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

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

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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 were SOS. HSCT related outcomes, relapse, OS and EFS did not different 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 canbalance between efficacy and toxicity of HSCT in children.

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A CASE OF POLYCYTHEMIA DIAGNOSED AS HEMOGLOBIN ANDREW-MINNEAPOLIS

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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 $5.8 \times 10^3/\mu L$ neu $3.3 \times 10^3/\mu L$ plt $174 \times 10^3/\mu L$), bilirubins and liver functions were within normal limits. On physical examination, conjunctiva and hands were pletoric, 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