as a third choice TKI. However, due to the deep neutropenia of the patient, it was possible to continue with a dose of 10 mg/m^2 from the 2nd week. With this dose, the neutrophil is around 0.8-1 $\times$ 10^3/μL. Under ponatinib treatment, BCR-ABL copy number was 6.6% IS at 1 month, 0.8% IS at 3 months, 0.09% at 5 months, and 0.05% at 6 months. No significant side effects were observed except neutropenia. **Conclusion:** There is no approved treatment in pediatric CML cases where the second choice TKI fails and there is no donor for transplantation. FDA approval for ponatinib in adult patients was obtained in December 2020. Ponatinib is a natural or mutant pan-BCR-ABL mutation inhibitor. It also inhibits VEGFR, FGFR,PDGFR,EPH and SRC kinases as well as KIT,RET,TIE2 and FLT3. The use of ponatinib should be evaluated by monitoring side effects/tolerance in pediatric cases where there is no other treatment option, and there is a need for studies on this subject.

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INHERITED BONE MARROW FAILURE DISEASES

PP 75

INVESTIGATION OF SALIVARY miR-9, miR-34a ve miR-196a LEVELS IN FANCONI ANEMIA AND ORAL SQUAMOUS CELL CARCINOMA PATIENTS

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Objective: Fanconi anemia (FA) is a rare bone marrow deficiency syndrome due to the DNA repair gene mutations, and Oral Squamous Cell Carcinoma (OSCC) is seen more frequently in FA patients than in the general population. The dysregulation of PI3K and Wnt signaling has been implicated in OSCC pathogenesis and abnormal expressions of miRNAs (a class of non-coding small regulatory RNAs) associated with these signaling pathways has been reported in OSHK patients. Salivary miRNAs are valuable biomarker candidates for OSCC development and prognosis. In this study, salivary levels of miR-9, miR-34a and miR-196a miRNAs related to PI3K and Wnt signaling pathways were examined in OSCC and FA patients and compared with the healthy control group. **Methodology:** Saliva samples were collected from 89 subjects including 25 OSCC patients, 24 FA patients and 40 healthy controls. Total RNA was isolated using Quick-RNA Miniprep Kit (Zymo Research) due to the kit instructions. cDNA was generated with miRCURY LNA miRNA PCR Assay (Qiagen, Hilden, Germany) and Quantitative real-time PCR was performed with miRCURY LNA SYBR Green PCR Kit (Qiagen, Hilden, Germany) according to the manufacturer's protocol. For the normalization of the expression levels of each miRNA, the mean expression U6 SnRNA was used as reference. The ΔΔCt value and the normalized miR-9, miR-34a and miR-196a salivary levels were calculated with Livak Method. **Results:** Our results showed that miR-9 and miR-34a levels in OSCC patients were significantly lower compared to healthy control groups (p= 0,01 and p= 0,012), and there was no significant difference in miR-196a levels (p> 0,05). In FA patients, miR-9 and miR-34 levels were lower than in control groups, likewise the OSCC patients (p =0,017 and p =0,014). There was no significant difference between miR-9, miR-34a, and miR-196a levels of FA patients and OSCC patients (p >0.05). **Conclusion:** According to our results, low levels of miR-9 and miR-34a in saliva are biomarker candidates that may be important for OSCC development. In FA patients, close follow-up of the levels of miR-9 and miR-34 would be appropriate considering OSCC development. Further studies are needed to confirm the potential of miR-9 and miR-34a as biomarkers for OSCC.

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PP 76

A NOVEL MISSENSE MUTATION OUTSIDE DNAJ DOMAIN OF DNAJC21 IS ASSOCIATED WITH SHWACHMAN-DIAMOND SYNDROME

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Shwachman-Diamond Syndrome (SDS) and related bone marrow failure disorders are characterized by early onset pancytopenia with a hypocellular bone marrow, short stature, and pancreatic insufficiency, along with an increased risk for myeloid malignancies. Recently, several cases with an SDS-like syndrome have been reported to harbor mutations in the DNAJ domain of *DNAJC21*. Here, we report an intriguing case

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

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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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PP 78

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