treatment responses of patients with high grade gliomas (HGG) in our center. Medical files of patients with malignant CNS tumors between 1987-2020 were analyzed retrospectively. There were 44 patients with HGG. **Case report:** Diagnosis of patients as follows: 21 pons glioma, 2 anaplastic astrocytoma, 11 anaplastic ependimoma, 7 glioblastoma multiforme, 1 glioblastoma, 2 gliomatosis cerebri. The median age at diagnosis was 6,5 yrs (7 – 17 yrs), M/F:25/19. Age distribution: <5 yrs 12 patients, 5-10 yrs 18 patients, 10-18 yrs 14 patients. The most frequent complaints for pons gliomas: cranial nerve paralysis