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ABSTRACTS

MENA ABSTRACTS

FREE PAPER SESSION

MENA-O-108 | **Demographic Patterns and Therapeutic Responses in Glanzmann Thrombasthenia:a Multi-Center Study**

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Abstract 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."

MENA-O-111 | **Diagnostic and Prognostic values of NCF4 rs1883112 Polymorphism Expression among Acute Lymphoblastic Leukemia in Egyptian Children**

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Abstract 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.

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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Abstract 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 x10^9/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.

MENA-O-121

Which Is More Effective: Weight-Based or Hemoglobin-Based Formula for Packed Red Cell Transfusion?

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Abstract 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 hours 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.

MENA-O-136

Deconvolution of Peripheral Blood Transcriptomes Reveals Prognostic Immune Shifts in Pediatric Acute Lymphoblastic Leukemia: A Machine Learning–Driven Study

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Abstract Background: Immunophenotypic variation in pediatric Acute Lymphoblastic Leukemia (ALL) influences therapeutic outcomes and relapse rates. However, comprehensive immune profiling via flow cytometry or single-cell
RNA-seq remains inaccessible in many settings. Machine learning (ML)-assisted transcriptomic deconvolution presents a promising alternative to derive immune cell proportions from bulk RNA-seq.

Objective:
To apply ML-based deconvolution on peripheral blood transcriptomic data to identify immune cell patterns predictive of minimal residual disease (MRD) in pediatric ALL.

Methods:
This retrospective study used publicly available gene expression data from pediatric ALL cohorts (GEO:GSE48558, n=78). CIBERSORTx was applied for deconvolution to estimate relative immune cell fractions. A random forest classifier was trained to predict MRD status based on these inferred cell proportions.Feature importance was evaluated using SHAP values.

Results:
Patients with positive MRD exhibited higher proportions of regulatory T cells (p=0.004) and monocytes (p=0.01), and reduced CD8+ T cells (p=0.002). The random forest model achieved an AUC of 0.91 (95% CI: 0.84–0.97) in distinguishing MRD-positive from MRD-negative patients. SHAP analysis highlighted CD8+ T cell depletion and Treg enrichment as the most predictive features. Cross-validation confirmed model
stability (accuracy = 87.5%, F1-score = 0.89).

Conclusion:
ML-guided deconvolution of blood RNA-seq enables non-invasive immune profiling in pediatric ALL, offering valuable prognostic insights. This method may assist in risk stratification where flow cytometry is limited, and lead the way for broader applications in hematologic malignancies.

Keywords: Acute Lymphoblastic Leukemia, deconvolution, machine learning, minimal residual disease,immune profiling.

MENA-O-140 | **Results of Combined Treatment of Non-Hodgkin Lymphoma of the Ileocecal Region in Children**

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Abstract Background: In children with non-Hodgkin lymphoma, damage to the ileocecal region is 1% to 2%. Complications such as intestinal perforation and peritonitis are common when this area is affected by lymphoma.
Study Purpose: To study the results of treatment of patients with lymphoma of the ileocecal angle.

Results:
We analyzed results of treatment of 6 patients with damage to the ileocecal angle from the pediatric oncology department of our center in 2024. The patients' ages ranged from 3 to 12 years, averaging 4.8 years. Of the patients, 4 (66.6%) were boys and 2 (33.3%) were girls. In cases of damage to the ileocecal
angle of the large intestine, in which signs of intestinal obstruction, increased risk of bleeding, intestinal perforation, and peritonitis developed, right-sided hemicolectomy and ileotransverse anastomosis was performed. In the postoperative period, patients underwent up to 6–8 courses of CHOP chemotherapy (vincristine, 1.4 mg/m2; cyclophosphamide, 750 mg/m2; doxorubicin, 30 mg/m2; prednisolone, 40 mg/m2) and CHOEP (vincristine, 1.4 mg/m2; cyclophosphamide, 750 mg/m2; doxorubicin, 30 mg/m2; etoposide, 100 mg/m2; prednisolone, 40 mg/m2). Chemotherapy was carried out against the background
of detoxification and hepatotropic, cardiotropic, and antibiotic therapy.
During the operation and postoperative treatments, no serious complications were observed.

Conclusion: In cases of damage to the ileocecal angle by malignant lymphoma in patients with a high risk of perforation and peritonitis, the use of surgical intervention at the initial stage of treatment improves

MENA-O-141

REAL WORLD DATA OF EMICIZUMAB PROPHYLAXIS IN YOUNG CHILDREN WITH SEVERE HEMOPHILIA A WITHOUT INHIBITORS

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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 (≤ 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 mth (6-35) at start of emicizumab were followed-up for median 24 mth (14-46). The cumulative follow-up was 342 mth. 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.(TABLE1) 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 yr (n=7) was zero.

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.

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 seconds) 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.

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.

MENA-O-149 | Redox-Dependent Modulation of IL-10 and TGF-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.

MENA-P-151 | Pediatric oncology

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BACKGROUND: Despite a well-known dose-dependent association between the risk of cardiac dysfunction and anthracycline, the risk of cardiac dysfunction for any given anthracycline dose varies between patients. CELF4 is an RNA binding protein mediating the alternative splicing of the TNNT2 gene, that encodes cardiac troponin T. The SNP rs1786814 in CELF4 is associated with anthracycline-induced cardiomyopathy in childhood cancer survivors (CCS) The aim of the study : was to detect the modifying effect of CELF4 (rs1786814) gene polymorphism on anthracycline related cardiotoxicity in survivors of childhood cancer.
METHODS: This cross-sectional study case control study included 53 CCS who had regular follow-up visits at the Pediatric Oncology Unit, Menoufia University Hospital compared to 53 age and sex matched healthy controls . CELF4 (rs1786814) gene polymorphism and conventional and speckle-tracking Echocardiography were done for all included children.
RESULTS: Regarding CELF4 (rs1786814) polymorphism , the homozygous mutant

GGgenotype and the mutant G allele were significantly predominant in CCS than controls . Conventional Echocardiographic study revealed significantly lower ejection fraction and end-systolic diameter in CCS compared to the controls . Speckle- tracking Echocardiography showed significant lower (GLPS-A4C) and (GLPS-LAX) in CCS than controls with no significant difference regarding (GLPS-A2), (GLPS-AVG) . Multivariate logistic regression analysis illustrated a statistically significant relation between cumulative anthracycline dose >300mg/m2 and CLEF4 (rs1786814) genotypes (GG and GA) and the risk of cardiotoxicity CONCLUSION: The presence of a mutant CELF4 (rs1786814) allele could be considered as a risk factor for cardiotoxicity in anthracycline treated CCS

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.

MENA-O-157 | Warm Autoimmune Hemolytic Anemia as a Gateway to Diagnosing XMEN syndrome in a Young Child

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.His277GlnfsTer8), 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.

MENA-O-158

Angiotensin Converting Enzyme Inhibitors (ACEIs) For Anthracycline-induced Cardiomyopathy: A Systematic ReviewAnd 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.

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 minutes) after the injection and, the second between 24 and 32 hours (+/- 1 hour) 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

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.

MENA-O-167

Prognostic Significance of PNH Clones in Aplastic Anemia Treated with Immunosuppression or Allogeneic HSCT

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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.
Method: 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.
Result: Among the 163 patients, 89 (54.6%) male and 74 (45.3%) were female, and the median age was 22(IQR
17-29) 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.

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 ERCC6L2,MYSM1 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.
Result: ERCC6L2 mutation:
18 patients from ten Saudi families with mutations of the ERCC6L2 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.

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.

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.

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.

MENA-O-173 | **Bleeding disorder**

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Beni Messous Hospital[1], beni messous hospital[2], beni messous hospital[3],beni messous hospital[4], beni messous hospital[5].

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.
Method : 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).

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.

MENA-O-182 | **Evaluation of TP53 Expression in pediatric B- Cell Acute Lymphoblastic Leukemia and Its Correlation with Clinicopathological Profiles**

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Background:TP53 is a tumor suppressor gene which impacts tumor progression and prognosis in various cancers. Alternative mechanisms of wild type TP53 protein inactivation such as over-expression of TP53 isoforms have been identified in many types of cancers. In the present study, we aimed to evaluate the expression of different TP53 isoforms in pediatric B-cell ALL (B-ALL) patients and explored its correlation with clinicopathological profiles.
Materials and methods: One hundred pediatric B-cell ALL patients (≤12 years) and 20 age matched healthy controls were enrolled for this study. Expression of TP53 full-length and isoform Delta40TP53,Delta133TP53, TP53Beta expressions was checked using qRT-PCR. Fold change was calculated using 2-Ct method. The expression of isoforms was correlated with various clinical parameters such as risk stratification, blast count, molecular cytogenetics etc.
Results: The median age of the patients was 5 years with male: female ratio of 1.2:1. Fifty two patients had high expression of TP53 full length gene expression with the maximum fold change of 10-fold. Isoform Delta40TP53, Delta133TP53 and TP53Beta was overexpressed in 17%, 53% and 6% of the patients, respectively. High expression of Delta133TP53 showed significant association with the presence of ETV6::RUNX1 (p=0.047) fusion, which is a marker of standard risk. The presence of KMT2A::AF4 which is linked with poor prognosis was also associated with higher TP53Beta expression (p=0.043). However, we could not find any significant association between expression of TP53 isoforms and other clinical parameters such as age, gender, TLC, MRD, or progressive disease etc. in pediatric B-ALL patients.

Conclusions: Our study has shown that the expression of full length TP53 and its isoforms is dysregulated in pediatric B-ALL patients. Additionally, the findings suggest that Delta133TP53 could further be explored as a potential biomarker for standard risk. While TP53Beta might serve as an indicator of poor risk in ALL and could possibly play a role in pathogenesis of high risk B-ALL, warranting further investigation on larger cohorts with longer follow ups.

MENA-O-186 | Renal Complications in Children with Sickle Cell Disease: A Retrospective Cohort Study

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Taibah University [1].

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.

MENA-O-187 | Cytogenetic findings and post induction MRD in B-Lymphoblastic Leukemia

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Background/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.

MENA-O-205 | **Prevalence and Diagnostic Delay of Inherited Bleeding Disorders in Women with Heavy Menstrual Bleeding**

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Background: Heavy menstrual bleeding (HMB) is a common gynecological complaint that significantly impacts women's quality of life. Inherited bleeding disorders (IBDs), such as von Willebrand disease and platelet function defects, are underrecognized causes of HMB in reproductive-age women. Despite their clinical importance, diagnostic delays remain frequent, leading to prolonged morbidity and inappropriate management. Current literature on the prevalence of IBDs among women presenting with HMB and the factors contributing to delayed diagnosis is limited, especially in real-world settings.
Objective: To determine the prevalence of inherited bleeding disorders and quantify the diagnostic delay among women presenting with heavy menstrual bleeding in a tertiary care setting.
Methods:
A retrospective cross-sectional study was conducted using medical records of women aged 15–45 years who presented with HMB to the gynecology and hematology clinics of Hospital Aballi Julio Trigo between January 2015 and December 2020. Inclusion criteria were documentation of HMB for ≥6 months and completion of standardized hemostatic evaluation, coagulation profile, von Willebrand factor assays, and platelet function tests. Women with structural uterine pathology affecting coagulation were excluded.
Main outcome measures included the prevalence of confirmed IBD diagnoses onset of HMB to confirmed diagnosis. Ethical approval was obtained from the institutional review board.
Results:
Of 312 eligible women, 104 (33.3%) were diagnosed with an inherited bleeding disorder. Von Willebrand disease was the most common (56 cases, 53.8%), followed by platelet function defects (31 cases, 29.8%) and other rare coagulation factor deficiencies (17 cases, 16.3%). The mean diagnostic delay was 6.2 ± 3.1 years (range: 1–14 years). Women with milder bleeding phenotypes experienced significantly longer delays compared to those with severe bleeding (7.5 vs. 4.3 years, $p < 0.01$). Prior history of epistaxis, postpartum hemorrhage, or family history of bleeding disorders was documented in 71% of diagnosed cases but was infrequently pursued for early investigation.
Conclusion:
Inherited bleeding disorders are highly prevalent among women with heavy menstrual bleeding, yet substantial diagnostic delays persist,

particularly in milder cases. These findings underscore the need for heightened clinical suspicion, standardized bleeding history assessments, and early referral to hematology to reduce morbidity and improve quality of life. Implementation of targeted screening protocols in gynecology clinics may significantly shorten time to diagnosis and optimize management strategies.

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.

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 ≤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.

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.

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.
Immune effector 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 (≥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 >400ng/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.

MENA-O-216 | **Unraveling novel complex genomic rearrangements in severe hemophilia A patients using optical genome mapping**

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Background. Around 50% of severe hemophilia A (sHA) is caused by recurrent inversions disrupting F8 gene. The underlying genetic cause cannot be precisely determined in ~5% of patients, using routine diagnostic (Dx) assays.
Aims. To decipher unusual results obtained with Dx assays in sHA families.
Methods. Dx assays do not detect aberrations outside F8. Optical Genome Mapping (OGM) uses enzymes to fluorescently tag high molecular weight DNA at specific sequence motif without breaking DNA,
preserving the architecture of CGRs. We evaluated, for the first time, the Dx capability of OGM in sHA.
Results. We identified the exact architecture of CGRs in two sHA families: (i) a 141.5 kb balanced inversion affecting intron 1 of F8 with breakpoints within known segmental duplications. None of the Dx assays can delineate the inversion with this resolution. (ii) IS-PCR results of Inv22 revealed an unusual pattern in the proband. OGM characterized the exact positions and orientation of a complex inversion: a novel Inv22 (~567 kb) concomitant with an extragenic duplication of ~193 kb upstream of F8. A critical gene in the region was RAB39B, a candidate gene for intellectual disabilities (ID) in the Xq28 duplication syndrome mediated by the int22h-related repeats. OGM showed the inversion breakpoint bisected one copy of
RAB39B, and another copy was left untouched, which explains the lack of ID in the proband.
Conclusion. OGM has the potential to become a next-generation Dx tool for sHA to detect both known recurrent as well as more complex genomic rearrangements as presented here.

MENA-O-217 | **Assessment of variabilities in clinical & 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.

MENA-O-219

Matched-Related Hematopoietic Stem Cell Transplantation in High-Risk Thalassemia Using a Reduced-Intensity Conditioning Regimen with Post-Transplant Cyclophosphamide for Graft-Versus-Host Disease Prophylaxis: Balancing Intensity and Safety.

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Background: Allogeneic hematopoietic stem cell transplantation (HSCT) remains the only reasonable curative modality for Thalassemia major (TM). Reduced-intensity conditioning (RIC) is associated with lower transplant-related toxicity. Post-transplant cyclophosphamide (PT/Cy) has improved engraftment and decreased the risk of graft-versus-host disease (GVHD).
Methods: This is a retrospective analysis of all patients with TM who received HSCT from matched related donor (MRD) at the Bone Marrow and Stem Cell Transplantation Center, Istishari Hospital, Jordan from March
Results: Five patients with median age of 12 years (range:7-17) were included. Three were males. All patients received Rabbit anti-thymocyte globulin (rATG) 0.5 mg/kg on day −10 and 1.5 mg/kg/day on days −9 to −7, Thiotepa 10 mg/kg on day -7, Fludarabine 30 mg/m2/day IV on days −6 to −2, Cyclophosphamide 14.5 mg/kg/day IV on days −6 to −5, and 300 cGy TBI in a single fraction on day-1. The graft versus host disease (GVHD) prophylaxis was with post-transplant Cyclophosphamide (PT/Cy) at 50 mg/kg/day IV on days +3 and +4 post-HSCT. Mycophenolate Mofetil and Tacrolimus were started on day +5 and stopped on days +28 and +365 post-HSCT, respectively. The median CD34+ cell dose was 7.3×106/kg (range:2.35-9.5×106/kg). Three patients received peripheral blood stem cells (PBSC).
The median time for Neutrophil and platelet engraftment was 14 (range:13-17) and 26 (range:15-43) days respectively. None of the patients needed ICU admission. Grade I–II mucositis occurred in all patients.Four patients experienced CMV reactivation, which was managed preemptively. One patient developed idiopathic interstitial pneumonitis, which was treated with high dose systemic steroids and other supportive measures. Only two patients developed acute grade I skin GVHD. No patients have developed chronic GVHD. At a mean follow-up time of 50 months (range: 31-77), all patients are alive, transfusion independent with donor-type Hb-electrophoresis.
Conclusion: The use of RIC with PT/Cy in MRD-HSCT for high-risk patients with TM is scarcely reported. This approach is safe, effective and associated with a favorable outcome. More prospective studies with a larger sample size are required.

MENA-O-220 | **Haploidentical Hematopoietic Stem Cell Transplantation as First-Line Therapy in Children with Severe Aplastic Anemia: A Single Center Experience from Jordan**

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Background: Severe aplastic anemia (SAA) is a rare but life-threatening hematologic disorder that can be cured with hematopoietic stem cell transplantation (HSCT). Although the outcomes with HLA-identical siblings are favorable, the majority of patients do not have HLA-identical donors.
Methods: We report on 9 consecutive patients with SAA who received upfront haplo-identical HSCT from October 2016 until May 2025 at the Bone Marrow and Stem Cell Transplantation center, Istishari Hospital, Amman-Jordan, which is considered the first private BMT center in Jordan.

Results: Nine patients with a median age of 7 years (range: 1.5-13) received haploidentical HSCT. Six were males. All patients received Rabbit anti-thymocyte globulin (rATG) IV 0.5 mg/kg on day −9 and 2 mg/kg/day on days −8 and −7, Fludarabine IV 30 mg/m2/day on days −6 to −2, Cyclophosphamide IV 14.5 mg/kg/day on days −6 to −5, and 300 cGy TBI in a single fraction on day -1. The graft versus host disease (GVHD)prophylaxis was with Cyclophosphamide (PT/Cy) at 50 mg/kg/day IV on days +3 and +4 post-HSCT.Mycophenolate Mofetil and Tacrolimus were started on day +5 and stopped on days +28 and +365 post-HSCT, respectively, giving no signs of GVHD. Seven patients (77%) received peripheral blood stem cells(PBSC), and two (22%) received GCSF-primed bone marrow (BM) grafts. Five donor-recipient pairs had major blood group incompatibilities.
The median time for neutrophil and platelet engraftment was 14 (range: 12–17) and 18 (range: 16–25) days, respectively. At a median follow-up time of 71 months (3–106), overall survival and event-free survival for the whole cohort are 100%. One patient developed primary graft failure and was successfully engrafted after a second transplant. None of the patients required ICU admission. Grade I- II acute GVHD occurred in six patients. Five patients had CMV reactivation, one EBV reactivation, and one BK virus cystitis. Two patients had chronic GVHD, of which one was extensive.
Conclusion: Matched related donor (MRD) HSCT is recommended for patients with SAA and has been shown to improve long-term survival. For those without an available MRD, upfront T-replete haploidentical HSCT offers an effective and safe alternative. Our findings suggest that this strategy may be feasible in resource-limited settings. Further prospective studies with larger cohorts are warranted to validate these results.

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 x109/ul to 1.8x109/ul, the mean increase in Hb was 2.15 g/dl, and the magnitude of increase in platelet count of 66x109/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.

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 cytopenia at 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/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.

MENA-O-225 | HIGH LEVELS OF SERUM AND CSF 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 69.3ng/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.

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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.