

# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA

CONFERENCE



CONFERENCES PRESIDENT

**PROF. MAJID ALFADHEL**



ASSOCIATION PRESIDENT

**PROF. FATMA ALJISMI**

**CLICK HERE**



[www.mena-mga.org](http://www.mena-mga.org)



**MOVENPICK GRAND ALBUSTAN  
DUBAI UNITED ARAB EMIRATES**

*Save the Date* **8-10 MAY 2025**





# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

Day 1

Thursday 8<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

07:30 - 08:00

Registration 



Time UAE

Topic & Speaker

**Moderator:** Prof. Majid Alfadhel - Prof. Fatma Aljasmí

08:00 - 08:30

**Opening Ceremony**  
MENA-MGA

08:30 - 08:40

**MENA-MGA who we are? and by-law of the organization**  
Prof. Fatma Aljasmí - UAE 

08:40 - 09:00

**Establishment of different committees:**  
- Genetic counselors committee  
- Guidelines committee  
- Metabolic Nutritions committee  
- Genomic committee  
MENA-MGA Board

09:00 - 09:20

**MENA-MGA current status and future plan**  
Discussion Panel MENA-MGA Board

09:20 - 09:45

**PhenoDB, GeneMatcher and VariantMatcher: Phenotypic and Genomic FederatedData Sharing To Facilitate Variant Classification and Disease Gene Discovery**  
Prof. Nara Sobreira - USA 

09:45 - 10:10

**Integrating Genomic Medicine Into Dubai's Academic Health Care System**  
Dr. Ahmad Abou Tayon - UAE 

10:10 - 10:40

**Rare Diseases and Precision Medicine: The Successive Paradigm**  
Prof. Tawfeg Ben-Omran - Qatar 

10:40 - 10:50

**Discussion**





# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

*Day 1*

Thursday 8<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

10:50 - 11:15

Coffee Break



Time UAE

Topic & Speaker

**Moderator:** Dr. Fuad Al Mutairi - Dr. Mohammed Almannai

11:15 - 11:35

**The RNA Exosome and Human Disease**

Almundher Al-Maawali - Oman 

11:35 - 11:55

**Whole Exome Sequencing for Fetal Structural Anomalies: Expanding the Diagnostic Yield in Prenatal Care - Royal Hospital Experiences**

Dr. Nadia Alhashmi - Oman 

11:55 - 12:15

**Unexplained Joint Pain: Rare Answers to Common Symptoms**

Dr. Ghada Ahmed Otaify - Oman 

12:15 - 12:25

**Discussion**

12:25 - 13:30

**Lunch Break**

**Moderator:** Prof. Moeenaldeen AlSayed - Dr. Almundher Al-Maawali

13:30 - 13:55

**POLG-related Disorders: Clinical and Molecular Phenotypic Features in Saudi Arabia**

Dr. Fuad Al Mutairi - KSA 

13:55 - 14:20

**SLC25A42-related Mitochondrial Disorder: an Update**

Dr. Mohammed Almannai - KSA 

14:20 - 14:45

**The Morbid Nuclear and Mitochondrial Genome of Mitochondrial Diseases in GCC Region**

Dr. Naif Almontashiri - KSA 



# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

*Day 1*

Thursday 8<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

14:45 - 15:15 | Coffee Break



**Moderator:** Dr. Nawal Makhseed - Dr. Khalid Al-Thihli



Time UAE

Topic & Speaker

15:15 - 15:35

**Progress in Strategies to Overcome Blood-Brain Barrier for Rare Diseases with Neurological Manifestations**

Prof. Maurizio Scarpa - Italy 

15:35 - 16:00

**Combined Pituitary Hormone Deficiencies: Identification of Novel Gene associated with Neuroendocrine Syndrome**

Prof. Malak Alghamdi - KSA 

16:00 - 16:25

**Beyond the Surface: Unraveling Gaucher vs ASMD Disease**

Dr. Hamda Abulhoul - UAE 

16:25 - 16:35

**Discussion**

18:00 - 20:00

**Gala Dinner  
All attendees**

20:00 - 20:05

**Closure of the first day**





# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

*Day 2*

Friday 9<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

08:30 - 09:00

*Registration* 



Time UAE

WORKSHOP

09:00 - 10:30

**Genomic database: A Practical Guide for Clinicians Workshop**  
(by Center of Arab Genomic Studies (CAGS))

Kindly ensure that you bring your laptop for Workshop  
First come first served Only for 40 attendees



Al Majlis Meeting Room

09:00 - 09:30

**Introduction to Genomics and Genetic Databases**

Pratibha Nair - UAE 

09:30 - 10:00

**Hands-on training**

Sami Bizzari - UAE 

10:00 - 10:30

**Discussion and conclusion**

10:30 - 11:00

**Coffee Break**





# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

*Day 2*

Friday 9<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

**Moderator:** Prof. Malak Alghamdi - Dr. Hiba Al Bloushi



Time UAE

Topic & Speaker

09:00 - 09:25

**Leveraging Machine Learning Models for Early Diagnosis of Rare Genetic Disorders: A Case Study on Fabry Disease**

Dr. Muhammad Jalal Khan - 

09:25 - 09:50

**Delineating the Cellular Mechanisms of Endoplasmic Reticulum-Retained Endoglin Mutants Causing Hereditary Haemorrhagic Telangiectasia Type 1**

Dr. Nesrin Gariballa - UAE 

09:50 - 10:20

**Therapeutic Benefit Uncovered through anecdotal evidence: Two Exemplary Cases from the Clinic**

Dr. Khalid Al-Thihli - Oman 

10:20 - 10:30

**Discussion**

10:30 - 11:00

**Coffee Break**







# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

*Day 2*

Friday 9<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

**Moderator:** Dr. Hamda Abulhoul - Dr. Maryam AlShehhi



Time UAE

Topic & Speaker

11:00 - 11:25

**Living Without Adipose Tissue: Uncovering the Genetic and Metabolic Puzzle of Congenital Lipodystrophy**

Dr. Jamal Aljubeih- UAE



11:25 - 11:50

**Evaluation of GeneReviews as a Resource for Rare Disease Treatment and Management for Machine Learning Applications: A Case Study on Inborn Errors of Metabolism (IEMs)**

Ms. Tabeer Fatima - UAE



11:50 - 12:20

**Future therapies for metabolic conditions: the race between gene editing and small molecules**

Prof. Johannes Häberle - Germany



12:20 - 12:45

**Precision in Prediction: Tailoring Machine Learning for Breast Cancer Missense Variants Integrating Dataset Selection with Model Interpretability for Pathogenicity Prediction**

Dr. Rahaf M. Ahmad - UAE



12:45 - 12:55

**Discussion**

12:55 - 14:00

**Friday Prayer & Lunch break**



# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

Day 2

Friday 9<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

**Moderator:** Dr. Nuha Alzaabi - Dr. Amal Altenaiji



Time UAE

Topic & Speaker

14:00 - 14:25

**The Power of Mendelian Genetics for Target Identification and Drug Development**

Prof. Bruno Reversade - KSA



14:25 - 14:55

**Transport Vehicle TM (TV) Platform and Tividenofusp Alpha in Treatment of MPS II**

Dr. Daniel Fertek - Switzerland



14:55 - 15:20

**How Can a Routine ALP Test Lead to Diagnosis of Rare Disease**

Dr. Mariam AIShehhi - UAE



15:20 - 15:50

**Update on clinical trials for inborn errors of metabolism**

Prof. Johannes Häberle - Germany



15:50 - 16:15

**GenRare: An AI Assistant Tool for Rare Genetic Diseases: Early Diagnosis and Management**

Dr. Aamna AIShehhi - UAE



16:15 - 16:30

**Discussion**

16:30 - 17:00

**Closure of the second day**





# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

Day 3

Saturday 10<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

08:30 - 09:00

Registration 

**Moderator:** Dr. Fatima Alali - Dr. Sara Belfaqeeh



Time UAE

Topic & Speaker

09:00 - 09:20

**Elucidating The Molecular Mechanisms of Human Monogenic Diseases Associated with *TGFB2***

Dr. Fadia Abu-Sailik - UAE 

09:20 - 09:40

**Cultural Dimensions in Genomic Science**

Dr. Hibat Omer - UAE 

09:40 - 09:50

**Discussion**

09:50 - 10:20

**Coffee Break**





# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

Day 3

Saturday 10<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

**Moderator:** Dr. Ali Al Beshri - Dr. Nadia Akawi



Time UAE

Topic & Speaker

10:20 - 10:40

**From Data to Decision: QPHI Experience**

Ms. Tasnim A. Fadl - Qatar 


10:40 - 11:00

**Hartnup Disease-Causing *SLC6A19* Mutations Lead to BOAT1 Aberrant Trafficking and ACE2 Mis-localisation Implicating the Endoplasmic Reticulum Protein Quality Control**

Dr. Nesreen Al Khofash - UAE 

11:00 - 11:20

**From Genetic Diagnosis to Prevention: The Impact of PGTA and PGTM on Reproductive Health**

Dr. Qamre Alam - Bahrain 

11:20 - 11:50

**At three hospitals done at different countries urine organic acids failed to diagnose a rare metabolic disease for 18 years**

Dr. Nawal Makhseed - Kuwait 

11:50 - 12:00

**Discussion**

12:00 - 13:00

**Lunch Break**







# MENA-MEDICAL GENETICS ASSOCIATION

## MENA-MGA CONFERENCE

*Day 3*

Saturday 10<sup>th</sup> May 2025

Clinical, Biochemical and Laboratory Genetics

**Moderator:** Dr. Nadia Alhashmi - Prof. Buruno Reversade



Time UAE

Topic & Speaker

13:00 - 13:30

**From gene to hope: Breakthrough therapies for the leukodystrophies**  
Dr. Laura Adang - USA

13:30 - 14:00

**Assessment of urea cycle function by pathway analysis using stable isotopes**  
Prof. Johannes Häberle - Germany

14:00 - 14:30

**Aicardi-Goutières syndrome (AGS) in the clinic: when genetic disease look like infection**  
Dr. Laura Adang - USA

14:30 - 14:40

**Discussion**

14:40 - 14:55

**Coffee Break**



**Moderator:** Prof. Majid Alfadhel



Time UAE

Topic & Speaker

14:55 - 15:05

**Award for the best poster**  
Prof. Johannes Häberle - Prof. Fatma Aljasmi

15:05 - 15:50

**Registration for MENA-MGA, Comments from audiences regarding MENA-MGA future plans**  
Discussion panel: MENA-MGA Board

15:50 - 16:00

**Closure of The Conference**