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Journal of Applied Hematology

(The Official Journal of Saudi Society of Hematology)

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CONTENTS

MENA Hematology Congress Abstracts
Madinah, KSA, 13–15 November 2025

ORAL PRESENTATIONS

MENA-O-108

Demographic Patterns and Therapeutic Responses in Glanzmann Thrombasthenia: a Multi-Center Study

Laila M. Sherief[1], Magdy El Ekiaby[2], Mahmoud El-Hawy[3], Eslam Elhawary[4], Amira A. Nazim[5], Samar M. Elbahy[6]

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BACKGROUND: Glanzmann thrombasthenia (GT) is a rare inherited bleeding disorder due to quantitative or qualitative defects of platelet integrin GPIIb/IIIa (CD41 and CD61). The prevalence of GT is unknown, but it is estimated to be about 1 per million and is more frequently encountered in populations with a higher rate of consanguinity. Phenotypically, bleeding sites and severity are quite variable. This study aimed to describe the demographic and clinical characteristics and response to treatment modalities used during bleeding episodes for patients diagnosed with GT in several hematology centers in Egypt.

PATIENTS AND METHODS: A cross-sectional multi-center study involved 87 children (<18 years old) diagnosed with Glanzmann thrombasthenia. Clinical data were recorded, such as bleeding manifestations and their response to various treatment modalities and laboratory results.

RESULTS: Eighty-seven children (61 females and 26 males) with a median age of 8 years were included in the study. Sixty-one children (55.2%) were born to consanguineous parents. The median age at diagnosis was 3 years (birth-14 years). The most frequent initial presenting symptom was epistaxis (39.1%), followed by cutaneous bleeding (29.9%), gastrointestinal bleeding (23%), heavy menstrual bleeding (19.5%), bleeding per gum (10.3%), and post-circumcision bleeding (5.7%), while one patient initially presented with intracranial hemorrhage (1.1%). In Addition, manifestations of recurrent bleeding were commonly reported as epistaxis (72.4%), cutaneous bleeding (44.8%), gastrointestinal bleeding (32.2%), and heavy menstrual bleeding (20.7%). Eighty children (92%) received treatment in the form of tranexamic acid, platelet transfusions, or recombinant factor VII. Platelet transfusions were given to 65 patients, 73.8% of them had a good response, and recombinant factor VII was given to 71 patients, 90.1% of them responded well to it. Type I Glanzmann thrombasthenia represented most of our patients (79.3%), followed by type III (11.5%), then type II (9.2%). No significant correlation was found between the expression of CD41 and CD61, and other parameters such as age of the patient, bleeding frequency, and response to treatment.

CONCLUSION: Glanzmann thrombasthenia is relatively common in communities with high rates of consanguineous marriages, making it a significant yet underestimated health concern in Egypt. In this study, Type I was the most prevalent subtype. The bleeding phenotype did not show significant variation across different types, with rFVIIa demonstrating a more effective treatment response compared to platelet transfusions. BMD findings and the frequency of admissions. Further research is needed to answer the proposed debate about the accuracy of DEXA scanning as diagnostic imaging in patients with SCD with low blood supply to the affected bone."

Keywords: Consanguineous marriages, bleeding phenotype

MENA-O-111

Diagnostic and Prognostic Values of NCF4 rs1883112 Polymorphism Expression among Acute Lymphoblastic Leukemia in Egyptian Children

Samar Mahmoud Elbahy, Walid Abdellatif Abdelhalim, Amira M. N. Abdelrahman, Adel Marzouk Ali, Omar Agha, Amr Fathy Mohamed Gad, Maha Rashwan

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BACKGROUND: Acute lymphoblastic leukemia (ALL) is the most prevalent pediatric malignancy. It has been linked to genes that impact the generation of reactive oxygen species. NCF4 rs1883112 is one of the implicated genes. This study aimed to evaluate the association between NCF4 SNP rs1883112 and susceptibility to developing ALL in Egyptian children, and its correlation with patient outcomes.

METHODS: This comparative case-control study included 100 Egyptian children newly diagnosed with ALL, collected from Benha University and Benha Children's Specialist Hospitals, Egypt. In addition, 100 healthy matched control children. A molecular study on peripheral blood for the detection of NCF4 rs1883112 polymorphism by real-time PCR was carried out. All patients were treated with the ST Jude Children's Research Hospital total XV protocol.

RESULTS: This study was conducted on 100 children with ALL; their mean age was 7.06 ± 4.62 years. The sample consisted

of 69% males and 31% females, in addition to 100 healthy children of matched age and gender. Complete remission was achieved in 95 patients; among them, 13 patients relapsed, and 6 patients died during the entire study period. The NCF4 rs1883112 polymorphism exhibited a protective effect against ALL susceptibility. Statistical analysis indicated that the GA and AA genotypes were associated with a lower risk of ALL, with odds ratios (ORs) of 0.50 (P = 0.001) and 0.54 (P = 0.015), respectively. The dominant model analysis further supported this finding, showing that the combined GA + AA genotypes had a lower risk of ALL (OR = 0.51, P = 0.001). The allele frequency analysis also revealed that the A allele is more common in the control group (53.5%) than in the case group (40.5%) and is associated with a decreased risk (OR = 0.72, P = 0.009). The AA genotype was significantly associated with a higher frequency of t(12;21) and those who achieved complete remission. There was no significant association between NCF4 (rs1883112) genotypes and other cytogenetics or immunophenotyping. The NCF4 rs1883112 dominant genotype hazard ratio was 0.15 for overall survival and 0.24 for disease-free survival by univariate analysis; this remained significant by multivariate analysis, suggesting that the GA/AA genotype was significantly associated with a lower risk of mortality and disease relapse.

CONCLUSION: It is suggested that the NCF4 rs1883112 (GA/AA genotype) has a significant protective effect against the development of ALL, which is associated with favorable outcomes in terms of overall survival and disease-free survival.

Keywords: NCF4 gene, rs1883112 polymorphism, acute lymphoblastic leukemia, diagnostic value, prognostic marker, Egyptian children

MENA-O-113

Evaluation of Safety and Efficacy of Romiplostim in the Treatment of Pediatric Immune Thrombocytopenia: A Cross-sectional study

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BACKGROUND: Evaluation of Safety and Efficacy of Romiplostim in the Treatment of Pediatric Immune Thrombocytopenia: A cross sectional study Purpose: Romiplostim is a thrombopoietin receptor agonist that is approved as a second-line therapy of primary immune thrombocytopenia (ITP). We aimed to evaluate the safety and efficacy of romiplostim in the treating children with persistent/chronic ITP.

METHODS: This observational cross-sectional study was carried at the Pediatric Hematology outpatient clinic of Benha University Hospitals, Egypt, during the period from December 2023 to March 2024, and enrolled 52 children with primary ITP on romiplostim therapy. The primary endpoint included the percentage of children who achieved a platelet response ≥ 50 × 10^9/L, for at least 2 successive weeks without rescue therapy, and the time to 1st platelet response, in addition to the reduction in bleeding episodes, especially significant bleeding, ITP-related hospital admissions, and the use of rescue therapy. The secondary endpoint included the percentage of children who attained a durable response, maintaining platelet response for ≥ 3-6 months without any rescue therapy, drug safety in addition to assessment of health-related quality of life (HRQoL) assessment using the PedsQL 4.0 Generic Core Scales.

RESULTS: The study involved 52 children diagnosed with primary persistent/chronic ITP having mean age of 10.4 ± 4.25 y (range, 3-16); with 28 (53.8%) males and 24 (46.2%) females. Romiplostim significantly decreased the frequency of significant bleeding episodes (from 76.9% to 34.6% of patients, P < 0.05), hospital admission rate (from 69.2% to 7.7% of patients, P < 0.05), and use of rescue medications (from 65.4% to 11.5% of patients, P < 0.05). Forty-eight patients (92.3%) achieved platelet response in a mean of 1.4 ± 3.2 years (range 0.25-15). Forty-two patients (80.8%) achieved a durable response. The most frequently observed side events were headache and joint pain; none of the patients developed serious adverse events. On assessment of HRQoL, scores were generally lower in parents' reports than children's reports at a mean of 78.6 ± 16.8 vs 81.8 ± 11.9, respectively. Platelet count was positively correlated with HRQoL in parent-proxy reports and platelet count ≥ 100 was a predictor of HRQoL in univariate linear regression analysis (P = 0.004).

CONCLUSION: We concluded that romiplostim is a safe and effective second-line therapy in the management of children with persistent/chronic ITP that significantly improved platelet count, reduced bleeding episodes, hospital stay rates, and the need to use rescue medications.

Keywords: Romiplostim, immune thrombocytopenia, pediatric patients, safety, efficacy, cross-sectional study

MENA-O-121

Which is More Effective: Weight-based or Hemoglobin-based Formula for Packed Red Cell Transfusion?

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BACKGROUND: Regular packed red cell transfusion remains the primary therapeutic approach for managing anemia in patients with transfusion-dependent thalassemia. However, these patients face multiple challenges, including limited availability of safe blood products and donors, which can affect the efficacy of transfusion therapy and overall quality of life.

AIM: To compare the effectiveness of weight-based versus hemoglobin-based formulas for calculating packed red cell transfusion volumes in transfusion-dependent thalassemia patients.

METHODS: This interventional study included 40 patients with transfusion-dependent thalassemia, who were randomly assigned to two groups and followed over a 6-month period.

- Group A received transfusions based on the traditional weight-based formula (20 ml/kg).
- Group B (intervention group) received transfusions based on the hemoglobin-based formula:

Volume (ml) =Weight (kg) × (Desired Hb (12 g/dL) – Actual Hb) ×3/Hematocrit of transfused unit.

All participants had their hemoglobin levels assessed before and within 24 h after each transfusion. Serum ferritin levels were measured at baseline and at the end of the 6-month follow-up.

RESULTS: Group B demonstrated a significantly higher post-transfusion hemoglobin level compared to Group A (12.80 ± 1.26 g/dL vs. 11.89 ± 1.16 g/dL; $P = 0.02$). Additionally, the total volume of packed RBCs transfused was significantly lower in Group B than in Group A (2274.0 ± 639 ml vs. 2535.0 ± 346 ml; $P = 0.043$). Notably, 60% of patients in Group B showed a significant reduction in both transfusion frequency and serum ferritin levels ($P < 0.001$).

CONCLUSION: The hemoglobin-based formula for calculating transfusion volume is more effective than the traditional weight-based method. It results in higher post-transfusion hemoglobin levels, reduced transfusion volume, and improved iron overload parameters.

Keywords: Packed red cells, Transfusion, Hemoglobin-based formula, transfusion efficacy, Thalassemia

MENA-O-141

Real World Data of Emicizumab Prophylaxis in Young Children with Severe Hemophilia A without Inhibitors

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Table 1: Baseline characteristics and characteristics post start of Emicizumab in the cohort

Characteristics		*n*=13 (%)
Baseline		
Age at onset in months median (range)		11 (6-18)
Sex	Male	13
Positive family history of HA		6 (46)
Prior treatment status		
	Treatment naive	5 (38)
	Minimal treatment	8 (62)
Type of prior treatment		
	FVIII	8 (62)
Bleed episodes before emicizumab		
	1-5	13 (100)
Indication of emicizumab*		
	Parental anxiety	0
	Difficult IV access	10 (77)
	Recurrent bleed on FVIII prophylaxis	5 (38)
	Poor compliance	5 (38)
Purpose of emicizumab		
	Bleed control	13 (100)
	De-novo	0
Type of bleed		
	Spontaneous	10 (77)
	Traumatic	3 (23)
	Procedural	0
Post Emicizumab		
Age at start of Emicizumab in months median (range)		16 (6-35)
Age Group		
	Age <1 year	4 (31)
	Age 1-2 year	3 (23)
	Age 2-3 year	6 (46)
Weight at start of Emicizumab in kg median (range)		10 (6-18)
Median follow-up in months (range)		24 (14-46)
Good compliance		12 (92)
Total no of bleeds post Emicizumab		2 (15)
Participants with >1 bleed		0
Participants with zero all bleeds		11 (85)
Total numbers of AE		4 (all grade 1)
Individual patients with AE		3 (23)

BACKGROUND: Young children (<3 year of age) with Hemophilia A (CwHA) are at increased risk of intracranial haemorrhage and vicious cycle of joint bleed leading to early arthropathy and inhibitor formation, thereby warranting prophylaxis at an early age. Young children are unique in view of physiological decreased production of vitamin K dependent coagulation factors, immunogenic naivety and increased clearance of emicizumab. Emicizumab is a bi-specific monoclonal antibody used in Hemophilia A and available in India since 2019. It has proven to be effective and safe in CwHA with and without inhibitors. Data on safety and efficacy of emicizumab in young children especially from low-middle income countries is limited.

AIMS: To study efficacy and safety of "standard-dose emicizumab prophylaxis" in young children with severe Hemophilia A without Inhibitor.

METHODS: We performed this retrospective analysis in young CwHA (<3 year of age at recruitment) started on emicizumab between Sep 2021 to Dec 2023. They were either treatment naïve or minimally treated ($\leq$5 exposure days). Inhibitor assay at the enrolment was done only for children with clinical suspicion. Emicizumab dose of 3 mg/kg/week for 4 weeks was followed by 6 mg/kg once/4 week was used. Doses were corrected to nearest vial strength. Demographic profile, disease and bleed related history was noted from case record forms. Emicizumab trough levels were estimated once at least 52-weeks after initiation.

RESULTS: Thirteen children with median age 16 month (6-35) at start of emicizumab were followed-up for median 24 month (14-46). The cumulative follow-up was 342 month. All children were symptomatic with 4/13 CwHA having joint bleeds. Five CwHA who had recurrent bleed on FVIII prophylaxis were screened for inhibitors and found to be negative. Post emicizumab, 11/13 CwHA had zero bleeds. Two children had minor traumatic bleeds (ecchymosis at hip region and dental bleed in one each). A total of four AE's (all grade 1) in three children were noted [Table 1]. One child had injection site swelling. Three CwHA had of emicizumab using Stago compact max-3 fully automated coagulometer. Local erythema at injection site. Mean emicizumab levels were 23.6 ± 10.5 µg/ml (95% CI: 17.24-30.01). Emicizumab levels of 10 children (77%) were below therapeutic range [Figure 1]. Hemophilia joint health score for all children > 4 year (n = 7) was zero.

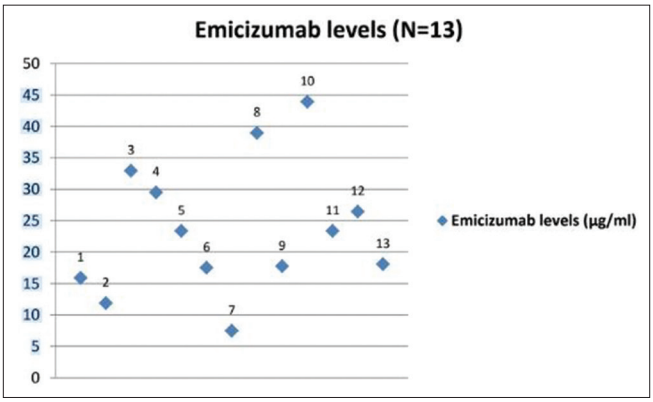

Figure 1: Emicizumab trough levels of all cases in the study

CONCLUSION: We found emicizumab prophylaxis safe and efficacious in young CwHA without inhibitors with good follow-up duration. Emicizumab levels lower than therapeutic-range was not associated with significant bleeds.

Keywords: Emicizumab Prophylaxis, hemophilia, inhibitors

MENA-O-144

Recurrent Epistaxis in a Child Due to Congenital Prekallikrein Deficiency: One of the Rarest Documented Bleeding Disorders Worldwide

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INTRODUCTION: Congenital prekallikrein (PK) deficiency is an extremely rare autosomal recessive coagulation disorder that typically presents with isolated prolongation of activated partial thromboplastin time (aPTT) in the absence of clinical bleeding. Symptomatic cases, especially in pediatric patients, are rarely reported worldwide and remain undocumented in Saudi Arabia to date.

AIM: To report a case of congenital PK deficiency presenting with recurrent epistaxis in a child from Saudi Arabia, highlighting one of the rarest inherited bleeding disorders seen in the region and the role of genetic testing in diagnosis.

METHODS: We evaluated a three-year-old male with spontaneous recurrent epistaxis and persistently prolonged aPTT. Coagulation factor assays and mixing studies were conducted. Due to normal factor levels and the absence of available contact factor testing locally, whole-exome sequencing was performed to establish the diagnosis.

RESULTS: Laboratory evaluation revealed significantly prolonged aPTT (121 s) with normal intrinsic pathway factor levels. Mixing studies confirmed the presence of a factor deficiency. Whole-exome sequencing identified a homozygous pathogenic KLKB1 frameshift mutation (c.451dupT), consistent with congenital PK deficiency. The patient was managed conservatively with nasal care, humidification, and local tranexamic acid application. Symptoms resolved, and no bleeding recurred during follow-up.

CONCLUSION: This case represents one of the rarest inherited bleeding disorders identified in Saudi Arabia, with no prior published reports of genetically confirmed PK deficiency from the country. It underscores the importance of considering contact factor deficiencies in children with isolated prolonged aPTT, even when clinical bleeding is mild. Genetic testing is critical for diagnosis, particularly where specialized assays are unavailable.

Keywords: Prekallikrein deficiency, recurrent epistaxis, congenital bleeding disorder, pediatric hematology, coagulation factors, rare bleeding disorders

MENA-O-147

Avascular Necrosis of the Femoral Head in Children and Adolescents with Sickle Cell Disease Presenting with Hip Pain

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BACKGROUND: Avascular necrosis is a progressive and potentially debilitating complication of sickle cell disease (SCD) which can affect any joint. However, in about 75% of cases it affects the hip joint as avascular necrosis of the femoral head (AVNFH), leading to chronic pain, disability, and reduced quality of life. Because of the limited literature on pediatric SCD patients with AVNFH, we analyzed clinical data to describe hematologic indices, treatments, and outcomes.

METHODS: We retrospectively analyzed 825 SCD patients to identify those with hip pain and at least one hip MRI. Clinical, laboratory, and imaging data were reviewed to assess AVNFH prevalence, risk factors, and outcomes. Surgical interventions and opioid use were tracked post-diagnosis. Group comparisons were conducted using chi-square and non-parametric tests. Healthcare utilization, laboratory markers, and treatment patterns were analyzed to identify associations with AVNFH. Statistical significance was set at $P < 0.05$.

RESULTS: Among 825 SCD patients, 104 (12.6%) with hip pain had undergone hip MRI; 57 (54.8%, overall prevalence 6.9%) had AVNFH. AVNFH patients were more likely male (73.7% vs. 36.2%, $P = 0.0001$) but did not differ in age (15.4 vs. 14.9 years, $P = 0.051$) or genotype ($P = 0.36$). They exhibited higher white blood cell count (9.1 vs. 7.0 × 103/mm3, $P = 0.042$), red cell distribution width (18.7% vs. 17.3%, $P = 0.011$), and lower MPV (8.7 vs. 9.2 fL, $P = 0.026$), with no differences in hemoglobin, HbF, or treatment history. AVNFH correlated with higher annual rates of ED visits (0.40 vs. 0.23, $P = 0.025$), hospitalizations (0.47 vs. 0.28, $P = 0.0052$), vaso-occlusive crises (0.65 vs. 0.34, $P = 0.0074$), and acute chest syndrome (0.16 vs. 0.06, $P = 0.021$). Of 57 AVNFH patients, 33 (57.9%) required surgery: 22 (38.6%) core decompression (median time to surgery: 6 months) and 13 (22.8%) total hip arthroplasty (THA; median time: 1 year). Sixty-one percent of decompression cases occurred within 3-18 months; one progressed to THA after 7 years. Opioid use increased non-significantly post-decompression (+3.5 morphine milligram equivalents/day [MME/day], 95%CI: −5.1, 12.1) but trended downward post-THA (−2.36 MME/day, 95%CI: −20.65, 0.11).

CONCLUSION: AVNFH is a common complication in SCD, particularly in males, and is linked to worse disease severity. Early diagnosis, individualized surgical planning, and optimized pain management are critical. Core decompression could serve as an intermediate step to delay THA, especially in growingchildren, where THA isn't an option. Future studies should explore whether early THA improves long-term pain control and reduces healthcare utilization.

Keywords: Sickle cell disease, avascular necrosis of the femoral head, risk factors, surgical interventions outcomes, opioid medication utilization, healthcare

MENA-O-149

Redox-Dependent Modulation of Interleukin-10 and Transforming Growth Factor-β Signaling in Pediatric Acute Lymphoblastic Leukemia

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BACKGROUND: Oxidative stress (OS) plays a pivotal role in the pathogenesis of Acute Lymphoblastic Leukemia (ALL), contributing to DNA damage and promoting leukemic cell survival. This study explores the interconnection between redox balance and the immunoregulatory cytokines Interleukin-10 (IL-10) and Transforming Growth Factor-β (TGF-β) in children with ALL.

METHODS: Serum samples from 100 ALL patients and 100 age-matched healthy controls were analyzed for levels of IL-10, TGF-β, Malondialdehyde (MDA), Catalase (CAT), and Superoxide Dismutase (SOD).

RESULTS: A significant inverse correlation between IL-10 and TGF-β ($\rho < 0.05$) was identified in the ALL group, suggesting opposing regulatory dynamics. TGF-β showed a positive correlation with antioxidant enzymes (CAT, SOD) and a negative correlation with MDA, indicating its potential association with reduced oxidative stress. In contrast, IL-10 levels were positively correlated with MDA and inversely related to antioxidant activity, reflecting a link to heightened oxidative damage.

CONCLUSION: These findings highlight a distinct redox-sensitive behavior of IL-10 and TGF-β in pediatric ALL and support their role as potential biomarkers for disease monitoring and therapeutic targeting. Further research is warranted to elucidate the mechanistic basis of these associations and their implications for redox-based interventions in ALL.

Keywords: Acute Lymphoblastic Leukemia, Oxidative Stress, IL-10, TGF-β, MDA, Catalase, Superoxide Dismutase

MENA-O-153

Assessing Maternal Micronutrient Levels and their Relationship with Cord Blood Quality Indicators

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BACKGROUND: Maternal micronutrients are crucial for fetal development and cord blood quality. Our study explored the relationship between maternal micronutrient levels and the quality of umbilical cord blood, which is used in transplants for genetic and blood disorders.

METHODS: This research involved pregnant women aged 18-36 who delivered via cesarean section. We analyzed cord blood units for various parameters, including volume, cell counts, and oxidative stress markers (SOD, CAT, GSH, and MDA). Additionally, we measured levels of essential micronutrients (Fe, Zn, Cu, and Se) to understand their impact on cord blood quality.

RESULTS: Our study of 50 pregnant women with a mean age of 27.20 ± 4.95 years found significant correlations between maternal micronutrient levels and cord blood parameters. Multivariate analysis showed that neonatal weight and specific micronutrients (Fe, Cu, and Se) had significant effects on cord blood quality, including volume, TNC, and CD34+ cells.

CONCLUSION: Our research demonstrates that a balanced intake of micronutrients during pregnancy is vital for cord blood quality and fetal development. Higher antioxidant micronutrient levels are associated with better-quality cord blood, which can improve transplantation outcomes.

Keywords: Cord blood, stem cells, micronutrients

MENA-O-157

Warm Autoimmune Hemolytic Anemia as a Gateway to Diagnosing XMEN Syndrome in a Young Child

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BACKGROUND: XMEN syndrome is a rare X-linked primary immunodeficiency caused by mutations in the MAGT1 gene. It is typically characterized by chronic Epstein-Barr virus (EBV) viremia, lymphoproliferative disorders, and immune dysregulation. While EBV infection is commonly associated with cold agglutinin autoimmune hemolytic anemia (AIHA), its association with warm AIHA is extremely rare.

CASE PRESENTATION: We report a case of a previously healthy 30-month-old boy who presented to the emergency department with jaundice, fatigue, and fever. Laboratory investigations revealed severe anemia (hemoglobin 2.8 g/dL), high reticulocyte count (76%), and a positive direct antiglobulin test (DAT) with IgG positivity, consistent with warm AIHA. Peripheral smear showed polychromasia, nucleated RBCs, and autoagglutination. The patient was admitted to the pediatric intensive care unit and initiated on pulse methylprednisolone, IVIG, and red blood cell transfusions. Due to persistent hemolysis, therapy was escalated to include rituximab. Further evaluation revealed persistent EBV viremia and low serum IgM, raising concern for underlying immunodeficiency. Whole-exome sequencing identified a hemizygous likely pathogenic MAGT1 variant: c. 828_829del (p. His 277GlnfsTer8), confirming the diagnosis of XMEN syndrome.

DISCUSSION: This case highlights an unusual and severe initial presentation of XMEN syndrome with warm AIHA, a rare phenomenon in both EBV infection and primary immunodeficiencies. The poor response to conventional therapy underscores the complexity of managing autoimmune cytopenias in immunodeficient patients. XMEN syndrome disrupts N-linked glycosylation and magnesium signaling, leading to impaired cytotoxic function of CD8+ and NK cells—key in controlling EBV-infected B cells. Identification of MAGT1 mutations and decreased NKG2D expression can aid in early diagnosis.

CONCLUSION: This case represents a unique pediatric presentation of XMEN syndrome with EBV-induced warm AIHA. It emphasizes the need to consider underlying genetic immunodeficiencies in cases of refractory AIHA, especially when linked with viral infections. Early recognition and genetic diagnosis are critical to optimizing management and considering curative strategies such as hematopoietic stem cell transplantation.

Keywords: Warm autoimmune hemolytic anemia, XMEN syndrome, immunodeficiency, pediatric hematology, genetic diagnosis, case report

MENA-O-158

Angiotensin Converting Enzyme Inhibitors (ACEIs) for Anthracycline-induced Cardiomyopathy: A Systematic Review and Meta-analysis of Randomized Controlled Trials

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INTRODUCTION: Anthracyclines are widely used chemotherapeutic agents in treating various hematological malignancies and solid tumors, but are limited by dose-dependent cardiotoxicity, leading to cardiomyopathy and heart failure. Their activation of the cardiac renin-angiotensin system increases oxidative stress, suggesting apotential protective role for angiotensin-converting enzyme inhibitors (ACEIs).This systematic review and meta-analysis evaluates the efficacy and safety of ACEIs in preventing anthracycline-induced cardiomyopathy.

METHODS: A systematic search of PubMed, Embase, Cochrane Library, and ScienceDirect identified randomized controlled trials (RCTs) assessing ACEIs for anthracycline-induced cardiotoxicity. The primary outcome was left ventricular ejection fraction (LVEF) change; secondary outcomes included incidence of heart failure, arrhythmias, and treatment-related adverse effects. A random-effects meta-analysis was performed using RevMan 4.5.1. software.

RESULTS: Nine RCTs involving 869 cancer patients were analyzed. At six months of follow-up, ACEIs significantly improved LVEF by 7.94% compared to controls (weighted mean difference (WMD): 7.93%, [95% CI] = [3.18-12.67], $P = 0.001$). At twelve months of follow-up, 4.08% improvement was observed, without statistical significance (WMD: 4.08%, 95% CI: [-0.53-8.69], $P = 0.08$). ACEIs were associated with non-statistically significant lower rates of heart failure and

arrhythmia development compared to the control (Odds ratio (OR) 0.20 [95% CI] = [0.03, 1.60], *P* = 0.13) and (OR 0.41 [95% CI] = [0.05, 3.50], *P* = 0.42) recepctively. No significant differences were noted in adverse events (OR 0.47, [95% CI] = [0.03-7.64], *P* = 0.60).The quality of evidence for the outcomes was assessed using the Grading of Recommendations, Assessment, Development, and Evaluation (GRADE) approach. Evidence was rated as high quality for LVEF at six months, moderate quality or LVEF at twelve months and heart failure, and low quality for arrhythmias and adverse events.

CONCLUSION: ACEIs show potential in reducing acute and short-term anthracycline-induced cardiotoxicity without significant adverse effects. This promising outcome, coupled with the need for ongoing research, underscores the potential of ACEIs to offer promising outcomes for patients undergoing anthracycline chemotherapy.

Keywords: Angiotensin converting enzyme inhibitors, anthracycline-induced cardiomyopathy,cardiotoxicity, systematic review, meta-analysis, randomized controlled trials

MENA-O-161

Half-life of Factor VIII in an Adult and Pediatric Population: Study at 03 Centers in Western Algeria

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On-demand treatment and even standard prophylactic regimens have their limitations. The approach of personalizing prophylaxis by studying the half-life of hemophilia A patients was born following the discrepancy observed between patients without spontaneous bleeding despite FVIII levels below 1%. The objective of our study is to determine the pharmacokinetic profile of our hemophiliac population in western Algeria and to evaluate the individual variability of our patient's estimated using PK modeling and Bayesian analysis. 39 patients with hemophilia A were followed at the Univeristy hospital of Oran, at the Pediatric establishment and at the Establishment of Tiaret. The Bayesian approach made it possible to calculate the difference between an individual and a population to predict an individual half-life. The most likely PK values for each patient were estimated from a limited number of FVIII levels using MyPKFiT, a web application that allows users to simulate dosing regimens using pharmacokinetic profiles (PK) individualized. The MyPKFiT application can be used with only 02 samples: the first between 03 and 04 hours (+/-30 min) after the injection and, the second between 24 and 32 h (+/-1 h) after the injection. The development of pharmacokinetics in the adjustment of individual prophylactic regimens for hemophilia A would most likely have a very significant medical and socio-economic impact in the management of the disease, particularly in developing countries. PK parameters are of great importance in the preventive treatment and management of hemophiliacs.

Keywords: Factor VIII, half-life, hemophilia A, pharmacokinetics, pediatric and adult population, Algeria

MENA-O-162

Correlation Study between Prognostic Scores GLATIT and Prognostic Scores ISTH-BAT over the Inherited Platelet Disorders: study in the Western of Algeria

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Inherited platelet disorders are a heterogeneous group of platelet diseases responsible for hemorrhagic manifestations of highly variable severity from patient to patient. For these platelet disorders, the only prognostic score is the GLATIT score valid only for Glanzmann thrombasthenia. We were inspired by the example of Von Willebrand's disease, many of whose clinical symptoms are similar to those of platelet disorder and where the ISTH-BAT score is recognized by the International Society of Hemostasis and Thrombosis ISTH to have a prognostic and diagnostic value to apply other scores belonging to other pathologies to our patients with inherited platelet disorders We recruited 60 patients including 34 Glanzmann thrombasthenia, 18 patients with Jean Bernard Soulier disease and 8 May-Hegglin patients. GLATIT is the only published score for the prognostic evaluation of Glanzmann thrombasthenia. We proposed to apply it to our Glanzmann thrombasthenia (TG) series, then to Jean Bernard Soulier's disease and May-Hegglin's disease. The ISTH-BAT score is a diagnostic and prognostic test, it allows a clinical assessment of the severity of bleeding in patients with von Willebrand disease. This score has been validated for Von-Willebrand disease. We used it for inherited platelet disorders. We studied the correlation between the GLATIT score and the ISTH-BAT score and the concordance rate and the Kappa test allow us to assess the strength of this agreement between 2 different scores (2 qualitative variables) during Inherited platelet disorders. The ISTH-BAT score is quite applicable to inherited platelet disorders, this will give us simple tools, accessible to any clinician, whatever his place of exercise and whatever the state and the performances of this equipment, to indicate screening, help to classify patients according to different degrees of severity and tailor treatments and could possibly be included among the criteria diagnoses in some cases.

Keywords: Inherited platelet disorders, prognostic scores, GLATIT score, ISTH-BAT score, correlation study, Algerian population

MENA-O-167

Prognostic Significance of PNH Clones in Aplastic Anemia Treated with Immunosuppression or Allogeneic Hematopoietic Stem Cell Transplantation

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BACKGROUND: Aplastic anemia (AA) is an immune-mediated bone marrow failure characterized by pancytopenia and hypocellular marrow. Small PNH clones are found in 50-60% of AA and other marrow failure syndromes. Treatment depends on severity and age; allogeneic hematopoietic stem cell transplantation (HSCT) is preferred for young patients with AA.

AIM: To evaluate the impact of PNH clone positivity on survival outcomes, transplant-related complications, and relapse rates among patients with AA treated with either immunosuppressive therapy (IST) or HSCT.

METHODS: Retrospective analysis of 163 patients of AA treated with either IST or HSCT from 2003 to 2023. Clinical data were obtained from the BMF and HSCT data base at KFSH&RC-Riyadh.

RESULTS: Among the 163 patients, 89 (54.6%) male and 74 (45.3%) were female, and the median age was 22(IQR17-29) years. In all, 122 (74.8%) patients were negative for PNH, while 41 (25.2%) patients with PNH positive. Overall, 104 (63.8%) patients underwent HSCT and 59 (36.2%) patients received IST. After median follow-up of 122.7 months (95% CI: 109-136), overall survival (OS) at 5 years was 100% for PNH-positive and 85.5% for PNH-negative patients, ($P = 0.01$). When further stratified for first-line treatment, 5-year OS was 100% for PNH-positive as compared to 86.5% for PNH-negative in IST, ($P = 0.09$). Among those undergoing HSCT, OS was 100% for PNH-positive and 85.3% for PNH-negative, ($P = 0.07$).In the PNH-positive and PNH-negative groups, event-free survival at 5 years was 83.9% versus 47.7%, respectively, ($P = 0.6$). In HSCT, the cumulative incidence of aGvHD in PNH-positive and PNH-negative patients was 9.5% and 15.6%, respectively, ($P = 0.4$), while cGvHD in PNH-positive and PNH-negative patients was seen in 5% and 21.7%, respectively, ($P = 0.06$). Graft failure rates were 13.9% in PNH-positive and 15.4% in PNH-negative patients, ($P = 0.7$), whereas the cumulative incidence of relapse was 16.1% in PNH-positive and 8.5% in PNH-negative patients in the IST group, ($P = 0.06$). Non relapse mortality was 0% in the PNH-positive group versus 6% in the PNH-negative group, ($P = 0.2$).

CONCLUSION: The survival rate of patients who had PNH-positive clones proved better than those who were PNH-negative. Factors associated with improved survival included upfront allogeneic HSCT and PNH clone positivity. The results indicate that PNH clone status should be considered as a prognostic factor in choosing the treatment for AA. Further studies are needed to address the prognostic effect of PNH in AA.

Keywords: Paroxysmal nocturnal hemoglobinuria clones, aplastic anemia, immunosuppressive therapy, allogeneic hematopoietic stem cell transplantation, prognostic significance, bone marrow failure

MENA-O-168

Expanding the Spectrum of Inherited Bone Marrow Failure: Genetic Mutations in Saudi Patients Carry Myeloid Malignancy Risk

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BACKGROUND: Inherited bone marrow failure (IBMF) are increasingly being recognized due to use of NGS. BMF secondary to mutations in ERCC6 L 2,MYSM 1 and OSM has been reported.

AIM: To highlight clinical characteristics, clonal progression and treatment outcome for Saudi patients with rare BMF syndromes.

METHODS: A retrospective analysis evaluated 29 patients diagnosed with germline mutations. Data collected from BMF registry and WES identified the mutations.

RESULTS: ERCC6 L2 mutation: 18 patients from ten Saudi families with mutations of the ERCC6 L2 gene. Six (33.3%) of the patients had BMF, 4 (22.2%) had AML, 3 (16.6%) Hypoplastic MDS, 3 (16.6%) MDS-EB, one (5.5%) had quadriplegia and one with normal CBC. Anemia was observed in 16 (88.8%), thrombocytopenia in 15 (83.3%), and leucopenia in 9 (50%) patients. Bone marrow was hypocellular in 12 (66.6%) and hypercellular in 3 (16.6%) cases. TP53 mutation was seen in 6 patients, complex karyotype in three patients and two patients had monosomy 7. Ten of the patients underwent HSCT. Among them, 4 patients with MSD, 2 patients with MUD; two patient underwent haploidentical HSCT and one patient received HSCT from umbilical cord blood. Seven of them have been treated conservatively or with immunotherapy. The 3-years-overall survival (OS) was 80.4%.

MYSM1 DEFICIENCY: Seven patients carried biallelic MYSM1 mutations presented with anemia in early childhood diagnosed with BMF at median age of six months. No-hematological features include facial dysmorphism in two, ASD and bone lesions associated with Immunological deficiency. Patients exhibited variable BMF severity with transient improvement; later transfusion dependence and progression to MDS/AML in 5 patients during 10-12 years since initial presentation with TP53 and CALR mutations and monosomy 7 and 5q deletion and t(1;19) suggest clonal evolution. 3 patients successfully underwent haploidentical HSCT for progressive BMF, transfusion dependency and MM with adverse cytogenetics.

OSM DEFICIENCY: Four female patients from consanguineous families were presented with anemia, thrombocytopenia, or pancytopenia between ages 13 months and 16 years. Bone marrow biopsy showed hypocellular marrows with megaloblastoid changes. Cytogenetic abnormalities including RUNX1 mutation and t(10;13) in two. WES identified homozygous OSM nonsense mutations. Two responded to eltrombopag and danazol while one patient received successful haploidentical HSCT due to high-risk cytogenetic abnormalities.

CONCLUSION: These cases show that newly recognized IBMFS in Saudi patients have variable clinical presentations

and heightened risk of myeloid malignancy. Early mutation detection and vigilant monitoring for clonal evolution are crucial. HSCT should be considered for progressive disease to improve outcomes.

Keywords: Inherited bone marrow failure, genetic mutations, myeloid malignancy risk, Saudi patient's hematologic genetics, bone marrow disorders

MENA-O-169

Unraveling the Prognostic Impact of CDKN2A/2B Gene Deletion in Children Diagnosed with Acute Lymphoblastic Leukemia, in Saudi Arabia: A Multicenter Study

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BACKGROUND: Deletions in the CDKN2A/2B gene locus are among the most common genetic alterations in pediatric acute lymphoblastic leukemia (ALL). Globally, these deletions have been associated with adverse outcomes, yet limited data exist regarding their impact in Middle Eastern populations.

AIM: To evaluate the clinical, biological, and prognostic impact of CDKN2A/2B deletions in pediatric ALL patients in Saudi Arabia through a multicenter retrospective study.

METHODS: This study analyzed 352 pediatric ALL cases diagnosed between 2015-2023. Patients were divided into two groups: those with CDKN2A/2B deletions (CDKNA group, n = 78) and those without (non-CDKNA group, n = 274). Demographics, clinical features, cytogenetics, treatment response, toxicities, and outcomes were compared using chi-square testing for categorical variables and Kaplan-Meier survival analysis. A *P* value < 0.05 was considered statistically significant.

RESULTS: Patients in the CDKNA group were significantly older at diagnosis (38.5% vs. 19.3% >10 years, *P* < 0.001) and had higher initial WBC counts (>50 × 109/L; 39.7% vs. 19.7%, *P* < 0.001). T-cell ALL was more frequent (9.0% vs. 2.9%, *P* = 0.028), and favorable cytogenetics (e.g., ETV6-RUNX1) were less common (16.7% vs. 27.7%, *P* = 0.047). CDKNA patients were more likely to be classified as high-risk (60.3% vs. 40.1%, *P* = 0.002).On day 15 of induction, poor marrow response (M3) was significantly more common in the CDKNA group (22.8% vs. 7.7%, *P* = 0.022). Treatment-related toxicity was higher, including infections (82.1% vs. 63.8%, *P* = 0.002) and asparaginase hypersensitivity (45.5% vs. 3.8%, *P* < 0.001). Outcomes were notably worse in the CDKNA group, including lower remission rates (65.4% vs. 83.8%, *P* < 0.001), higher relapse (30.8% vs. 15.7%, *P* = 0.003), and mortality (25.6% vs. 8.8%, *P* < 0.001). Kaplan-Meier curves confirmed significantly reduced overall survival and event-free survival (*P* < 0.001 and *P* = 0.007, respectively).

CONCLUSION: CDKN2A/2B deletions are associated with aggressive disease features, poor early treatment response, higher toxicity, and worse survival in pediatric ALL patients. Identifying this deletion at diagnosis is essential to guide intensified or alternative therapy approaches in high-risk subgroups. Further prospective validation is warranted.

Keywords: CDKN2A/2B gene deletion, acute lymphoblastic leukemia, pediatric leukemia, prognosticmpact, genetic abnormalities, Saudi Arabia, multicenter study

MENA-O-171

Impact of Cytoreductive Nephrectomy on Outcomes in Metastatic Renal Cell Carcinoma Patients with Concurrent Hematological Malignancies Receiving Immune Checkpoint Inhibitor Therapy: A Multi-institutional Retrospective Cohort Analysis

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INTRODUCTION: The best timing of cytoreductive nephrectomy in metastatic renal cell carcinoma (mRCC) patients with concurrent hematological malignancies receiving immune checkpoint inhibitors (ICIs) remains undefined. This study evaluated the impact of upfront nephrectomy on the outcomes in these targeted population of interest.

METHODS: We conducted a retrospective cohort analysis using the TriNetX database from total of 144 institutions from different countries around the world, but mainly in the United States. We identified 726 patients with mRCC and concurrent hematological malignancies receiving ICI therapy between January 2015 to June 2025. Patients were according to nephrectomy status and hematological malignancy presence into four groups. Primary outcome was overall survival; secondary outcomes included severe adverse events and hospitalizations. Propensity score matching (PSM) and multivariable modeling were performed.

RESULTS: After PSM, 107 patients each were analyzed in the primary comparison groups (Heme + Neph + vs Heme + Neph-). Median overall survival was 34 months versus 32 months respectively (HR 0.874, 95% CI 0.580-1.317, *P* value = 0.520). The nephrectomy group demonstrated significantly higher hospitalization rates (100% vs 48.5%, absolute risk difference + 51.5%, *P* value < 0.001, NNH = 2). Severe adverse event rates were similar between groups (49.3% vs 48.9%, *P* value = 0.476). Among patients without hematological malignancies, nephrectomy showed a deviation toward better survival benefit (HR 0.851, *P* value = 0.056).

CONCLUSIONS: In mRCC patients with concurrent hematological malignancies, upfront cytoreductive nephrectomy

does not provide significant survival benefit but significantly increases healthcare utilization. These findings support individualized treatment decisions focusing on the importance of systemic therapy prioritization.

Keywords: Cytoreductive nephrectomy, metastatic renal cell carcinoma, hematological malignanciesimmune checkpoint inhibitors, treatment outcomes, retrospective cohort study, multi-institutional study

MENA-O-172

Acute Admissions in Sickle Cell Disease in Saudi Children in Eastern Province

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BACKGROUND: Sickle cell disease (SCD) in eastern province (EP) of Saudi Arabia is common, due different haplotype of SCD in this area, this study conducted to see the complication of SCD and compare it with other studies.

PATIENTS AND METHODS: Retrospective study done March 2016 and September 2020 for 702 patients with tracing all admissions related complication and its sequences.

RESULTS: During study period, 1415 admission (5% from total admission) for patients with SCD, 91.5% have sickle cell homozygous. Painful crisis still the commonest complication (65%), where acute splenic sequestration is significantly high in Eastern Province comparing to SCD from western province (WP) of Saudi Arabia. Hyper hemolytic crisis is not uncommon due to high prevalence of G6PD deficiency in this area. Three deaths occurred during the study period only one of them is related to SCD (hyper hemolytic crisis). No significant between gender and in the hemoglobin or hemoglobin electrophoresis.

CONCLUSIONS: Sickle cell disease in eastern province came with less severe clinical picture but due to of high incidence in this area, as well as it is huge burden and health care challenge.

Keywords: Sickle cell disease, acute admissions, pediatric hematology, Saudi children, Eastern Province, hospitalization outcomes

MENA-O-173

Bleeding Disorder

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INTRODUCTION: Hereditary F VII deficiency is clinically and biologically heterogeneous. The severity of hemorrhagic signs is variable and poorly correlated with plasma FVII: C levels. The majority of studies note discrepancies between the factor level and the importance of hemorrhagic signs; moreover, common hemostasis tests are poorly predictive in terms of hemorrhagic risk. In addition, a proportion of patients remain asymptomatic, which can make the prediction of bleeding risk and its management difficult. Two severity scales are currently available (Peyvandi, Jain).

OBJECTIVE: Analyze the relationship between the hemorrhagic phenotype and the plasma level of factor VII.

METHODS: This is a retrospective study, which includes all patients with factor VII deficiency followed at the Hemophilia Center (CTH) of the Beni Messous University Hospital. The various data were collected from patient files. patients: age; history (ATCDS) of bleeding; the level of factor VII. We classified the severity of the deficit according to two scales: Peyvandi: A Jain: B.

RESULTS: One hundred and forty-nine patients with factor VII deficiency are followed at the CTH. Their average age is 36.9 years (18-89), including 46 men and 103 women. The sex ratio is 2.2. Scale A: for a severe deficit 23.8% of patients were asymptomatic, for moderate and minor 65.42% presented a hemorrhagic syndrome and 4.67% observed postoperatively. Scale B: For a severe rate, 100% of our patients presented with hemorrhagic ATCDS; for moderate and minor rates 66.9% presented hemorrhagic ATCDS and 4.22% observed in postoperative Comments In the case of a severe deficit according to scale B; a correlation exists between the rate and the phenotype, but for higher rates, the bleeding risk is poorly predictive. Furthermore, according to scale A, a discordance is found for all factor VII levels, and we observed that some patients presented a hemorrhagic syndrome postoperatively despite high levels and the absence of all hemorrhagic ATCD.

CONCLUSION: The direct relationship between the plasma level of factor VII and the hemorrhagic phenotype remains unproven. It is necessary to establish a score including parameters which can better guide therapeutic indications (hemorrhagic ATCD, the site of the intervention, comorbidities, the age of symptomatological presentation).

Keywords: VII deficiency, score, management

MENA-O-176

Assessment of Cognitive Function in Children with Beta Thalassemia Major: A Cross-sectional Study

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BACKGROUND: Multiple risk factors contribute to cognitive impairment in children with b-thalassemia major.

AIM: For a more refined understanding of this issue, we attempted to evaluate cognitive function in b-thalassemia major patients and identify the relationship between possible cognitive dysfunction and the following: demography, transfusion and chelation characteristics, iron overload, and disease complications.

METHODS: We studied 100 b-thalassemia major children and 100 healthy controls who matched well in terms of age, sex, and socioeconomic status. All participants underwent psychometric assessment using Wechsler Intelligence Scale for Children–Third Edition, Arabic version.

RESULTS: The mean Full-Scale IQ and Performance IQ of patients were significantly lower than those of controls, whereas no significant difference was found for Verbal IQ. No significant relationship existed between IQ and any of the assessed parameters.

CONCLUSION: We concluded that Performance IQ, not Verbal IQ, was significantly affected in b-thalassemia major patients, but there was no clear association between IQ and any of the parameters.

Keywords: Beta thalassemia major, cognitive function, pediatric hematology, neurocognitive assessment, cross-sectional study, children

MENA-O-186

Renal Complications in Children with Sickle Cell Disease: A Retrospective Cohort Study

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BACKGROUND: Sickle cell disease (SCD) is a hereditary hemoglobinopathy associated with multi-organ complications, including early-onset renal impairment. In Saudi Arabia, SCD is a growing public health concern, particularly in regions like Al-Madinah Al-Munawwarah.

OBJECTIVES: To assess the prevalence and clinical patterns of renal complications in pediatric SCD patients, with a focus on glomerular hyperfiltration and genotype-specific differences.

METHODS: A retrospective cohort study was conducted on 230 pediatric and adolescent patients with confirmed SCD. Clinical and laboratory data were collected from two regional medical centers. Renal function was assessed using age-specific eGFR formulas: Full Age Spectrum (FAS) for those under 18 years and CKD-EPI for those 18 and older.

RESULTS: Hyperfiltration was identified in 84.7% of patients based on minimum eGFR, with the highest prevalence in children aged 4-10 years (97.3%). Only two patients met criteria for CKD stages 2 or 3. The HbSS genotype was significantly associated with higher rates of hospitalization, transfusion, and frequent vaso-occlusive crises ($P < 0.05$). Multivariable logistic regression showed that age < 10 years and ≥ 4 VOC episodes annually were independent predictors of hyperfiltration. The HbAS genotype was associated with lower odds of hyperfiltration.

CONCLUSION: Hyperfiltration is common in pediatric SCD, especially among younger children and those with severe clinical phenotypes. These findings support the need for early renal screening and genotype-guided monitoring to prevent long-term kidney complications.

Keywords: Sickle cell disease, renal complications, pediatric nephrology, retrospective cohort study,kidney dysfunction, children

MENA-O-187

Cytogenetic Findings and Post Induction MRD in B-Lymphoblastic Leukemia

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BACKGROUND AND OBJECTIVES: Cytogenetic and molecular genetics play a crucial role in the diagnosis of B-lymphoblastic leukemia (B-ALL). These genetic alterations are decisive in risk stratification and chemotherapy protocols. Conventional karyotyping and fluorescence *in situ* hybridization (FISH) are widely used diagnostic tools. Despite their complementary nature, discrepancies between these techniques can pose challenges, particularly when predicting post-induction minimal residual disease (MRD). This study aims to predict the gap between karyotyping and FISH by evaluating their concordance and investigating how combined cytogenetic findings correlate with post-induction MRD status.

METHODS: A retrospective study was conducted in the Cytogenetics laboratory of Indus Hospital, Karachi. All newly diagnosed patients of B-ALL (1-17 years) from January 2021 to September 2024. Cytogenetic including karyotyping by G-banding and FISH for BCR-ABL1, ETV6-RUNX1, and KMT2A gene rearrangement were done on a semi-automated system MB8 Leica Biosystems, Germany. Post-induction MRD was done by 8-color flow cytometry on BD FACS CANTO-II using post-induction day 35 bone marrow aspirate.

RESULTS: This study included 976 patients, with a median age of 7.00 years (IQR: 5.00-11.00) with M/F ratio 1.5:1. In the BCR-ABL1 analysis, the most frequent finding was gain of BCR1 followed by BCR/ABL1 positivity with a concordance rate of 61% and 76% respectively when compared with karyotype. The ETV-RUNX1 analysis revealed that the most frequent abnormality was RUNX1 gain followed by the ETV-RUNX1 fusion with a concordance rate 27% for both. KMT2A gain was observed in 8.73% of cases, with a concordance rate of 72.4% followed by KMT2A rearrangement. All negative cases were concordant across both FISH and karyotype. Statistical analysis confirmed a significant association between FISH

findings and karyotype (p = <0.001), with Cramer's V values of 0.61, 0.793, and 0.812 for BCR-ABL1, KMT2A, and ETV-RUNX1, respectively. Post-treatment minimal residual disease (MRD) was detected in 27% of the cohort. A detailed analysis revealed MRD positivity rates highly associated with specific genetic alterations.

CONCLUSION: In this study, FISH and karyotyping demonstrated moderate to strong concordance for key cytogenetic abnormalities. Post-induction MRD revealed strong associations with BCR-ABL1 and KMT2A abnormalities, highlighting their potential as predictors of residual disease risk. These findings underscore the complementary role of FISH and karyotyping in B-ALL diagnosis and risk assessment.

Keywords: B-lymphoblastic leukemia, cytogenetic findings, minimal residual disease, post-inductiontherapy, pediatric leukemia, prognostic markers

MENA-O-206

Overcoming Surveillance Gaps: Deep Learning for Accurate Detection and Chronicity Classification of Hospital-acquired Pulmonary Embolism

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INTRODUCTION: Hospital-acquired venous thromboembolism (HA-VTE) is one of the most preventable causes of in hospital death. Accurate detection is essential for evaluating the effectiveness of thromboprophylaxis and guiding safety interventions. At our institution, radiology reports are unstructured, limiting the effectiveness of automated tools. Prior efforts using ICD codes and trigger-based systems were inaccurate, and manual chart review—though more reliable—proved too labor-intensive for sustained surveillance. Moreover, existing systems do not reliably distinguish between acute and chronic PE, which is critical for identifying new, hospital-acquired events. These limitations drove the development of an AI based solution to extract and classify VTE events directly from radiology reports and support timely identification of HA-VTE.

METHODS: We developed a deep learning-based binary classification system using BiomedNLP-PubMedBERT-base uncased-abstract-fulltext, fine-tuned on radiology reports. The deep neural network architecture included a pre-trained transformer encoder, dropout layer (0.3) for regularization, and linear classifier for binary PE detection. Deep learning training employed class-balanced loss weighting, AdamW optimization, and linear scheduling with warmup over 15 epochs. Performance was evaluated using accuracy, precision, recall, F1-score, and AUC metrics on a held-out test set. We included all CT pulmonary angiography reports from January 2024 to the end of June 2025.

RESULTS: The binary PE detection model achieved exceptional performance with 99.47% accuracy, F1-score of 0.99, precision of 0.98, and perfect recall of 1.00 on the test dataset containing 2,679 radiology reports (35.27% Positive). The acute/chronic of the positive reports classification model demonstrated strong performance with 96.48% accuracy, F1-score of 0.893, precision of 0.913, and recall of 0.875. Error analysis revealed only 2 misclassified cases in binary detection and 5 cases in acute/chronic classification, indicating robust model performance.

CONCLUSION: Our AI-based tool accurately detects hospital-acquired PE from unstructured radiology reports, addressing key limitations of existing surveillance methods. In addition to binary detection, it accurately classifies the chronicity of PE, enabling more precise identification of hospital-acquired events. Future work will extend this approach to lower limb DVT, automate real-time triggering within clinical workflows, and integrate it with hospital data systems to identify predictors of hospital-acquired VTE and guide targeted prevention strategies.

Keywords: Hospital-acquired pulmonary embolism, deep learning, medical imaging, chronicity classification, automated detection, clinical surveillance

MENA-O-209

Hodgkin Lymphoma

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BACKGROUND: In advanced-stage classical Hodgkin lymphoma (cHL), achieving a negative interim PET (iPET) after two cycles of ABVD is generally considered a favorable prognostic marker. Most patients with early metabolic response proceed to complete therapy without residual disease. However, a subset of patients experience discordant end-of-treatment (EOT) findings, with evidence of the diseases despite an initial complete response.

AIMS: This study aimed to identify baseline clinical features that might help predict a discordant EOT.

METHODS: A retrospective review was conducted of 122 patients with stage III or IV cHL treated at our institution between 2016 and 2024. All patients achieved complete metabolic response (Deauville score $\leq$ 3) on Ipet following two cycles of ABVD. Clinical and laboratory data, including IPS score, bone marrow and extranodal involvement, were collected. The primary outcome was residual disease identified at EOT-PET. Logistic regression was used to identify predictive factors. Disease-free survival (DFS) was estimated using Kaplan-Meier analysis.

RESULTS: Of the 122 patients with negative iPET scans, 21 (17.2%) were found to have residual uptake on EOT-PET. The median age was 28 years; 63% were male, and 70.5% had nodular sclerosis subtype. Most patients (69.7%) had stage IV disease. In univariate analysis, no baseline variable—including IPS score, bone marrow involvement, extranodal disease, or B symptoms—was significantly associated residual PET positivity. However, in multivariate analysis, stage IV disease emerged as an independent predictor, with a statistically significant association with residual disease at EOT (OR 3.32; 95% CI, 0.97-11.37; $P = 0.041$). The median follow-up was 28.8 months. The median DFS was not reached, and the estimated two-year DFS was 83%.

CONCLUSIONS: While early metabolic remission on interim PET is reassuring, a notable proportion of patients still demonstrate residual disease by the end of therapy. Stage IV disease was independently associated with this outcome when adjusted for other variables. No significant associations were identified with IPS score, bone marrow, or extranodal involvement. These findings suggest that relying solely on interim PET may overlook residual risk in a subset of patients, particularly those with extensive disease burden at baseline.

Keywords: Hodgkin lymphoma, lymphoid malignancy, chemotherapy, radiotherapy, treatment outcomes, prognosis

MENA-O-210

Hemophilia

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BACKGROUND: Haemophilia A management has advanced significantly with the introduction of extended half-life (EHL) recombinant factor VIII (FVIII) therapies, notably efmoroctocog alfa. Efanesoctocog alfa further extends FVIII half-life by decoupling it from the von Willebrand Factor, allowing once-weekly dosing with sustained FVIII activity to normal or near normal levels in children and adults for 3 and 4 days after injection, respectively. This report evaluated treatment patterns and outcomes of switching from efmoroctocog alfa to efanesoctocog alfa prophylaxis in children with haemophilia A in the United Arab Emirates (UAE).

METHODS: This retrospective, single-centre report evaluated 22 paediatric, male haemophilia A patients (median age = 9.0 years [IQR: 7.3-15.5]) transitioning from efmoroctocog alfa to efanesoctocog alfa prophylaxis at Tawam Hospital, UAE. All patients received efmoroctocog alfa prophylaxis during the pre-switch period (January 2023 to July 2024) before transitioning to efanesoctocog alfa in August 2024 and were followed up until March 2025 (observation period). All patients were inhibitor-negative at baseline. Primary efficacy was assessed through annualised bleeding rates (ABR). Secondary outcomes included joint bleeding frequency, inhibitor occurrence, and dosing patterns. Adherence to efanesoctocog alfa was monitored through infusion logs and pharmacy refill records. Patients were considered fully adherent if they received all scheduled doses throughout the observation period.

RESULTS: Twenty-one patients (95.5%) had severe disease. During the pre-switching period, half of the cohort experienced joint bleeding episodes; eight patients (36.4%) experienced one joint bleed, and three patients (13.6%) experienced two or more bleeds. The knee was the most frequently affected joint (54.6%), followed by the elbow (18.2%). The median prophylactic dose of efanesoctocog alfa was 57.1 IU/kg (IQR: 50.0-63.9). Patients received efanesoctocog alfa for eight months. During the observation period, 100% of patients achieved a zero ABR, with no reported treatment-emergent adverse events or discontinuations. All patients demonstrated 100% adherence to the prescribed once weekly efanesoctocog alfa regimen. No active inhibitors or treatment-emergent adverse events were observed.

CONCLUSIONS: Switching from efmoroctocog alfa to once weekly efanesoctocog alfa prophylaxis demonstrated highly effective bleeding prevention and a favourable safety profile in paediatric patients. These findings highlight efanesoctocog alfa's potential to improve adherence and long-term clinical outcomes in routine paediatric haemophilia A management, warranting further exploration through larger, prospective studies.

Keywords: Hemophilia A, Hemophilia B, inherited bleeding disorder, coagulation factor deficiency, pediatric hematology, factor replacement therapy

MENA-O-215

IL-6 and Ferritin Levels Predict the Severity of ICAHT post CAR T-cell Treatment with Axi-cel for B cell Lymphoma

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BACKGROUND: CAR T-cell therapy is a promising treatment for hematologic malignancies but is associated with toxicities. ctor cell-associated hematotoxicity syndrome (ICAHT) which is serious but incompletely understood complication of CAR T-cell therapy.

AIM: To evaluate the incidence and risk factors for ICAHT.

METHODS: A retrospective study of 85 B-cell lymphoma patients treated with Axi-cel CAR T-cell therapy evaluated demographics, survival, ICANS, CRS, and cytopenias, identifying key risk factors for severe ($\geq$grade 3) and prolonged (>30 days) cytopenias.

RESULTS: The cohort included male 53 (63%) and female 32 (37.6%) patients with a median age of 53 (IQR: 36-52) years.

The 12-month OS was 75%.CRS was observed in 90.7% of patients, Grade 1-2 (64.1%), Grade 3-4 (35.9.1%). ICANS occurred in 83.7% of cases (Grade 1-2) 65.3%, Grade 3-4 (34.7%). Thrombocytopenia was reported in 62.8% of patients at different grades (Grade 1-2 (50%), Grade 3-4 (50%) while neutropenia was seen in 54.7% (Grade 1-1(27.6%), Grade 3-4 (72.4%). Thrombocytopenia persisted for ≥ 30 days in 55.3% of cases and < 30 days in 44.7%. Neutropenia lasted ≥ 30 days in 52.9% and < 30 days in 47.1%. Anemia persists for ≥ 30 days was seen in 58.8% and < 30 days in 41.2% of patients. Risk factors for grade 3-4 thrombocytopenia included pre Axi-cel infusion IL-6 > 12 pg/mL (3-fold risk, $P = 0.026$), ferritin > 400 ng/mL (9-fold risk, $P < 0.001$), time from last chemotherapy to lymphodepletion < 45 days (5-fold risk, $P = 0.009$), bone marrow involvement (2-fold risk, $P = 0.05$), and pre-lymphodepletion thrombocytopenia (14-fold risk, $P < 0.001$).Bridging radiation therapy was protective against neutropenia (0.4-fold risk, $P = 0.03$).Prolonged cytopenias were associated with pre Axi-cel infusion IL-6 > 12 pg/mL (4-fold risk for anemia, $P = 0.003$), ferritin > 400 ng/mL (2.5-fold risk for anemia, $P = 0.037$; 4-fold risk for thrombocytopenia, $P = 0.008$), time from last chemotherapy to lymphodepletion < 45 days ($P = 0.04$), and high LDH > 400 ($P = 0.01$ for anemia).In multivariate analysis, IL-6 was the only significant factor for anemia (OR = 2.95, 95% CI 1.1-7.8, $P = 0.02$), and ferritin was the only significant factor for prolonged thrombocytopenia (OR = 3.5, 95% CI 1.3-9. 8, $P = 0.01$). For severe thrombocytopenia, significant factors included ferritin (OR = 8.5, 95% CI 1.9-37.5, $P = 0.004$), time from last-chemotherapy to lymphodepletion < 45 days (OR = 0.23, 95% CI 0.06-0.9, $P = 0.03$), and pre-lymphodepletion thrombocytopenia (OR = 0.1, 95% CI 0.01-0.5, $P = 0.006$).

CONCLUSION: CAR T-cell therapy is associated with significant cytopenias, with IL-6 and ferritin levels predicting anemia and thrombocytopenia, respectively. Recent chemotherapy and bone marrow involvement further increase the risk of cytopenias. These findings highlight the need for tailored monitoring and management strategies to mitigate hematologic toxicities in CAR T-cell therapy patients.

Keywords: Interleukin-6, ferritin, ICAHT, CAR T-cell therapy, Axi-cel, B-cell lymphoma, cytokinebiomarkers

MENA-O-217

Assessment of Variabilities in Clinical and Laboratory Features in Siblings with Sickle Cell Disease at Sultan Qaboos University Hospital

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BACKGROUND AND PURPOSE: Sickle cell anemia (SCA) is one of the most prevalent autosomal recessive genetic disorders worldwide. Similarly, in Oman, the disease is very prevalent as well. Although they share the same parental beta-globin genes, but there is a variation in behavior of SCA among siblings although they share the same parental beta-globin genes. The study aims to identify the clinical and laboratory variables that impact the variation in behavior of the disease in the siblings.

METHODOLOGY: We performed a retrospective analysis to address the laboratory and clinical findings of groups of sickle cell siblings seen in Sultan Qaboos University Hospital (SQUH). Accordingly, we studied 85 families which have more than one sibling known to have SCA. A Chi-square test had been used to test the consistency of clinical features, whereas intra-class correlation coefficient test had been used to assess the variability of laboratory features.

RESULTS AND DISCUSSION: Results of intra-class correlation coefficient test showed that there are significant variability and low consistency in laboratory features between siblings, in which correlation coefficient (r) of Mean Corpuscular Volume (MCV) was (r = 0.422; $P = .007$), Hemoglobin(Hb) was (r = 0.309; $P = .049$), Lactate Dehydrogenase (LDH) was (r = 0.368; $P = .023$), Hemoglobin S (HbS) was (r = 0.368 ; $P = .014$). since a correlation coefficient of these features is less than (0.5), so this indicates that there are low consistency and concordance in these variables among siblings. In other hand, results of Chi-square test in clinical features showed no significant variability among groups of siblings.

CONCLUSIONS: Our findings suggested that there is a variability in laboratory parameters in siblings although they share similar parental parental beta-globin genes, so further researches and studies needed to concern the factors which affect the values of laboratory parameters among siblings.

Keywords: Concordance, sickle cell diseas, siblings

MENA-O-221

Metformin versus Oxymetholone in Pediatric Patients with Fanconi Anemia

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BACKGROUND: Fanconi anemia (FA) is a rare inherited bone marrow failure syndrome with cancer susceptibility. Metformin is a potential candidate for treatment FA to ameliorate DNA damage and bone marrow failure.

AIM: To assess the efficacy and safety of metformin in improving hematological parameters in non-diabetic patients with FA.

METHODS: In a prospective interventional non-randomized controlled clinical trial [NCT06519786], 30 non-diabetic children and adolescents with FA and at least one cytopenia were recruited over a period of two years. Sixteen patients received oxymetholone (OXY), while 14 patients received

metformin (MET) [500 mg twice daily for patients < 10 years, and 1000 mg twice daily for patients > 10 years] for 6 months. Hematological response (HR) was assessed based on modified Myelodysplastic Syndrome International Working Group criteria.

RESULTS: The mean age was 10.5 ± 3.8 years for MET group and 9.8 ± 3.9 years in OXY group. Hematologic response was comparable in both groups including absolute neutrophil count (ANC), hemoglobin (Hb) and platelet counts (P = 0.738, 0.229 and 0.440 respectively). Four (30.7%) out of 13 evaluable patients (30.7%) achieved HR in MET arm (ANC n = 1, Hb n = 2, platelets n = 1), where ANC increased from 0.46 × 109/ul to 1.8 × 109/ul, the mean increase in Hb was 2.15 g/dl, and the magnitude of increase in platelet count of 66 × 109/ul. On the other hand, 9 patients (56.25%) in OXY arm achieved HR (ANC n = 4, Hb n = 6, platelets n = 4) with median (IQR) increase in ANC of 0.8 (0.6) x109/ul, a median (IQR) rise in Hb of 3.45 (2.3) g/dl, and a median (IQR) platelet increase of 47.5 (19.5) x109. Gastrointestinal symptoms were the most encountered adverse events in the MET group, most of them were grade 1 and presented in the first month of therapy: 38.5% had diarrhea, 38.5% vomiting, 30.8% abdominal pain, and 7.7% dyspepsia. Moreover, 46.2% and 38.5% suffered from loss of appetite and weight loss respectively. None of the patients developed hypoglycemia or metabolic acidosis. On the other hand, the majority of patients (75%) in OXY group suffered from elevated liver enzymes, and two male patients (12.5%) had signs of virilization.

CONCLUSION: Metformin was inferior to oxymetholone in achieving hematological response in patients with FA. Gastrointestinal side effects and weight loss were the most common toxicity with MET as opposed to elevated liver enzymes in OXY group.

Keywords: Fanconi anemia, pediatric hematology, Metformin, Oxymetholone, comparative therapy, bonemarrow failure

MENA-O-222

Clinical Spectrum of an Egyptian Cohort of Children with Myelodysplastic Syndrome

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BACKGROUND: Distinguishing inherited from acquired and reactive conditions predisposing to childhood MDS is challenging and directly impacts therapeutic options. An underlying genetic predisposition is increasingly being recognized.

AIM: To characterize clinical features and outcome of childhood MDS in a group of Egyptian children.

METHODS: We retrospectively analyzed data from 27 patients with MDS diagnosed over a period of 5 years between January 2020 and December 2024.

RESULTS: The study included 14 males and 13 females, with a mean age at presentation of 7.4 ± 5.0 years. Parental consanguinity was found in thirteen (44%) patients. All patients had single or multilineage cytopeniaat diagnosis. Other presenting manifestations included repeated infections (40.7%) or autoimmune manifestations (11%). One child presented with myeloid sarcoma and his bone marrow examination showed dysplastic features and positive trisomy 8. Fifteen patients (55.5%) had an underlying inheritedbone marrow failure syndrome (IBMFS): six with Fanconi (FA) anemia, 3 patients with dyskeratosis congenita (DC). Other IBMFS included GATA2 deficiency, Pearson syndrome, Schwachman-Diamond syndrome, Diamond-Blackfan anemia, and Noonan syndrome. Acquired causes were recognized in seven patients: 3 (11%) patients with autoimmune/autoinflammory diseases and three (11%) patients with primary immune deficiency, while one patient suffered from underlying chromosomal abnormality and global developmental delay who developed transient MDS related to antiepileptic medication. The underlying condition was not identified in five patients (18.5%). Three patients had cytogenetic abnormalities: FA with + 1, Noonan syndrome with monosomy 7, and MDS with trisomy 8. Four patients went on to develop AML with rapid progression. Overall, there were 6 mortalities: four with progressive AML, one with sepsis and one with transfusion related acute lung injury.

SUMMARY AND CONCLUSION: Unlike adult MDS, IBMFS represent the main cause of MDS in children. Clonal evolution and progression to AML remain a major cause of mortality, thus necessitating early therapeutic interventions.

Keywords: Myelodysplastic syndrome, pediatric hematology, clinical spectrum, Egyptian cohort, bonemarrow disorders, childhood MDS

MENA-O-225

High Levels of Serum and Cerebrospinal Fluid Interleukin-18 in CNS Hemophagocytic Lymphohistiocytosis and Other Inflammatory Demyelinating CNS Diseases

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BACKGROUND: Differentiating CNS-HLH, especially in the CNS-restricted form, from other inflammatory demyelinating diseases (IDD) of the CNS represents a diagnostic challenge owing to the overlapping clinical picture and neuroradiological findings, and to the lack of the proper diagnostic tools.

AIM: We investigated the role of interleukin-18 (IL-18) as a biomarker in differentiating CNS-HLH from other CNS-IDD.

METHODS: We measured serum and cerebrospinal fluid (CSF) levels of IL-18 in children with familial HLH and in patients with other CNS-IDD as confirmed on brain MRI images and compared them to 31 age-and sex-matched healthy controls.

RESULTS: Among the 26 enrolled subjects, 14 (48.2%) patients had familial HLH with CNS disease (group-1), while 12 patients suffered from other CNS-IDD (group-2)diagnosed as follows: two patients with each of acute necrotizing encephalopathy, multiple sclerosis, neuromyelitis optica, and clinically isolated syndrome, and one patient with each of acute disseminated encephalomyelitis, transverse myelitis, Anti-MOG syndrome, and Rasmussen encephalitis. The majority (10/14) of familial HLH patients carried mutations in PRF1 [6 (43%) patients] and UNC13D [4 (28.6%) patients] genes. Levels of IL-18 were significantly higher in the sera of both group-1 and group-2 as compared to healthy controls (median 40.6 and 37.4 ng/ml versus 6.8 ng/ml respectively, P=<0.001). On *post-hoc* analysis, both group 1 and group-2 had comparable median serum IL-18 levels ($P = 0.797$).Similarly, the median CSF IL-18 levels where comparable between group-1 and group-2 (53.3 versus 69.3 ng/ml, $P = 0.612$). Cerebrospinal fluid (CSF) IL-18 levels positively correlated with CSF protein (r = 0.508, $P = 0.022$), while this was not true for serum IL-18 (r=-0.195, $P = 0.329$).

CONCLUSION: Children with inflammatory CNS demyelination exhibit higher levels of serum IL-18 than healthy controls. Both serum and CSF levels of IL-18 were not able to differentiate CNS-HLH from other inflammatory demyelinating CNS disease.

Keywords: Interleukin-18, CNS hemophagocytic lymphohistiocytosis, inflammatory demyelinating CNS diseases, serum biomarkers, cerebrospinal fluid analysis, neuroinflammation

MENA-O-226

Interim Data from Phase 2 Study of Sutacimig for Prophylactic Treatment in Glanzmann Thrombasthenia

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BACKGROUND: Glanzmann thrombasthenia (GT) is a severe inherited bleeding disorder that disrupts platelet aggregation and healthy clot formation. Impairment in clotting leads to frequent bleeding events ranging from low-volume mucocutaneous bleeding, debilitating to life-threatening hemorrhages, contributing to iron deficiency anemia and impaired quality of life. Sutacimig is an investigational bispecific antibody designed to restore healthy clot formation in GT. Sutacimig's dual mechanism of action achieves both accumulation of endogenous FVIIa and targeting of activated platelets via TLT-1 receptor binding, driving localized thrombin generation and fibrin formation at the surface of activated platelets.

AIMS: This Phase 2 study evaluates the safety, tolerability, pharmacokinetcs (PK), pharmacodynamics (PD), and efcacy of sutacimig in GT.

METHODS: Phase 1 assessed single ascending doses (0.2-1.25 mg/kg). The ongoing Phase 2 study involves multple-ascending doses (0.3-0.9 mg/kg every two weeks) in GT partcipants afer a run-in period to document bleeding episodes. Key endpoints include safety, PK, PD, and assessment of bleeding events.

RESULTS: Phase 1 ($n = 7$) data demonstrated that sutacimig was well tolerated and achieved a signifcant reducton in ATBR, with a mean reducton exceeding 50% during the 30-day follow-up from baseline. Preliminary Phase 2 data indicate consistency with the reducton observed in Phase 1. Contnued evaluaton of sutacimig, including data antcipated from Phase 2 Parts B, expected mid-2025, will further elucidate sutacimig's profle. PD data demonstrated a dose-dependent increase in endogenous total FVII (a) and FVIIa actvity, along with improved thrombin generaton comparable to standard rFVIIa dosing, indicating efectve coagulaton actvaton. The PK profle supports a convenient, infrequent dosing schedule.

CONCLUSIONS: Interim results show sutacimig signifcantly reduces frequency of bleeding events with a favorable safety profle, supportng its potental as a novel prophylactc treatment for GT. Sutacimig's subcutaneous dosing ofers a proactive alternatve to reactve GT care, addressing a critcal unmet need.

Keywords: Glanzmann thrombasthenia, Sutacimig, prophylactic treatment, phase 2 clinical trial, inherited platelet disorder, hemostasis therapy

POSTER PRESENTATIONS

MENA-P-105

Hemolytic Disease of the Newborn and Neonatal Transfusion

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Hemolytic Disease of the Newborn (HDN) is a serious immunohematologic condition caused by maternal alloantibodies that cross the placenta and destroy fetal red blood cells, most commonly due to Rh incompatibility. Advances in immunoprophylaxis and prenatal care have significantly reduced the incidence of Rh-mediated HDN, yet cases due to other alloantibodies (e.g., Kell, Duffy) continue to present challenges in neonatal care.

OBJECTIVE: This study aims to review the pathophysiology, diagnostic approaches, and management of HDN, with a focus on the role of neonatal transfusion in preventing and treating anemia and hyperbilirubinemia in affected newborns.

METHODS: A retrospective review of clinical cases from 2015 to 2024 was conducted at a tertiary care center. Data included maternal antibody screening, fetal monitoring outcomes, transfusion interventions, and neonatal morbidity rates. Literature from PubMed and WHO databases was also reviewed.

RESULTS: Out of 72 HDN cases identified, 25% required intrauterine transfusion, and 40% underwent exchange transfusion postnatally. The use of antigen-negative, irradiated,

and leukoreduced red blood cells was associated with reduced incidence of transfusion reactions. Early identification and aggressive phototherapy reduced the need for invasive transfusion in mild to moderate cases.

CONCLUSION: Neonatal transfusion remains a critical component in the management of HDN, especially in severe cases with anemia and hyperbilirubinemia. Continued emphasis on antenatal antibody screening and appropriate blood product selection is essential for optimizing neonatal outcomes.

Keywords: Neonatal intensive care, Exchange transfusion, Neonatal outcomes

MENA-P-107

Multiple Myeloma

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Multiple Myeloma (MM) is a clonal plasma cell malignancy characterized by excessive production of monoclonal immunoglobulins and infiltration of the bone marrow. It accounts for approximately 10% of hematologic cancers and presents with bone pain, anemia, renal impairment, and hypercalcemia. Despite therapeutic advances, MM remains incurable, with inevitable relapse in most patients.

OBJECTIVE: This study aims to evaluate the current diagnostic tools, therapeutic approaches, and emerging challenges in the clinical management of multiple myeloma, with a focus on personalized treatment strategies and outcomes.

METHODS: A retrospective review was conducted on 120 patients diagnosed with MM between 2018 and 2024 at a tertiary care center. Data analyzed included clinical presentation, laboratory findings, treatment regimens, response rates, and progression-free survival (PFS). Diagnostic strategies, including serum free light chain assay, bone marrow biopsy, and imaging studies, were assessed for diagnostic accuracy.

RESULTS: Initial diagnosis was most commonly prompted by anemia (65%) and bone pain (55%). First-line therapy included bortezomib-based regimens in 78% of patients, with an overall response rate of 82%. Autologous stem cell transplantation was performed in 36% of eligible patients. Median PFS was 28 months. Relapse occurred in 48% of patients during the follow-up period, with resistance noted in those with high-risk cytogenetics.

CONCLUSION: Multiple myeloma remains a complex and evolving disease. While novel therapies have improved response rates and survival, relapse and resistance continue to challenge long-term management. Tailored treatment approaches and incorporation of next-generation therapies are essential to improve outcomes.

Keywords: Anemia, immunoglobulins, involvement, multiple myeloma, monoclonal, m-protein, plasma cell dyscrasia

MENA-P-109

Diffuse Large B-Cell Lymphoma Development Post-COVID-19 Vaccination in a Sickle Cell Patient: A Complex Clinical Scenario

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BACKGROUND: Unlike the general population, sickle cell disease (SCD) patients have a 2-to 11-fold higher risk of hematological malignancies including lymphoma.

CASE PRESENTATION: In the era of coronavirus disease 2019 (COIVD-19), a 26-year-old non-smoker overweight male diagnosed as SS genotype SCD with hyperuricemia and iron overload, presented with cervical, axillary lymphadenopathy and lower neck mass with compressive chest symptoms approximately six weeks after encountering COIVD-19 infection that was preceded by receiving a single dose of BNT162b2, COVID-19 mRNA vaccine. A cervical lymph node True-cut biopsy with immunohistochemistry revealed diffuse large B-cell lymphoma (DLBCL); germinal center B-cell (GCB-subtype). Pan-CT scan and BMB revealed DLBCL (Stage IIx) with a good R-IPI.

MANAGEMENT: The patient was initially managed by blood transfusion, IV methylprednisolone as a pre-phase, followed by six cycles of R-CHOP-21 without administration of granulocyte colony-stimulating factor (G-CSF) to avoid precipitation of sickle cell crisis. During immunochemotherapy hydroxyurea was held to avoid cytopenias and top-up blood transfusion was used as an alternative management for SCD whenever indicated. Outcome: The patient is still on follow-up after three years of his last chemotherapy, with good health condition and persistent complete remission, as confirmed by PET/CT restaging at the end of successful treatment despite the diagnostic and therapeutic challenges due to the overlapping initial findings and lack of hydroxyurea or G-CSF administration throughout immunochemotherapy. Interestingly, vaso-occlusive crisis (VOCs) did not occur during R-CHOP. However, upon follow up in the last three years, he had recurrent hospitalizations due to acute chest syndrome, and painful VOCs.

DISCUSSION: In hemoglobinopathies, multiple blood transfusions increase the risk of alloimmunization and exposure to blood-borne viruses with increased risk for malignancy. Moreover, iron overload represents a chronic proinflammatory state that leads to iron-induced carcinogenesis. Also, baseline hyperuricemia was reported to be an early feature of undiagnosed hematological malignancy. Post-COVID vaccine DLBCL including (GCB subtype) was reported in three patients, even after a single dose of BNT162b2-vaccine. Also, 1ry lymphomas was reported after SARSCoV-2 (a potentially oncogenic virus).

CONCLUSION: To the best of our knowledge, we are the first to report DLBCL/GCB in young adult male with SCD

that developed after BNT162b2 vaccination and COVID-19 infection. This sequence of events raises questions about a potential causal relationship. However, the simple association is still a possible scenario. Thus, further research using molecular profiling for SARS-CoV-2 spike protein and nucleocapsid protein, will be helpful in discrimination.

Keywords: Sickle cell disease (SCD), COVID-19 infection, COVID-19 vaccination, Diffuse large B-cell lymphoma (DLBCL)

MENA-P-114

Epidemiology of Vascular Thrombosis in the Eastern Province of Saudi Arabia: A Single Center Study and Comparison with National Data

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BACKGROUND: Despite the growing evidence on the thrombophilia in Saudi population along its diverse regions, there have been no studies on thrombotic events in the Eastern Province of Saudi Arabia. This single-center study aimed to investigate the prevalence of venous thromboembolism (VTE) in a hematology clinic in the Eastern Province of Saudi Arabia between January 2015 and May 2023. The objective of this study was to investigate the clinical characteristics of VTE and compare them with national data.

METHODS: This is a retrospective, observational, single-center study conducted in the Eastern Province from January 2015 to May 2023. After applying the inclusion and exclusion criteria and the prevalence rate calculation an analysis of n = 170 patients was conducted to compare the epidemiological results of the current study with national data published in other provinces of Saudi Arabia. Data collected included demographics, comorbidities and location with thrombosis recurrence.

RESULTS: The prevalence rate of VTE in this cohort was 3.16%. Women made up 70% of the population. About 80.5% of cases were obese or overweight, 37.6% of cases had comorbidities that may increase the risk of thrombosis, and 12.9% of cases were associated with smoking. The most common site of VTE (56.5%) and the site of higher recurrence of VTE (21.8%) was the lower extremities. In addition, recurrent VTE was observed in 28.2% of cases, reflecting a significantly higher recurrence rate compared with other national studies ($P = 0.001$). Compared with other national statistics, the most significant risk factors for thrombosis in the Eastern Province were smoking, obesity and family history ($P = 0.000$). Compared with national statistics, this study demonstrated significantly higher rates of VTE in pregnancy, patients on hormonal therapy, and patients with rheumatological/autoimmune diseases ($P = 0.001$).

CONCLUSION: The incidence of VTE can be reduced by changing lifestyles and creating educational programs to educate people about the dangers of obesity and smoking.

Keywords: Venous thromboembolism, pulmonary embolism, epidemiology

MENA-P-122

Netrin-1 and Clusterin: Valuable Markers for Early Detection of Kidney Injury in Children with Transfusion-dependent Thalassemia

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BACKGROUND: Advances in the management of transfusion-dependent thalassemia (TDT) have led to improved patient outcomes but also revealed new complications, including renal abnormalities. Clinical studies have reported mild tubular dysfunction and altered glomerular filtration rate (GFR) in children with TDT. Additionally, progressive increases in serum creatinine have been observed following exposure to certain iron chelators.

AIM: To detect early renal tubular dysfunction as a complication of transfusion-dependent thalassemia.

METHODS: A total of 100 children were enrolled in this study: 50 with TDT and 50 age- and gender-matched healthy controls. All participants underwent thorough medical history-taking, clinical examination, and laboratory investigations including complete blood count, serum creatinine, blood urea, serum ferritin, urinary creatinine, urinary albumin, and measurement of serum Netrin-1 (NTN-1) and Clusterin (CLU) levels.

RESULTS: Serum Netrin-1 levels were significantly elevated in TDT patients compared to controls (68.6 ± 28 pg/ml vs. 14.3 ± 2.5 pg/ml; $P < 0.001$), demonstrating 98.00% sensitivity and 84.00% specificity for detecting renal injury. Similarly, serum Clusterin levels were significantly higher in TDT patients (101.94 ± 37 ng/ml) than in controls (16.58 ± 6.7 ng/ml; $P < 0.001$), with 88.00% sensitivity and 86.00% specificity. Notably, all participants had normal serum creatinine, urea, uric acid, and urine albumin-creatinine ratios.

CONCLUSION: Serum Netrin-1 and Clusterin are promising early biomarkers for renal tubular injury in children with transfusion-dependent thalassemia, even before conventional kidney function tests show abnormalities.

Keywords: Netrin-1, Clusterin, kidney injury, transfusion-dependent thalassemia, early biomarkers

MENA-P-129

Neutrophil CD64: early Predictor for Sepsis in Cancer Patients during Febrile Neutropenia

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BACKGROUND: Febrile neutropenia (FN) is a common treatment-related complication in pediatric cancer patients with substantial morbidities and mortalities. Previous studies reported that neutrophil CD64 (n CD64) had higher diagnostic accuracy for infection with high sensitivity and specificity in neonates, pediatrics and adult patients.

AIM: To evaluate the usefulness of neutrophil CD64 expression as an early diagnostic marker of sepsis in children with cancer during episodes of FN.

METHODS: A case control study was conducted on 100 children (50 patients with hematological malignancies and febrile neutropenia, 25 patients with hematological malignancies without febrile neutropenia and 25 apparently healthy children as a control group). Routine laboratory investigations including blood culture were done in patients with cancer according to our local standards. Procalcitonin level and Neutrophil CD64 expression by flow cytometry were measured for all study participants.

RESULTS: n CD64 expression was significantly higher in patients with cancer and FN compared to other groups ($P > 0.001$). At a cutoff value of $\geq$ 17.82%, serum n CD64 had 94% sensitivity and 72% specificity. n CD64 expression level was negatively correlated to absolute neutrophil count (ANC) during episode of FN (r= (-0.359, $P = 0.01$). A positive correlation was found between nCD64 expression and both of CRP and procalcitonin. Blood culture was positive in 54% in patients with cancer and FN. The most common isolated organism was Klebsiella pneumonia. Among patients with cancer and FN, n CD64 expression level was significantly higher in patients with positive blood culture compared to those with negative cultures.

CONCLUSION: Neutrophil CD64 expression seems to be a reliable marker in early detection of sepsis during episodes of febrile neutropenia in children with haematological malignancies.

Keywords: Neutrophil, sepsis, febrile neutropenia

MENA-P-133

Assessment of Audiometric Abnormalities in Children with Gaucher Disease

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NEUTROPHIL CD64: Early predictor for sepsis in cancer patients during febrile neutropenia.

BACKGROUND: Gaucher disease (CD) is the most common autosomal recessive lysosornal storage disorder characterized by insufficiency Of the enzyme glucocerebrosidase. Although sensorineural hearing loss has been described due to neurological involvement of the auditory pathway in types II and III GD disease. All patients with GD should undergo audiologic;al assessment to screen for sensorineural hearing loss as part of their multidisciplinary management. Aim: to asses and describe the audiological affection in patient with gaucher disease trying to improve quality of life in patients witn GD.

PATIENTS AND METHODS: This cross-sectional study was conducted at gaucher clinic at hematology unit at Pediatrics Department and Audio-vestibular unit at E.N.T department. Faculty of Medicine, Zagazig University. The study included 24 children With Gaucher disease types I and II. they were 13 females (54,2%) and 11 males (45.8%). with a median age of 6.5 years, ranging from 2-5 to 18 years. Pure Tone Audiometry, Tympanometry, Otoacoustic Emissions, Auditory Brainstem Response (ABR) were used to test for audiological affection in GD patients.

RESULTS: A statistically significant' positive correlation was found between level Of glucocerebrosidase enzyme and result Of acoustic emission at 3000Hz and 4000 Hz, also, There is A statistically significant negative correlation between level Of enzyme and latency (V 40) and interpeak (IIIV 40).

CONCLUSION: The study highlighted significant audiological and neurological implications associated with the disease, particularly in patients with Type III GO. The auditory brainstem response (ABR) analysis revealed latency delays, especially in lower enzyme levels, pointing to retrocochlear involvement and compromised auditory pathway integrity.

Keywords: Gaucher disease, audiological affection, tympanometry, otoacoustic emissions. auditory brainstem response

MENA-P-138

Awareness of Iron Deficiency Anemia among School-aged Children and Adolescents in Madinah, KSA

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BACKGROUND AND AIM: Iron deficiency anemia (IDA) is a common dietary condition that can cause serious medical conditions, especially in teenagers. It is crucial to understand the awareness levels surrounding this condition to implement effective educational and prevention strategies. This study aims to evaluate the awareness about IDA among students aged 9-18 years in Madina, focusing on their knowledge of its symptoms, causes, and preventive measures. The findings of this study can help to improve understanding, prevention and management of this condition within the community.

METHODS: In a cross-sectional study conducted among school

aged students in Madinah we assessed awareness of IDA. A structured questionnaire collected data on demographics, dietary habits, physical activity, and IDA knowledge. Data is cleaned in Excel and analyzed via IBM SPSS 29.0.0.

RESULTS: In this study (n = 109), most participants were females (n = 65, 59.6%), Saudis (n = 93, 85.3%), and aged 13-18 years (n = 64, 58.7%). IDA was reported in 73 participants (67.0%). High awareness of IDA was observed in 64.2% (n = 70), while 35.8% (n = 39) had low awareness. Awareness was significantly associated with age (P < 0.001), gender (higher in females, n = 53, 81.5%; P < 0.001), nationality (non-Saudis n = 14, 87.5%; P = 0.035), BMI (normal n = 38, 80.9%; obese n = 4, 100.0%; P < 0.001) and fast food intake frequency (n = 20, 95.2%; P = 0.002). However, income level (P = 0.422), history of anemia (P = 0.960), and chronic disease history (P = 0.491) did not significantly impact awareness. Only 31.2% took iron supplements, and just 3.7% identified worm infections as a cause.

CONCLUSION: The current study revealed that there is relatively high awareness of IDA among adolescents in Madinah, especially among older females with normal or higher BMI. However, practical knowledge and preventive behaviors remain limited. Significant associations were found between awareness and age, gender, BMI, and fast-food habits, highlighting the need for targeted, behavior-focused educational interventions across schools and communities.

Keywords: Iron deficiency, adolescents, awareness, dietary habits, anemia prevention

MENA-P-143

Frequency of Anemia and Possible Risk Factors among Sudanese Children with End Stage of Renal Disease

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Anemia is a common Feature of chronic kidney disease, but the management of anemia in children is complex. Erythropoietin and Supplemental iron are used to maintain hemoglobin levels. The aim of this study to determine the Frequency of anemia and possible Risk Factors Among children with End stage renal disease.

METHODS: A total of 96 children, 61 males (63.5%) and 35 Females (36.5%), were attended at hemodialysis units in Khartoum state were enrolled in the study and Frequency of anemia was estimated by analyzing CBC on blood counter (sysmex). The concentration of iron profile, C-reactive protein and parathyroid hormone was measured using COBAS INTEGRA 400 PLUS and COBAS E411.

RESULTS: 99% of children were anemic, 4.17% of the them were suffering from iron deficiency anemia and also there is other causes contributing to anemia in ESRD patients which are inflammation and hyperparathyroidism.

CONCLUSION: The prevalence of anemia in children on hemodialysis in Sudan appears to be higher than that reported in other studies in spite of extensive use of rHuEPO and iron supplementation.

Keywords: RBC disorder, anemia, renal disease

MENA-P-148

Investigating Stroke Risk in Microcytic Hypochromic Anemia through the Role of Red Blood Cell Size and Aggregation

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BACKGROUND: Microcytic hypochromic anemia is traditionally linked to reduced red blood cell (RBC) aggregation due to smaller cell size and lower hemoglobin. However, emerging data suggest plasma-phase factors may significantly influence aggregation, potentially increasing thrombotic risk. This study explores the paradox of enhanced RBC aggregation in such patients.

AIMS: To assess RBC aggregation behavior in microcytic hypochromic anemia and evaluate its relationship with RBC indices. The study also aims to determine whether increased aggregation may represent an underrecognized risk factor for thrombosis.

METHODS: A case-control study was conducted on 102 patients diagnosed with microcytic hypochromic anemia, compared to healthy controls. CBC parameters were measured, and RBC aggregation was analyzed using the Laser-Assisted Optical Rotational Cell Analyzer (LORRCA). Key parameters included Aggregation Index (AI%), kinetic indices (T1/2, T1/2 ER, TR, TR SD), and amplitude (AMP, AMP SD).

RESULTS: Despite significantly lower MCV, MCH, Hb, and HCT, the anemic group showed elevated AI% (74.9% vs. 67.1%). Kinetic parameters were prolonged, indicating delayed and disorganized aggregation. While AMP was slightly decreased, its variability (AMP SD) increased, suggesting inconsistent aggregate strength. RDW was also elevated, indicating anisocytosis. These findings suggest plasma components—rather than RBC morphology—may play a dominant role in aggregation behavior.

CONCLUSIONS: This study challenges the conventional view that RBC aggregation is reduced in microcytic states. Instead, results reveal heightened aggregation likely mediated by plasma-phase factors such as fibrinogen. The use of LORRCA enabled detailed real-time analysis, supporting the presence of a hyperaggregable state in microcytic hypochromic anemia. These findings underscore the need to consider both cellular and plasma elements when assessing thrombotic risk in anemic patients. Future studies should investigate inflammatory markers and plasma protein levels to better understand the mechanisms behind this aggregation pattern.

Keywords: Microcytic anemia, Hypochromic anemia, red blood cell

MENA-P-152

Investigating the Role of B-cell Activating Factor (BAFF) in Multi-transfused Children with Thalassemia

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BACKGROUND: Frequent blood transfusions in thalassemia patients can lead to alloimmunization, with B-cell activating factor (BAFF) playing a crucial role in this process. We studied BAFF levels in pediatric thalassemia patients with multiple transfusions to understand its relationship with red blood cell alloimmunization.

METHODS: A total of 150 participants, including 100 thalassemia pediatric patients and 50 healthy children, were enrolled. Venous blood samples were drawn for CBC, blood grouping, and BAFF measurement via ELISA at baseline and 12 months later. Antibody screening and identification were conducted using column agglutination technology at both baseline and follow-up.

RESULTS: A significant positive correlation was observed between BAFF and the BAFF-to-ALC ratio at both time points. ALC was notably higher in splenectomized patients and those without baseline RBC antibodies. Furthermore, elevated BAFF and BAFF-to-ALC ratio were independently predictive of baseline RBC antibodies, while higher ALC levels were associated with a decreased risk of antibody development.

CONCLUSION: Our study shows that BAFF levels are elevated in thalassemic patients, especially those with RBC alloantibodies, and increase over time. These findings indicate that BAFF may be a useful biomarker for predicting red blood cell alloantibody formation, with potential clinical benefits.

Keywords: B-cell activating factor, thalassemia, transfusion, allo-antibodies

MENA-P-154

The Role of Radiotherapy in Improving Outcomes for Neuroblastoma Patients Undergoing Autologous Bone Marrow Transplantation

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BACKGROUND: Neuroblastoma remains a significant clinical challenge, particularly in high-risk cases. Bone marrow transplantation and radiotherapy are important treatment strategies, but their combined effect on prognosis and survival requires further study.

AIM: This study aimed to examine the outcomes of neuroblastoma patients who underwent autologous bone marrow transplantation followed by radiotherapy.

METHODS: This study enrolled 31 pediatric neuroblastoma patients and assessed the impact of BMT and radiotherapy on treatment outcomes.

RESULTS: The male-to-female ratio was 2.1:1, and most patients were preschool-aged. Bone marrow metastasis occurred in 67.7% of cases. BMT was associated with improved outcomes, with a lower relapse rate (46.7%) compared to non-transplanted patients (87.5%). The difference in outcomes between the two groups was statistically significant ($P = 0.05$). The addition of radiotherapy to BMT did not significantly impact outcomes in this study.

CONCLUSION: Autologous bone marrow transplantation for high-risk neuroblastoma is associated with improved patient outcomes, including increased survival and reduced recurrence rates. In this context, radiotherapy does not seem to have a significant impact on patient prognosis or survival.

Keywords: Neuroblastoma, Autologous bone marrow transplantation, radiotherapy

MENA-P-160

The Role of Radiotherapy in Improving Outcomes for Neuroblastoma Patients Undergoing Autologous Bone Marrow

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BACKGROUND: The goal of this study was to create and validate a Sanger sequencing-based assay using the SeqStudio Genetic Analyzer to detect mutations in the HFE gene, specifically p. C282Y, p. H63D, and p. Glu 277 Lys. These mutations are linked to hereditary hemochromatosis (HH), an autosomal recessive condition characterized by iron overload. Prior to in-house validation, the HFE gene mutation assay was sent to Bio Scientia (Germany) for qPCR analysis. While this method reliably detects common mutations, it frequently misses rare variant mutations, indicating the need for a more efficient and cost-effective method of detecting these mutations. The development of this assay provided a promising solution by reducing TAT from three weeks to six days, making it cost-effective, and allowing for novel mutation detection through broad screening.

METHODS: Genomic DNA was extracted from peripheral blood. The HFE gene (NM_000410.4) was analyzed by PCR amplification of exons 2 and 4 using a VeritiPro ™ Thermal Cycler (Applied Biosystems, USA). Sanger sequencing was performed using the BigDye ™ Terminator v3.1 Cycle Sequencing Kit and sequenced in the SeqStudio Genetic Analyzer (Applied Biosystems, USA). The patient's sequence was compared to the reference HFE gene sequence.

RESULTS: 134 patient samples were analyzed for three variants in the HFE gene: c.187C>G (p. His 63Asp), c.845G>A (p. Cys 282Tyr/C282Y), and c.829G>A (p. Glu 277 Lys) using Sanger sequencing. Out of 134 samples, 59 (44%) had one or more HFE gene mutations, while 75 (56%) tested negative for all HFE gene mutations. The distribution of mutations among positive samples is shown below: c.187C>G (p. His63Asp) heterozygous 37 cases & homozygous 8 cases; C282Y heterozygous (c.845G>A): 1 case; C282Y homozygous: 1 case; c.829G>A (p. Glu277 Lys)

heterozygous 8 cases; while compound heterozygous [c.187C>G (p. His63Asp), c.845G>A (p. C282Y)] one case & (p. His63Asp-p. Glu277 Lys) three cases. The presence of homozygous C282Y—the most clinically significant genotype in hereditary hemochromatosis was uncommon in this cohort.

CONCLUSION: The study developed and validated a Sanger sequencing-based assay to detect HH-associated HFE gene mutations (c.187C>G, c.845G>A, and c.829G>A). This study shows that Sanger sequencing is a reliable, cost-effective in-house method for mutation detection with a wider mutation detection capacity and faster turnaround time than outsourced qPCR testing. Due to the rare p. C282Y mutation and common p. H63D mutation in Saudis, more research is needed. A larger sample size of clinically well-characterized Saudi patients with HH may reveal the role of p. H63D or novel HFE mutations.

Keywords: HFE mutations, hemochromatosis, HFE gene

MENA-P-164

Brain Iron Accumulation With Neurodegeneration

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Neuroferritinopathy is a rare form of neurodegeneration with brain iron accumulation (NBIA), caused by mutations in the FTL gene. We present the case of a 35-year-old woman initially diagnosed with non-anemic iron deficiency syndrome (NAIDS), who developed acute neuropsychiatric symptoms following intravenous (IV) iron therapy. Magnetic resonance imaging (MRI) revealed iron deposition in basal ganglia and cerebellar nuclei, and genetic testing confirmed FTL mutation. This case underscores the importance of considering NBIA in patients with atypical neurological symptoms post iron infusion.

BACKGROUND: Non-Anemic Iron Deficiency Syndrome (NAIDS) refers to a state of iron depletion with normal hemoglobin levels, often resulting in symptoms like fatigue, cognitive impairment, and dyspnea. While oral iron is first-line therapy, IV iron is used when intolerance occurs. However, IV iron can rarely lead to hypersensitivity reactions or worsen undiagnosed neurodegenerative conditions.

CASE PRESENTATION: A 35-year-old female presented with fatigue, poor concentration, dyspnea, and menstrual irregularities. Her only medical history was mild allergic rhinitis. Laboratory investigations revealed microcytosis (MCV 75 fL) and severe iron deficiency (serum ferritin 2.5 ng/mL) with a normal hemoglobin of 12.1 g/dL. She was diagnosed with NAIDS and prescribed oral iron, which she could not tolerate. She received two IV iron sucrose infusions (200 mg each). Five days after the infusions, she developed diffuse myalgia, perioral numbness, tremors, and progressed to seizures, dystonia, and ataxia. Laboratory workup was unremarkable. Brain MRI showed hypointense signals on T2 and T2*-weighted imaging in the globus pallidus, substantia nigra, caudate, and cerebellar nuclei, consistent with iron deposition. Genetic testing revealed an FTL gene mutation, confirming neuroferritinopathy.

DISCUSSION: Neuroferritinopathy is a rare autosomal dominant NBIA subtype marked by progressive movement disorders and cognitive decline. The FTL mutation causes aberrant iron storage and neuronal damage. Although IV iron is generally safe, this case suggests that iron infusion may trigger or exacerbate latent neurodegenerative processes in genetically predisposed individuals. This case also illustrates the importance of considering NBIA in patients who develop new neurological symptoms after iron therapy, especially when imaging shows basal ganglia iron deposition.

CONCLUSION: In patients with unexplained extrapyramidal symptoms following IV iron therapy, clinicians should consider underlying NBIA, particularly neuroferritinopathy. Early recognition through neuroimaging and genetic testing can prevent misdiagnosis and guide future management. Publication Consent : obtained from patient.

Keywords: Neuroferritinopathy, NABIA, intravenous Iron, FTL gene and movement disorders

MENA-P-170

CLPB-related 3-Methylglutaconic Aciduria Type VII: A Crucial Consideration in Severe Congenital Neutropenia with Epilepsy

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CLPB (caseinolytic peptidase B) deficiency is a rare mitochondrial disorder combining neurologic dysfunction and congenital neutropenia. We present a diagnostically challenging case of a 4-month-old girl initially labeled as hypoxic-ischemic encephalopathy (HIE), with seizures and persistent neutropenia. Whole exome sequencing revealed a homozygous pathogenic CLPB variant (c.1760A>G; p. Tyr 587Cys), confirming 3-methylglutaconic aciduria type VII (MGCA7). This case emphasizes the importance of recognizing CLPB-related disorders in infants with overlapping neurologic and hematologic findings.

INTRODUCTION: CLPB deficiency is an autosomal recessive mitochondrial chaperonopathy affecting stress-induced protein disaggregation. It presents as a spectrum ranging from severe infantile encephalopathy with neutropenia to milder isolated neutropenia. Severe forms resemble HIE but involve congenital neutropenia, which may not respond fully to G-CSF therapy. Early recognition is essential for appropriate management and genetic counseling.

CASE PRESENTATION: A 4-month-old Yemeni girl was referred for persistent neutropenia and developmental delay. Born at term following presumed perinatal asphyxia, she was initially diagnosed with HIE. Her clinical course was marked by intractable epilepsy, global developmental delay, hypotonia, left-sided hemiparesis, and oxygen dependency. MRI showed

bilateral thalamic and basal ganglia hyperintensities, corpus callosum thinning, and cerebral atrophy. EEG demonstrated modified hypsarrhythmia. Her ANC ranged from 0.03-0.16 × 10□/L, peaking transiently at 0.71 × 10□/L following G-CSF. Bone marrow was non-specific. Ferritin was 5266, and EBV PCR was positive. Whole exome sequencing revealed a homozygous CLPB mutation (c.1760A>G; p. Tyr 587Cys), consistent with MGCA7.

DISCUSSION: This case represents a severe MGCA7 phenotype, reinforcing the link between CLPB mutations, neurologic dysfunction, and neutropenia. The c.1760A>G variant is documented as pathogenic. Incomplete response to G-CSF is consistent with published cases. CLPB testing is critical in infants with combined neurologic and hematologic findings to prevent delays in diagnosis and care.

CONCLUSION: CLPB deficiency should be included in the differential diagnosis for infants with unexplained encephalopathy and neutropenia. Early genetic testing avoids misdiagnosis, facilitates proper counseling, and improves family planning options.

Keywords: CLPB deficiency, Aciduria, Congenital Neutropenia

MENA-P-178

Clinical and Laboratory Outcomes of First and Second Episodes of COVID-19 in Patients with Sickle Cell Disease: A Retrospective Comparative Cohort Study

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BACKGROUND: Sickle cell disease (SCD) is an inherited hemoglobinopathy marked by chronic hemolysis, vaso-occlusive crises, and heightened susceptibility to infections. Individuals with SCD are particularly vulnerable to severe complications from COVID-19 due to their immunocompromised state. While the effects of an initial COVID-19 infection in SCD patients have been explored, limited data exist regarding reinfection outcomes.

OBJECTIVE: This study aims to compare the clinical, laboratory, and outcome parameters of SCD patients during their first and second episodes of COVID-19 infection.

MATERIALS AND METHODS: A retrospective cohort study was conducted at Sultan Qaboos University Hospital (SQUH) from January 2020 to December 2023. Patients with confirmed SCD and at least one documented episode of COVID-19 were included. Clinical symptoms, laboratory parameters, treatment details, and outcomes were analyzed using descriptive statistics and comparative analysis. Differences between the first and second infections were assessed using Chi-square and Student's t-tests, with statistical significance set at $P < 0.05$.

RESULTS: Among 265 SCD patients, a total of 27 SCD patients who experienced two episodes of COVID-19 were included in the study. The median age was 30.6 years, with a male-to-female ratio of 10:17. Hospitalization rates were higher during the second infection (92.6% vs. 70%, $P = 0.07$), but disease severity was milder, with fewer ICU admissions (0% vs. 3.7%, $P = 0.22$) and reduced ventilatory support requirements (0% vs. 7.4%, $P = 0.43$). Hemoglobin levels declined significantly in both episodes ($P < 0.01$), while inflammatory markers remained elevated. Despite increased morbidity, there were no reported deaths.

CONCLUSION: SCD patients with COVID-19 reinfection experienced higher hospitalization rates but a less severe disease course, potentially due to immune adaptation or improved management strategies. These findings highlight the need for continued surveillance, early intervention, and further research to understand long-term outcomes in SCD patients with recurrent COVID-19 infections.

Keywords: COVID-19, hematological changes, immune adaptation, reinfection, retrospective cohort study, sickle cell disease

MENA-P-188

Assessment of Hemophilia Severity and Associated Clinical Outcomes: Experience from Pakistan

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BACKGROUND: Hemophilia is a rare bleeding disorder which results from a decreased level of specific clotting factors, mainly factor VIII (Hemophilia A) or factor IX (Hemophilia B).The clinical severity of hemophilia is directly related to the concentration of these factors in the blood. The absence of a national registry, coupled with limited access to diagnostic facilities and a lack of public awareness, contributes to the underestimation of hemophilia in Pakistan.

AIMS: This study aims to describe the hemophilia spectrum, along with its clinical consequences, in a resource-limited region of Pakistan, utilizing an underdiagnosed multidisciplinary approach.

METHODS: A descriptive retrospective study was performed at the Fatmeed Foundation (hemophilia treatment centers in Pakistan). Patients diagnosed with hemophilia A or B from 2019 to 2024 ($N = 90$) were included. The collected information comprised age, sex, factor VIII/IX quantification classifying them into different severities, bleeding episodes, longitudinal study of arthropathy, chronic joint changes, Hepatitis and HIV serology. Participants' clinical outcomes were evaluated based on the severity of the disease and the resources available.

RESULTS: Among the study population of 90,94.4% were males and 5.5% were females. In our study cohort 88% had hemophilia A, and 12% had hemophilia B. According to the severity classification, 43.3% of the patients had severe hemophilia, 37.7% had moderate hemophilia, and 18.8% had mild hemophilia. 13.3% of the patients had hemophiliac arthropathy, primarily those with severe degree of hemophilia. In addition, 14.4% of patients had substantial impairment and chronic pain along with recurrent episodes of bleeding. Due

to the restricted availability of factor concentrates, which were mostly used for severe bleeding episodes, on-demand therapies were given. As a result, fresh frozen plasma transfusions were given to numerous patients. Serological evaluations revealed that 4.4% of patients were positive for Hepatitis C.

CONCLUSION: Pakistan's low-income setting sharpens the focus on hemophilia's features, such as high incidence of severe disease, late diagnosis, and grave clinical consequences stemming from inadequate access to comprehensive care specially preventative treatment. This highlights the need for immediate action directed towards strengthening the healthcare infrastructure, improving the diagnostic systems, ensuring reliable access to factor concentrates, and establishing systematic care models aimed at mitigating the long-term morbidity and mortality in a low-and middle-income settings like Pakistan. This research was supported by Fatmeed foundation however they have no role in analyzing data and conceptualization of this research.

Keywords: Hemophilia, clinical outcomes, bleeding disorder

MENA-P-193

Prevalence of Myeloproliferative Neoplasms (MPNs) and its Molecular Biomarkers in Saudi Population in Al-Madinah Region

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Myeloproliferative Neoplasms (MPNs) are hematological disorders characterized by increased production of myeloid lineage blood cells. MPNs are categorized as Philadelphia (Ph) chromosome-positive, including Chronic Myeloid Leukemia (CML), Ph chromosome-negative, Polycythemia Vera (PV), Essential Thrombocythemia (ET), and Primary Myelofibrosis (PMF). Limited data exist on the frequency of MPNs and their molecular markers in the Saudi population. This study aimed to identify the common MPN subtypes and their associated molecular markers in Saudi citizens residing in the Al-Madinah Region. We retrospectively analyzed the clinical data of 60 patients between 2014 and 2023. Bone marrow samples were analyzed for mutations in the BCR-ABL, JAK2, CALR, and MPL genes using karyotyping, specific FISH panels, and various mutation detection methods, including Sanger sequencing. Our findings revealed that MPNs were more prevalent (78%) than Acute Myeloid Leukemia (AML; 11.6%) and Acute Lymphoblastic Leukemia (ALL; 10%) in the study population. Among MPNs, CML was the most common(34%), followed by equal rates of PV and ET (27.6% each), with PMF showing the lowest incidence (10.6%). Molecular biomarker analysis demonstrated BCR-ABL-positive mutations in all CML cases, JAK2-positive mutations in all PV cases, and the most frequent mutation in PMF cases. ET and PMF cases exhibited various mutation patterns, with triple-negative status for JAK2, CALR, and MPL being the most frequent molecular alterations in ET. This study represents the first estimation of Ph chromosome-negative MPN incidence and identification of common molecular biomarkers used for diagnosis in Saudi Arabia. Further studies with larger sample sizes and broader regional coverage are required to confirm these findings and to provide a more comprehensive understanding of MPNs in the Saudi population.

Keywords: Myeloproliferative neoplasms, Saudi Arabia, Al-Madinah Region, Molecular markers; BCR-ABL, JAK2; CALR, MPL

MENA-P-198

Clinical and Therapeutic Profile of Non-Hodgkin's Lymphoma: A Retrospective Study from a Najran Oncology Center

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BACKGROUND AND OBJECTIVE: Children diagnosed with cancer are more likely to have vitamin D problems due to compromised health before diagnosis, the disease itself, and cancer treatments. Previous studies, albeit limited in scope, has shown an elevated prevalence of vitamin D deficiency in pediatric oncology patients. This study aims primarily to describe the prevalence of vitamin D insufficiency among children with cancer in Lebanon.

SUBJECTS AND METHODS: This was an unmatched case control study comparing vitamin D levels among Lebanese children diagnosed with cancer (cases) to children without cancer (controls) in Lebanon. Serum vitamin D levels were determined for both cases and controls. Vitamin D status was evaluated by categorizing patients into two groups: hypovitaminosis D, defined as 25(OH)D levels < 30 ng/ml, and normal levels, defined as 25(OH)D levels ≥ 30 ng/ml. Additionally, patients were classified according to the endocrine society recommendations: deficiency (25(OH)D ≤ 20 ng/ml), insufficiency (25(OH)D 21-29 ng/ml), and normal levels (25(OH)D ≥ 30 ng/ml).

RESULTS: This unmatched case-control study included a total of 268 patients with 67 oncology pediatric patients (cases) and 201 non-oncology pediatric patients (control). The prevalence of vitamin D insufficiency and deficiency among cancer children were 16.45% and 80.6%, respectively. In contrast, among non-oncology patients, 32.4% showed vitamin D insufficiency, and 38% had vitamin D deficiency. We found a significant association between having cancer and the classification of vitamin D levels. Age and the duration of chemotherapy were significantly associated with vitamin D levels in children with cancer, with older children and those receiveing chemotherapy for less than one year having lower vitamin D levels.

CONCLUSION: Among children diagnosed with cancer, a notable prevalence of vitamin D deficiency is observed, highlighting this subgroup as particularly high-risk and, therefore, underscoring the significance of early detection and supplementation in this specific high-risk population.

Keywords: Vitamin D, oncology, Endocrinology

MENA-P-201

The Role of Platelet-to-Neutrophil Ratio (PNR) as a Biomarker for Pulmonary Hypertension and Echocardiographic Changes in Sickle Cell Disease Patients: A Retrospective Cohort Study

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BACKGROUND: Sickle cell disease (SCD) is a chronic hemoglobinopathy characterized by persistent inflammation, vaso-occlusion, and progressive organ damage. Pulmonary hypertension (PH), affecting 6-10% of adults with SCD, is a serious complication defined as a mean pulmonary artery pressure (mPAP) >20 mmHg. Echocardiographic features such as right ventricular dilation (RVD) and right atrial enlargement (RAE) are frequently associated with PH. The platelet-to-neutrophil ratio (PNR), representing platelet activation and neutrophil-driven inflammation, has emerged as a potential biomarker in SCD. However, its relationship with PH and echocardiographic changes remains unexplored.

AIMS: This study investigates associations between PH and PNR in SCD and evaluates whether both correlate with hydroxyurea use, hematologic parameters, complications, and echocardiographic changes.

METHODS: This retrospective cohort study was conducted at King Fahd Hospital, Al Khobar, Saudi Arabia, and included 115 SCD patients aged > 14 years with echocardiogram reports. Data were extracted from electronic medical records January 2019 to January 2025. PH was defined as mPAP > 20 mmHg, estimated using pulmonary artery systolic pressure and acceleration time per 2023 American Society of Echocardiography (ASE) guidelines. PNR was calculated by dividing absolute platelet count by absolute neutrophil count. Additional variables included age, gender, echocardiographic findings (RVD, RAE), history of complications (acute chest syndrome, pulmonary embolism, stroke, heart failure), and laboratory parameters (hemoglobin, hemoglobin S, fetal hemoglobin). Statistical analyses were performed using Chi-square and ANOVA in SPSS v26. Ethical approval was obtained (IRB-UGS-2024-01-730).

RESULTS: PH was significantly associated with hydroxyurea use(P = 0.024), ACS(P = 0.009), RVD (P = 0.041), higher HbS (P = 0.015), and lower HbF (P = 0.016). PNR was not significantly associated with PH (P = 0.137). Stratified analysis revealed that higher PNR levels correlated with prior pulmonary embolism (P = 0.011) and lower HbF (P = 0.049).

CONCLUSIONS: Although PNR was not independently linked to PH in SCD, tertile analysis revealed associations with prior pulmonary embolism and lower HbF, suggesting it may reflect chronic vascular complications rather than predict PH directly. Echocardiographic abnormalities and markers such as ACS, HbS, and HbF remain stronger predictors of PH.

Keywords: Sickle cell disease, platelets, neutrophils, pulmonary hypertension

MENA-P-207

A Quality Initiative to Improve IVC Filter Use and Retrieval: Translating Guidelines into Practice

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INTRODUCTION: IVC filters are used to prevent pulmonary embolism in patients with venous thromboembolism who cannot receive anticoagulation. However, their use has often extended beyond evidence-based indications. Guidelines also emphasize prompt retrieval once anticoagulation can be safely resumed.

WHY THIS PROJECT: Baseline analysis revealed 27% of IVC filters were inserted without an appropriate indication based on our, which align with ASH recommendations. Additionally, 24% of filters were not retrieved. The aim of this project was to improve the use of IVC filters and to increase the rate of retrieval.

METHODS: We applied the Harvard 7-step and PDSA cycles to address both overuse and under retrieval. Outcomes were measured over two periods: a pre-intervention (March–September 2024) and a post-intervention (October 2024–May 2025). Fisher's exact test to assess statistical significance and Statistical process control (SPC) were constructed to identify special cause variation using Western Electric rules.

INTERVENTIONS: (1) Establishment of policy restricting insertion to Hematology: Baseline data showed that hematology consultation was associated with significantly higher appropriateness of IVC filter insertion (92.3% vs. 70.6%). We implemented a policy requiring hematology approval prior to insertion. (2) Automatic Consultation to IVC filter team Upon Filter Request: An automatic E-mail alert system triggered by any filter order in the EHR. The thrombosis quality specialist receives the alert and informs the IVC filter team, who assess appropriateness, and communicate directly with the primary team to guide evidence-based decisions. (3) Weekly Review: A dedicated IVC filter team—comprising a hematology consultant, fellow, and quality specialist —conducts weekly rounds to review all inpatients with filters. Each case is reassessed, a retrieval plan is documented, and the primary team is contacted to ensure implementation. (4) Education of Healthcare Providers.

RESULTS: Following the intervention, the appropriateness of filter insertion improved from 72.7% (48/66) to 91.4% (64/70), *P* = 0.0028. SPC analysis demonstrated special cause variation, triggered by Rule # 3.Similarly, the retrieval rate increased from 76% (38/50) to 92.3% (48/52), *P* = 0.0062. SPC analysis showed special cause variation triggered by Rule #4. The median time to IVC filter retrieval decreased from 35 days (IQR 18-60) to 22 days (IQR 14-51), *P* = 0.180, Mann–Whitney U test.

CONCLUSION: This quality improvement initiative successfully translated clinical guidelines into practice through a multifaceted strategy. The intervention led to significant improvements in IVC filter appropriateness and timely retrieval. Next steps include scaling the model to other centers to support broader guideline adherence in thrombosis care.

Keywords: IVC, guidelines, thrombosis

MENA-P-208

A Quality Initiative to Improve IVC Filter Use and Retrieval: Translating Guidelines into Practice

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BACKGROUND: Bone marrow biopsy is a key diagnostic tool in hematology, yet safety data in anticoagulated patients remain limited. Practice varies widely, with some clinicians interrupting anticoagulation and others proceeding without changes. This systematic review and meta-analysis aimed to clarify the safety profile of bone marrow biopsy in anticoagulated patients.

METHODS: Following PRISMA guidelines, we searched PubMed, Embase, Cochrane Library, and Web of Science from 1990 to December 2024. Eligible studies reported bleeding outcomes in anticoagulated patients undergoing bone marrow biopsy. The primary outcome was major bleeding; secondary outcomes included minor bleeding, thrombotic events, and bleeding risk factors. Random-effects meta-analysis was performed using R software.

RESULTS: Seven studies (78,574 procedures; 1,078 anticoagulated patients, 1.4%) were included. The pooled major bleeding rate was 0.053% (95% CI: 0.040-0.072%), or fewer than 1 in 1,800 procedures. In anticoagulated patients, the rate was 0.278% (95% CI: 0.095-0.815%), about 1 in 360 procedures. Heterogeneity was negligible (τ^2 =0.000, I^2 =0%). Significant risk factors included myeloproliferative neoplasms (OR 4.12, 95% CI: 1.67-10.15) and severe thrombocytopenia < 50 × 10^9/L (OR 3.45, 95% CI: 1.23-9.67). Anticoagulation status alone was not significantly associated with increased bleeding risk (OR 0.85, 95% CI: 0.23-3.12).

CONCLUSIONS: Bone marrow biopsy has an excellent safety profile, with major bleeding rates < 0.1% overall and < 0.3% in anticoagulated patients. Findings do not support routine interruption of anticoagulation before biopsy. Risk assessment should focus on established predictors such as myeloproliferative neoplasms and severe thrombocytopenia. Results support a shift toward more liberal biopsy practices in anticoagulated patients.

Keywords: Bone marrow biopsy, anticoagulation, bleeding risk

MENA-P-212

Telomere Length and Response to Imatinib Therapy in Newly Diagnosed Chronic Myeloid Leukemia Patients – Chronic Phase: Measured by Flow-FISH Technique

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BACKGROUND: Telomere length measurement has been assessed by many previous studies In CML patients. Telomere shortening was found to correlate with disease progression. The aim of our study was to measure Relative Telomere Length (RTL) in chronic phase CML patients before imatinib therapy and correlate it with their molecular response after 6 months.

METHODS: This study included 38 CML patients in chronic phase and 40 normal adult controls. RTL was assessed in leukocytes by Flow-FISH technique.

RESULTS: This work revealed significant reduction of the RTL in CML patients than normal control. RTL showed negative correlation with BCR-ABL gene expression measured at diagnosis (p=>0.001). Patients who did not show complete molecular remission had longer telomere length (mean 0.24 ± 0.05) than patient got complete molecular remission (mean 0.011 ± 0.04), and the difference was statistically significance (*P* = 0.048).

CONCLUSION: Shortening of TL may have a role in the pathogenesis of the disease, and may be used as a predictor to patient´s molecular response to Tyrosine Kinase Inhibitors therapy (TKIs).

Keywords: Leukemia, imatinib therapy, CML

MENA-P-218

Experience and Outcomes from the First Comprehensive Pediatric Private Hematopoietic Stem Cell Transplantation Center in Jordan

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BACKGROUND: Hematopoietic stem cell transplantation (HSCT) is a well-established treatment for various conditions. In the Eastern Mediterranean region, pediatric HSCT services face distinct challenges, including a large pediatric population and high rates of consanguinity, which contribute to a higher prevalence of genetic diseases and increased demand for transplantation. The current availability of transplant teams in the region falls short of meeting this growing need. Incorporating private transplant services could help address this demand.

METHODS: A retrospective review of all patients under the age of 18 years who received HSCT at the Bone Marrow and Stem Cell Transplantation Center, Istishari Hospital, Jordan.

RESULTS: In June 2016, this private center of excellence was established with the mission of providing comprehensive and personalized care to patients suffering from a wide range of hematological disorders and malignancies. Serving not only the Jordanian population but also individuals from surrounding countries, the center aims to deliver advanced, individualized treatment plans tailored to each patient's specific needs, ensuring the highest standards of care. Since then, 65 patients received 68 hCTs ; 58 (85%) were allogenic and 10 (15%) were autologous. The median age of the cohort was 6.5 (0.6-17) years; 40 (61%) were males. The primary indication for allogeneic HSCT was leukemia (*n* = 23, 40%), followed by severe aplastic anemia (SAA; n = 12, 21%), hemoglobinopathies (*n* = 10, 17%), bone marrow failure syndromes (*n* = 5, 8%), primary immunodeficiency (*n* = 5, 8%), inherited metabolic diseases (*n* = 1, 2%), and lymphoma (*n* = 1, 2%). For autologous HSCT, neuroblastoma was the main indication (*n* = 6, 60%). Among the allogeneic transplants, 30 (52%) utilized matched related donors, while 28 (48%) were haploidentical. Peripheral blood stem cells were the graft source in 44 transplants (64%). Primary graft failure occurred in 1 patient with SAA who later engrafted after a second transplant. At a median follow-up of 50 (3-106) months, 52 patients were alive and free of primary disease, yielding an overall survival rate of 80% with transplant-related mortality (TRM) of 4.6%. A remarkable 100% survival was observed in patients with benign hematological diseases.

CONCLUSION: We observed good outcomes that are comparable to international data. Establishing private HSCT services allows more patients to benefit from this potentially life-saving treatment.

Keywords: Stem Cell, Transplantation, bone marrow