



POSTER PRESENTERS

- 1 Comprehensive Newborn Screening of Lysosomal Diseases: Insights from the LysoNeo Study**
Prof. Abdellah Tebani, Professor / Clinical Academic Consultant, Rouen University Hospital / Metabolic Biochemistry Department
- 2 Myxedema Coma in Neuronal Ceroid Lipofuscinosis Type 6: a life-threatening complication in a rare disease**
Mr. Ans Ahmed Mahmood, Medical Student, Gulf Medical University
- 3 Multisystem Inflammatory Syndrome in Neonates (MIS-N): A Case Report and Clinical Insights**
Mr. Yasser Abd Alsamad, Medical Student, University of Sharjah
- 4 Variant-Specific Proteostasis Defects Define Pathogenic Mechanisms in Rare TGFBR2-Associated Aortopathies**
Ms. Fadia Mahmoud Aayed Abu Sailik, PhD student, UAE University
- 5 Hematological Manifestations and Platelet Abnormalities in Pediatric DiGeorge Syndrome: A Retrospective Study on Thrombocytopenia and GPIIB Association**
Ms. Dana Haj Mohammed (1), Student, University of Sharjah
- 6 The Enigma of West Syndrome: A Case of Infantile Spasms Without Genetic Clues**
Dr. Ali Raafat Ammar, Physician, Faculty of Medicine, Ras Al Khaimah Medical and Health Science University
- 7 Elucidating the Cellular and Molecular Mechanisms of Discoidin Domain Receptor 2 (DDR2) Disease-Causing Mutations**
Mrs. Anjana Raj, PhD Student, United Arab Emirates University
- 8 Kaposi's Sarcoma in Patients Exposed to Calcineurin Inhibitors or Systemic Corticosteroids: A Two- Decade Real-World Safety Assessment.**
Dr. Sara Omar Abu Aleenein, Medical Student, Jordan University of Science and Technology, Medicine
- 9 ExomePilot: An Interactive AI-Ready Platform for Streamlined Rare Disease Genomic Diagnostics**
Dr. Nour al dain Marzouka, Assistant Professor, Department of Genetics & Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al Ain, United Arab Emirates
- 10 Severe Neonatal Nemaline Myopathy Associated with NEB Gene Mutation: A Case Report**
Dr. Aaisha Mohammed Rafi (1) Medical Resident, Aster Hospital Dubai
- 11 Fanconi-Bickel syndrome presenting with neonatal hyperglycemia: A case report**
Dr. Aaisha Mohammed Rafi (2) Medical Resident, Aster Hospital Dubai
- 12 FNAIT in the Middle East: A Confirmed Case Highlighting Immunogenetic Diversity and Evidence-Based Neonatal Management.**
Dr. Hamza Haj Mohamad, Pediatric resident, AL Jalila Hospital
- 13 Clinical and Genetic Evaluation of Childhood-Onset Epilepsy: A Comprehensive Analysis**
Dr. Alfiya Fasaludeen, Post doctoral researcher, Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al Ain, United Arab Emirates
- 14 Surgical Management of Hemimegalencephaly: A Systematic Review and Meta-Analysis of Seizure Freedom and Complications**
Ms. Shriya Karlapudi Year 3 Medical Student, Faculty of Life Sciences and Medicine, King's College London, UK
- 15 Combined Endoscopic Submucosal Dissection and Complete Endoscopic Hand Suturing for Solitary Rectal Ulcer Syndrome: A Novel Global Case Report**
Dr. Shahd Faris Alqudah, MBBS, University of Sharjah
- 16 Paramyotonia Congenita in an Adolescent With SCN4A Mutation: The Importance of Clinical Phenotyping & Early Genomic Testing**
Dr. Aisha Sajjad (1), Medical Intern, Kings College Hospital



POSTER PRESENTERS

- 17 **Cholestatic Hepatitis in Pediatric Stevens–Johnson Syndrome/Toxic Epidermal Necrolysis: A Diagnostic Challenge**
Dr. Aisha Sajjad (2), Medical Intern, Kings College Hospital
- 18 **Late Onset Glutaric Aciduria Type II: Case Report and Literature Review**
Dr. Mohammed Naji AlAbsi, Medical Intern, Sheikh Khalifa Medical City
- 19 **A Novel Loss-of-Function variant in TNS2 provides further evidence for TNS2-related nephrotic syndrome**
Ms. Sara Alhmoudi, Medical Research Specialist, UAE University
- 20 **Diagnostic Odyssey – What Comes After Negative Genome?**
Dr. Aya Abuelhaija, Clinical Genetics Physician, Boston Children's Hospital, Harvard Medical School
- 21 **Neurodevelopmental Disorder Associated with Heterozygous HNRNPD Variant in an 8-Year-Old Boy: A Case Report**
Dr. Zahra Mohamed Yusef Majichian, Pediatric resident, 1 Pediatric Residency program, Sheikh Khalifa Medical City, Abu Dhabi
- 22 **Coexisting Persistent Hyperinsulinemic Hypoglycemia of Infancy and epileptic encephalopathy: A Diagnostic Challenge in an Infant with Refractory Seizures**
Dr. Maitha Abdulla Mohammed Alhay Alshamsi, Medical Intern, Sheikh Shakhbout Medical City (SSMC)
- 23 **Where Rare Cancer Diagnostics Fail: A Comparative Analysis of Biomarker Testing Vulnerabilities**
Ms. Nooran Sabati, BSc. Medical Laboratory Sciences Student, Abu Dhabi University
- 24 **Population Representation and Enrichment of Pathogenic ATP7B Variants in Middle Eastern Populations**
Dr. Fahd Tayyab, BSc Student in Biomolecular and Medical Genetics (Undergraduate Researcher), Biomedical Sciences Department, College of Health Sciences, Abu Dhabi University, Al Ain, UAE
- 25 **Analysis of Whole Exome Sequencing Data for the Diagnosis of Prenatal Disease Variants in a UAE Cohort**
Ms. Lana Mashal, Student, Abu Dhabi University
- 26 **Reversing Acute Cardiomyopathy With Coenzyme Q10 Supplementation in Cobalamin B Disease: A Case Report and Literature Review**
Dr. Dalia Ra'a Said (1), Clinical Genetics Fellow, Tawam Hospital
- 27 **Hereditary Spastic Paraplegia: A 15-Year Retrospective Cohort Study of Clinical and Genetic Findings from Tawam Hospital, UAE**
Dr. Dalia Ra'a Said (2), Genetics Fellow, Tawam Hospital-SEHA
- 28 **When Neutropenia Unmasks DADA2: A Rare Intronic ADA2 Variant Identified by Whole-Exome Sequencing in an Infant: A Case Study**
Dr. Nadia Salaheldin Hamid Elsayedelfeil, Pediatric Resident, Al Qassimi Women's and Children's Hospital
- 29 **Primary immunodeficiency disorders in consanguineous populations: Diagnostic Challenges and Outcomes**
Aarifa Banu, Doctor, Tbilisi State Medical University, American Programme, Tbilisi, Georgia
- 30 **Multisystem Cardiovascular Complications and Therapeutic Challenges in a Patient with Marfan Syndrome**
Dr. Mustafa Elsheikh Farah Mohamed Elhassan (1), Medical Intern, Fakeeh University Hospital
- 31 **UAE–GCC–Global AI-Enabled Rare Disease Registry: A Transformative Platform for Precision Medicine and Societal Impact**
Advisor Mohamad Ahmd Layla, Chief Operations Officer for Global Alliance for Artificial Intelligence (GAFAI), Global Alliance for Artificial Intelligence (GAFAI)
- 32 **When Hypocalcemia Hides: Atypical Symptomatic Hypocalcemia in an Adult with DiGeorge Syndrome**
Dr. Doaa Elsheikh Farah Mohamed Elhassan (1), Internal Medicine Resident, Dubai Health
- 33 **The role of antenatal ultrasound in early detection of alobar holoprosencephaly**
Dr. Mariam Elsayed Mohamed (1), Intern physician, Fujairah hospital, EHS, Fujairah hospital, EHS



POSTER PRESENTERS

- 34 **MT-ND5–Associated Mitochondrial Disease Presenting with Multisystem Neurological and Ophthalmologic Manifestations in a Pediatric Patient: A Case Report**
Dr. Doaa Elsheikh Farah Mohamed Elhassan (2), Internal Medicine Resident, Dubai Health
- 35 **Joubert Syndrome (JS) with Intellectual Disability in a 28-Year-Old - The First Documented Case in the UAE**
Dr. Prof. Madhusudan Deepak Thalitaya, Consultant Psychiatrist and Head of Department, Intellectual Disability Psychiatry, Professor of Psychiatry, GMU and MBRU, EHS, Al Amal Psychiatric Hospital, Dubai, UAE
- 36 **Beyond Binary: Frequency-based Symptoms for Fabry Disease Detection using Machine Learning Algorithms**
Dr. Muhammad Jalal Khan, Postdoctoral Fellow, United Arab Emirates University
- 37 **Spontaneous Hemothorax Secondary to Hereditary Multiple Exostosis: A Rare Complication Masquerading as Pulmonary Tuberculosis**
Mr. Abdulrahman Abbas Yusuf Mohammed, General Medical Practitioner, Port Sudan Teaching Hospital
- 38 **Integrative Genomic and Pathway Analysis of Inherited Ocular Diseases Reveals Anatomical and Molecular Signatures Across the Eye**
Waruni Nawagamuwa, PhD Scholar, Department of Genetics and Genomics- UAE University
- 39 **Resolving Diagnostic Pitfalls in Inherited Ocular Disease Using Long-Read Sequencing and Integrated Genomic Analysis in an Emirati Cohort**
Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al-Ain, United Arab Emirates
- 40 **Three-Arm Phenotype: Neurodevelopmental, Autoinflammatory, and Immunodeficiency Features From 6q27 Deletion**
Dr. Rana Haitham Salman, Pediatrician, Al Qassimi Women and Children's Hospital
- 41 **Uncharted Territory: Defining the Clinical, Radiologic, and Genetic Landscape of Pediatric Congenital Brain Malformations in the UAE**
Dr. Faizah Hashimy (1), Specialist fellow, Tawam Hospital
- 42 **Efficacy of Hydroxychloroquine in Managing Respiratory Complications in Niemann-Pick Disease Type C**
Dr. Faizah Hashimy (2), Specialist fellow, Tawam Hospital
- 43 **Emanuel Syndrome: A Physiotherapy Overview**
Ms. Shahwar Fatima Ansari, Master Student, Institute of Public Health, College of Medicine and Health Sciences, United Arab Emirates University, Al Ain, United Arab Emirates
- 44 **Beyond Common Arrhythmias: Discovering Brugada Syndrome in a Patient With Nonspecific Symptoms**
Dr. Noel George Cherian (2), Medical Intern, Thumbay University Hospital
- 45 **Peri induction Cardiovascular Collapse and Delayed Respiratory Failure in MPS I: Anesthetic Challenges and Successful Epidural Analgesia Despite Severe Scoliosis**
Dr. Thoayba Gaafer Mahgoub Elhassan, PHYSICIAN / SPECIALIST, Al Qassimi Woman Children Hospital
- 46 **A retrospective cross-sectional study on newborn screening and prevalence of disorders among UAE population**
Ms. Khadija Shafique, Genetic Counselor, United Arab Emirates University, Department of genetic and genomics, College of Medicine and Health Sciences
- 47 **a. Dual Chromosomal Imbalance from Familial t(15;20): A Neonatal Case of Monosomy 15q and Trisomy 20q with Severe Multisystem Involvement**
b. Unbalanced translocation with monosomy 15q26.2–qter and trisomy 20q12–qter in a late preterm neonate born to a familial balanced t(15;20): expanding the genotype–phenotype spectrum"
Dr. Sukena Susnerwala, Specialist, Latifa Hospital
- 48 **Refractory cyclic vomiting syndrome in TUBB3 R262H syndrome: a case report**
Dr. Rosul Adel Makkiyah, Pediatrics resident doctor, Al Jalila Children's Speciality Hospital



POSTER PRESENTERS

- 49 **Paired-Like Homeobox 2B (PHOX2B) Mutation and the Hidden Endocrine Puzzle: Hyperinsulinism in Congenital Central Hypoventilation Syndrome**
Dr. Mohamad Sabsabee (1), Pediatric resident, Al Jalial children's Speciality Hospital
- 50 **When One Gland Speaks First: Autoimmune Polyendocrinopathy Syndrome Type 1 (APS-1) Unmasked by Isolated Hypoparathyroidism**
Dr. Mohamad Sabsabee (2), Pediatric resident, Al Jalial children's Speciality Hospital
- 51 **Follistatin as a Potential Biomarker for diabetes in patients with β -thalassemia major: A Cross-Sectional Study**
Dr. Alaa Al Hajji, Medical tutor, University of Sharjah, University of Sharjah
- 52 **Multidisciplinary Management of Severe Hidradenitis Suppurativa in a Specialized Wound Care Service: A Clinical Case Report**
Ms. Aisha Mostafa Hassan (2), Charge nurse/wound management, SKHF hospital
- 53 **Retrospective Study of Clinical and Genetic Profiles of Alpha-Mannosidosis Patients From the UAE**
Mr. Ali Kassem Saad, PhD Student, UAEU
- 54 **A case of report late-onset biotinidase deficiency presenting as neuromyelitis optica spectrum disorder**
Ms. Anna Chudaikina, Clinical Geneticist, Morozov Children's City Clinical Hospital
- 55 **Knowledge, Attitudes, and Perceptions to the Genome Program: A Cross-Sectional Study of UAE Adults**
Dr. Reem Mohamed Alabsi, 4th year medical student, College of Medicine - University of Sharjah
- 56 **Perioperative Management of a Patient with MELAS Syndrome and Hypertrophic Cardiomyopathy Undergoing Ureteroscopic Stone Removal Under Spinal Anaesthesia: A Rare Case Report**
Dr. Shirang Nagorao Bamne, Consultant Critical Care Medicine, Nmc Specialty Hospital, Abu Dhabi
- 57 **Assessing the Current Public Knowledge, Attitudes, and Perceptions Regarding Consanguineous Marriages in the United Arab Emirates**
Dr. Shahd Alali, Medical school graduate, University of Sharjah
- 58 **Beyond a Single Diagnosis: A Child with Coexisting DiGeorge Syndrome and Ataxia-Telangiectasia**
Dr. Sara Mohamed Ali, Department of Pediatrics, Sheikh Shakhbout Medical City (SSMC), Abu Dhabi, United Arab Emirates, Sheikh Shakhbout Medical City (SSMC)
- 59 **From short-read genome sequencing to targeted treatments for rare diseases through the CincyKidsSeq study**
Dr. Anne Slavotinek Professor and Division Director, Human Genetics, Cincinnati Children's Hospital Medical Center, Cincinnati, OH
- 60 **a. Congenital Harlequin Ichthyosis: A Case Report**
Dr. Jini Kottuvala Narayanan, Specialist Pediatrics, LLH Hospital Abu Dhabi, Burjeel holdings
- 61 **Dual Molecular Diagnosis of Late-Onset Leigh Disease and Nephronophthisis Type 3 in an Emirati Woman**
Dr. Fatima AlKindi, Internal Medicine, Sheikh Tahnoon Bin Mohammed Medical City and Tawam Hospital SEHA, PureHealth, Al Ain, UAE"
- 62 **Genetic Molecular background of Prolonged QT syndrome in patients of Al Ain region diagnosed in the years between 2014 and 2024**
Dr. Waed Sumairi, Fellow, Tawam hospital
- 63 **The Role of Pirfenidone in Parkinson's Disease: Mechanisms and Therapeutic Potential**
Ms. Mouza Ali Hasan AlQaishi Alshehhi, PhD student, UAEU collage of medicine
- 64 **Treatment of Reactivation and Progression in Langerhans Cell Histiocytosis (LCH): A 17-Year Single Center Experience from Tertiary Care Hospital in Pakistan**
Dr. Muhammad Rafie Raza, Consultant, Indus Hospital & Health Network, Karachi (IHHN)



POSTER PRESENTERS

- 65 **X-Linked Agammaglobulinemia Masquerading as Hyper-IgE Syndrome: An Unusual Diagnostic Challenge**
Dr. Aysha Dilsha Mullakkantakath, Junior Resident Doctor, Department of Pediatrics, Lady Hardinge Medical College and Kalawati Saran Children's Hospital, New Delhi, India
- 66 **Beyond Malnutrition: A Rare Diagnosis of 5,10-Methenyltetrahydrofolate Synthetase Deficiency in a Child Presenting With Severe Acute Malnutrition and Global Developmental Delay**
Dr. Brenda Syngkon (1), Junior Resident, Department of Paediatrics Lady Hardinge Medical College And Kalawati Saran Children's Hospital, New Delhi, India
- 67 **Unmasking Fanconi Anemia: A Three-Case Series of Clinical Clues and Molecular Diagnosis**
Dr. Brenda Syngkon (2), Junior Resident, Department of Paediatrics Lady Hardinge Medical College And Kalawati Saran Children's Hospital, New Delhi, India
- 68 **Mapping the PAH Variant Landscape in Phenylketonuria Across the Middle East and North Africa: A Systematic Review**
Dr. Nusrat Ferdouse Primary Care Physician, Master's Candidate in Genomic Medicine, UAE university, Alain, UAE
- 69 **Beyond Ocular Disease: Severe Neurodevelopmental Impairment and Neurodegeneration in SIX6-Related Disorder**
Dr. Sana Al-Rawi, Pediatric Specialist, NMC Royal Hospital
- 70 **Prevalence and Characteristics of Factor VIII Inhibitors in Children with Hemophilia A: First National Data from Uzbekistan**
Dr. Dilorom S. Makhmudova, Scientific Secretary, Scientific and Practical Medical Center of Pediatric Oncology, Hematology and Immunology and International Academic Hub for Childhood Cancer, Scientific and Practical Medical Center of Pediatric Oncology, Hematology and Immunology, Tashkent, Uzbekistan
- 71 **The Other Side of CAMT: Unveiling an Exceptionally Rare THPO Mutation**
Dr. Tanisha Negi, Senior Resident, Department of Paediatrics, Kalawati Saran Children's Hospital, LHMC, New Delhi, Department of Paediatrics, Kalawati Saran Children's Hospital, LHMC, New Delhi
- 72 **Transparent Phenotype-Driven Variant Re-Analysis for Unsolved Rare Disease Cases in a Middle Eastern Cohort**
Ms. Fathimath Nubla Latheef, Department of Cancer and Genomic Sciences, University of Birmingham Dubai, Dubai, UAE
- 73 **Diagnostic Delay and Its Determinants in Rare Neuropsychiatric Genetic Disorders in the UAE: A Comparative Analysis with International Literature**
Ms. Fatima Ahmad, B.sc. Molecular and Medical Genetics student, Abu Dhabi University
- 74 **Perioperative outcomes in pediatric hemophilia A on emicizumab prophylaxis: a systematic review and meta-analysis**
Ms. Noor Ismail Abu Tobeh (2), Medical student, University of Sharjah
- 75 **Teaching Machines to Spot Zebras: AI-Assisted Diagnosis in Rare Disease**
Mr. Syed Aasim Syed Nasir (2), 6th Year Medical Student, Faculty of Medicine, Tbilisi State Medical University, Tbilisi, Georgia
- 76 **Successful treatment of genetically proven neonatal diabetes mellitus (NDM): case series from the UAE**
Dr. Juman Babi, Resident Pediatrics, SEHA-SSMC
- 77 **Gamifying Public Health Genomics Education: Development and Pilot Evaluation of the Jackaroo Game for Students and Healthcare Professionals**
Dr. Azhar Talal Rahma, Instructor at the Institute of Public Health, College of Medicine and Health Sciences, UAE