

CRASH
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1st MEGMA

BASIC GENETICS AND INBORN ERRORS OF
METABOLISM DIAGNOSTICS AND THERAPEUTICS



5-6 JUNE
2026

HYATT REGENCY RIYADH OLAYA HOTEL

Hybrid Conference
Physical **Free** of Charge

VIRTUAL 500 SAR



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TARGET AUDIENCE

1

**Medical Genetics
Fellows and
Consultants**

2

**Clinical Lab
Scientists**

3

**Genetic Counselors
and Nurses**

4

**Pediatrician
and Neurologists**

5

Physicians



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OBJECTIVES:

1. Education about advanced therapeutic options for genetic defects and inborn errors of metabolism disorders, including enzyme replacement and gene therapies.
2. Know the basic genetics concepts, common genetics syndrome.
3. Identify clinical signs and symptoms of common Inborn Errors of Metabolism (IEM).
4. Training on Bayesian analysis to assess genetic risk.
5. Interpret biochemical laboratory tests for accurate diagnosis including tandem MS, plasma amino acids and urine for organic acids.
6. Interpret Cytogenetics laboratory tests for accurate diagnosis including karyotype, FISH and CGH microarray.
7. Interpret molecular laboratory tests for accurate diagnosis including whole exome sequencing (WES) and whole genome sequencing (WGS).
8. Execute emergency management plans for acute metabolic decompensation.
9. Provide effective genetic counseling and ethical guidance to affected families.



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SPEAKERS



Prof. Majid Alfadhel



Prof. Malak Alghamdi



Prof. Fuad Almutairi



Prof. Zuhair Al-Hassnan



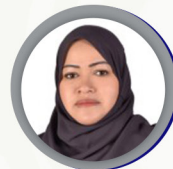
Dr. Abdullah Alfalah



Dr. Ahmed Alodaib



Dr. Ali Tabish



Dr. Aqeela Alhashim



Dr. Bader Almuzaini



Dr. Hajar Alakeel



Dr. Hamad Althiyab



Dr. Maha Alotaibi



Prof. Mohammed Al-Dubayee



Dr. Mohammed Almuqbil



Dr. Mohammed AlMannai



Dr. Muhammad Umair



Dr. Najim Ameziane

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Day1
Friday 5th June

Clinical, Biochemical and Laboratory genetics

07:30 - 08:30 *Registration*

08:30 - 09:00 | **Opening Remarks**
Prof. Majid Alfadhel

09:00 - 09:20 | **Important Concepts in Genetics**
Dr. Mohammed Almuqbil

09:20 - 09:40 | **Syndromes that Every Geneticist Should Know**
Dr. Mohammed Almuqbil

09:40 - 10:00 | **Rare Lipid Disorders in Pediatric Patients**
Prof. Mohammed Al-Dubayee

10:00 - 10:10 | **Discussion**

10:10 - 10:25 | **Coffee break**

10:25 - 10:45 | **Clinical Cytogenetics -1**
Dr. Bader Almuzaini

10:45 - 11:05 | **Clinical Cytogenetics -2**
Dr. Bader Almuzaini

11:05 - 11:25 | **Updates in the Management of Primary Hyperoxaluria Type 1**
Prof. Majid Alfadhel

11:25 - 11:30 | **Discussion**

11:30 - 13:10 | **Friday Prayer and Lunch Break**





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Day1
Friday 5th June

Clinical, Biochemical and Laboratory genetics

- | | |
|---------------|--|
| 13:10 - 13:30 | Metachromatic Leukodystrophy (MLD): Disease Burden and Recent Therapeutic Advances
Dr. Mohammed Almuqbil |
| 13:30 - 13:55 | Molecular Genetics - 1
Dr. Abdullah Alfalah |
| 13:55 - 14:20 | Molecular Genetics - 2
Dr. Abdullah Alfalah |
| 14:20 - 14:45 | Bayesian Analysis and Risk Assessment
Dr. Hajer Alaqeel |
| 14:45 - 15:05 | Restoring Hope: The Therapeutic Journey in AADC Deficiency
Dr. Aqeela Alhashim |
| 15:05 - 15:15 | Discussion |
| 15:15 - 15:30 | Coffee break |
| 15:30 - 15:55 | Newborn Screening and Tandem Mass Spectrometry (TMS) - 1
Dr. Mohammed Almanai |
| 15:55 - 16:20 | Plasma Aminocids - 1
Prof. Fuad Almutairi |
| 16:20 - 16:45 | Plasma Aminocids - 2
Prof. Fuad Almutairi |
| 16:45 - 16:55 | Discussion |
| 16:55 - 17:00 | Closure Of The First Day
Prof. Majid Alfadhel |

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Day 2

Saturday 6th June

Clinical, Biochemical and Laboratory genetics

- | | |
|---------------|---|
| 08:30 - 08:55 | Newborn screening and Tandem Mass Spectrometry (TMS)-2
Dr. Mohammed Almannai |
| 08:55 - 09:20 | Urine for Organic Acids -1
Dr. Ahmed Alodaib |
| 09:20 - 09:45 | Urine for Organic Acids-2
Dr. Ahmed Alodaib |
| 09:45 - 10:10 | The Art of Molecular Matchmaking: Primer Design how to do it?
Dr. Muhammad Umair |
| 10:10 - 10:20 | Discussion |
| 10:20 - 10:40 | Coffee break |
| 10:40 - 11:05 | Biochemical Genetics -1
Prof. Malak Alghamdi |
| 11:05 - 11:30 | Biochemical Genetics -2
Prof. Malak Alghamdi |
| 11:30 - 11:55 | Update on the Diagnosis and Treatment of Inborn Errors of Metabolism
Prof. Majid Alfadhel |
| 11:55 - 12:15 | Interpretation of WGS/WES
Dr. Najim Ameziane |
| 12:15 - 12:25 | Discussion |
| 12:25 - 13:30 | Lunch break |





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Day 2

Saturday 6th June

Clinical, Biochemical and Laboratory genetics

- | | |
|---------------|--|
| 13:30 - 13:50 | Basic Genetics-1
Prof. Zuhair Alhassnan |
| 13:50 - 14:10 | Basic Genetics-2
Prof. Zuhair Alhassnan |
| 14:10 - 14:30 | Whole Exome Sequencing and Whole Genome Sequencing Interpretation
Dr. Ali Tabish |
| 14:30 - 14:50 | Lysosomal Storage Disorders
Prof. Malak Alghamdi |
| 14:50 - 15:20 | LC-FAOD Overview
Dr. Maha AlOtaibi |
| 15:20 - 15:30 | Discussion |
| 15:30 - 15:45 | Coffee break |
| 15:45 - 16:05 | Reproductive Genetics
Prof. Majid Alfadhel |
| 16:05 - 16:25 | MCQ Preparation Tips
Dr. Hamad Althyab |
| 16:25 - 16:45 | Hunter Syndrome (MPS II): Disease Burden and Recent Therapeutic Advances
Dr. Mohammed Almannai |
| 16:45 - 17:05 | Epidermolysis Bullosa Highlights
Dr. Hamad AlThiyab |
| 17:05 - 17:15 | Discussion |
| 17:15 - 17:30 | Closure Of The Course
Prof. Majid Alfadhel |



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Prof. Majid Alfadhel

Chairman, Genetics & Precision Medicine Department, Professor of Medical Genetics, King Abdullah Specialized Children Hospital (KASCH), King Saud bin Abdulaziz University of Health Sciences (KSAU-HS), King Abdulaziz Medical City (KAMC), Ministry of National Guard Health Affairs (MNG-HA), Riyadh, Saudi Arabia

Prof. Alfadhel is currently Deputy Executive Director of King Abdullah International Medical Research Centre (KAIMRC), Chairman of Genetics and Precision Medicine department and Director of Medical Genomic lab and Professor of Pediatrics and Genetics at King Saud bin Abdulaziz University for Health Sciences (KSAU-HS). He contributed to discovery of more than 70 novel genes and link it for the first time to a human disease, in addition, he discovered two syndromes, first one with with dysmorphic features, intellectual disability, and speech delay, which named Alfadhel syndrome due to a defect in RAPIGDS1 gene and other novel neurodevelopmental disease with poor or absent speech, dysmorphic facies, and behavioral abnormalities named Umair-Alfadhel Neurodevelopmental disorder due to a defect in NAV3 gene. Both registered in the OMIM database, the official website for all genetic disorders. Additionally, he is founder and editor in chief of Journal of Biochemical and Clinical Genetics, Editorial board member of several other international journals. Authors of three books and one chapter in a book. Has more than 250 publications in high impact factor journals and more than 200 abstracts. He is supervisor and teachers for plenty of fellows, residents and medical students. His foresight and passion for innovation, earned him an award for inventing a new application in the Apple store and Goggle play for the treatment of inborn errors of metabolism (IEM Drugs). Finally, Prof. Alfadhel has been recognized with two of the highest prestigious awards in genetics: the Canadian College of Medical Geneticist (CCMG) Founders' Award for Career Achievement (2024) and the Arab Award in Genetics (2026).

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Prof. Malak Alghamdi

Senior Medical Genetics Consultant & Professor, Medical Genetics Division, Department of Pediatrics, King Khalid University Hospital (KKUH), King Saud University, Riyadh, Saudi Arabia.

Prof. Malak earned her MBBS degree from King Saud University and holds Arab and Saudi Board certifications in Pediatrics, in addition to Canadian Board certification in Medical Genetics (FCCMG, Canada).

Prof. Alghamdi completed a fellowship in Clinical Biochemical Genetics at British Columbia Children's Hospital, Canada. She established the clinical and academic Medical Genetics Division at KSUMC, with a strong focus on neurogenetic disorders and inborn errors of metabolism.

Her academic and clinical contributions include plenty of publications in the field of medical genetics. She is also actively involved in mentoring fellows, residents, and medical students, supporting the advancement of genetics education and training in Saudi Arabia.



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Prof. Fuad Almutairi

Senior Medical Genetics Consultant & Professor, Genetics and Precision Medicine Department (GPM), King Abdullah Specialized Children Hospital (KASCH), King Saud bin Abdulaziz University of Health Sciences (KSAU-HS), King Abdulaziz Medical City (KAMC), Ministry of National Guard Health Affairs (MNG-HA), Riyadh, Saudi Arabia.

Prof. Fuad Almutairi earned his MBBS degree from King Saud University and completed his Pediatrics Residency at Prince Sultan Military Medical City (PSMMC). He later pursued fellowship training in Biochemical Genetics at the University of British Columbia, Vancouver, Canada.

Prof. Almutairi is actively involved in clinical genetics education and training and currently chairs the National Medical Genetics Fellowship Program at the Saudi Commission for Health Specialties (SCFHS). He is also a member of the Board of Directors of the Saudi Society of Medical Genetics and contributes to several regional and international scientific initiatives. His research interests focus on neurogenetic disorders, inborn errors of metabolism, and the discovery of novel disease-associated genes through local and international collaborations. He has published numerous scientific articles in high-impact journals and continues to contribute extensively to research and precision medicine initiatives in Saudi Arabia.



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Prof. Zuhair Al-Hassnan

Senior Medical Genetics Consultant & Professor, Department of Medical Genomics Genomic Medicine Centre, Alfaisal University, King Faisal Specialist Hospital & Research Centre(KFSHRC), Riyadh, Saudi Arabia.

Prof. Zuhair completed his fellowship in Medical Genetics at Johns Hopkins University and holds multiple board certifications in clinical biochemical and molecular genetics.

Prof. Al-Hassnan has extensive expertise in clinical genomics, newborn screening, and rare genetic disorders. He leads several national and international research initiatives and actively contributes to policy development and ethics in genomic medicine. He is the chairman of Saudi National Committee for Genetic Diseases. Throughout his career, he has received numerous awards recognizing his achievements in genetic research and clinical innovation in Saudi Arabia. He has published numerous scientific articles in high-impact journals and continues to contribute extensively to research in Saudi Arabia.



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Prof. Mohammed Al-Dubayee

Senior Pediatric Endocrinology consultant & Professor, Department of Pediatrics, King Abdullah Specialized Hospital - Al Qasim, Saudi Arabia.

Professor Mohammed Al-Dubayee is a Saudi physician specializing in Pediatric Endocrinology and Diabetes, with a special interest in lipid disorders, inherited metabolic diseases, and precision medicine in children and adolescents. He is actively involved in advancing pediatric endocrine and metabolic healthcare services in Saudi Arabia through clinical practice, academic leadership, research, and medical education. Professor Al-Dubayee has contributed to several scientific and collaborative initiatives focused on pediatric diabetes, dyslipidemia, obesity, and genomic medicine, with particular interest in improving early diagnosis and long-term outcomes for children with rare and hereditary metabolic disorders. He is also actively engaged in postgraduate medical training, healthcare quality improvement, and the development of specialized pediatric service.



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Dr. Mohammed AlMannai

Senior Medical Genetics Consultant, Genetics and Precision Medicine Department (GPM), King Abdullah Specialized Children Hospital (KASCH), King Saud bin Abdulaziz University of Health Sciences (KSAU-HS), King Abdulaziz Medical City (KAMC), Ministry of National Guard Health Affairs (MNG-HA), Riyadh, Saudi Arabia.

Dr. Mohammed AlMannai completed his Medical Doctor (MD) degree at Arabian Gulf University. He completed his medical internship and Pediatrics Residency at Salmaniya Medical Complex, Ministry of Health, Bahrain, before continuing his pediatric training at the University of South Alabama. He later received specialty training in Medical Genetics and Medical Bio-chemical Genetics at Baylor College of Medicine.

Dr. AlMannai currently serves as a Consultant in the Genetics and Precision Medicine Department at King Abdulaziz Medical City, Riyadh, and is the Program Director of the Clinical Genetics and Genomics Fellowship Program at the same institution.

He is actively involved in scientific research and has contributed to numerous publications in the fields of medical genetics and genomic medicine.

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Dr. Mohammed Almuqbil

Pediatric Neurology & Neurogenetics Consultant & Associate Professor, King Abdullah Specialized Children Hospital (KASCH), King Saud bin Abdulaziz University of Health Sciences (KSAU-HS), King Abdulaziz Medical City (KAMC), Ministry of National Guard Health Affairs (MNG-HA), Riyadh, Saudi Arabia.

Dr. Mohammed Almuqbil is a Consultant in Pediatric Neurology and Neuro-genetics at King Abdullah Specialized Children's Hospital (KASCH), Riyadh, Saudi Arabia, and an Associate Professor at King Saud bin Abdulaziz University for Health Sciences (KSAU-HS).

He specializes in pediatric neurological and neurogenetic disorders, with expertise in medical genetics, biochemical genetics, and rare neurological diseases.

Dr. Almuqbil completed his Pediatric Neurology residency at McGill University in 2016, followed by board training in Medical Genetics and Genomics at Harvard Medical School in 2018, and a fellowship in Medical Biochemical Genetics at Johns Hopkins University in 2019.

He is actively involved in clinical research, medical education, and international scientific collaborations, with numerous publications in high-impact journals focusing on neurogenetics, mitochondrial disorders, leukodystrophies, and rare genetic diseases.

Dr. Almuqbil also serves on the boards of several scientific societies and contributes extensively to resident and fellow training programs.



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Dr. Maha AlOtaibi

**Medical Genetics & Genomic Medicine Consultant, King Saud Medical City,
Riyadh, Saudi Arabia.**

Dr. Maha is actively involved in clinical genetics practice and precision medicine initiatives, with a strong focus on improving diagnostic pathways for patients with rare and hereditary disorders.

Dr. AlOtaibi has contributed to several research projects and scientific publications in the fields of neurogenetics, metabolic disorders, and genomic medicine. She also participates in national initiatives related to genetic disease awareness and newborn screening programs.



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Dr. Abdullah Alfalah

Medical and Molecular Genetics Consultant, Riyadh, Saudi Arabia.

Dr. Abdullah Alfalah is a physician specializing in medical and molecular genetics, with particular expertise in rare diseases, genomic diagnostics, and clinical genomics.

He is actively involved in genomic research and multidisciplinary rare disease initiatives, with a focus on advanced genomic interpretation, reanalysis strategies, and improving diagnostic yield in complex genetic disorders.

Dr. Alfalah has contributed to several research projects and scientific presentations in the fields of pediatric genetics, neurogenetics, and precision medicine. He is also passionate about integrating genomics into clinical care and advancing medical education in the field of genetics.



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Dr. Ahmed Alodaib

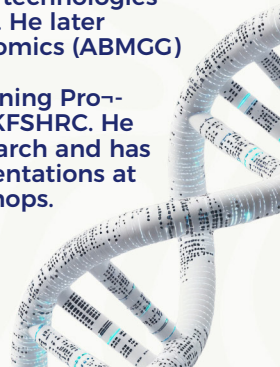
Clinical Scientist Consultant in Clinical Biochemistry & Biochemical Genetics, Head of Metabolomics Section, Genomic Medicine Centre, King Faisal Specialist Hospital & Research Centre(KFSHRC), Riyadh, Saudi Arabia.

Dr. Alodaib is a Consultant Clinical Scientist in clinical biochemistry and biochemical genetics and currently serves as the Head of the Metabolomics Section, including the Biochemical Genetics Unit, within the Precision Medicine Laboratory Department at the Genomic Medicine Center of Excellence at King Faisal Specialist Hospital & Research Centre (KFSHRC). He is also an Adjunct Assistant Professor at the University of Pittsburgh and Alfaisal University.

Dr. Alodaib graduated from the College of Science, Department of Biochemistry at King Saud University in 2003. In 2008, he completed his Bachelor's Honors degree in Health Science at the University of Melbourne, followed by a Master's degree in Human Genetics from the University of Sydney in 2010.

In 2015, he earned his PhD in Human Genetics from the University of Sydney, where he utilized advanced next-generation sequencing technologies to identify disease-causing genes in rare Mendelian disorders. He later completed the American Board of Medical Genetics and Genomics (ABMG) Clinical Fellowship Training in Biochemical Genetics in 2019.

Dr. Alodaib is the Founder and Director of the Fellowship Training Program in Clinical Biochemical Genetics (Laboratory-based) at KFSHRC. He has played an active role in regional and national health research and has published extensively in leading scientific journals, with presentations at numerous national and international conferences and workshops.



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Dr. Ali Tabish

Medical Genetics Consultant, Department of Pathology and Laboratory Medicine, King Abdulaziz Medical City (KAMC), Ministry of National Guard Health Affairs (MNG-HA), Riyadh, Saudi Arabia.

Dr. Ali hold an MD and PhD with a strong academic and clinical focus on clinical genetics and genomic medicine.

He has a special interest in advancing the application of genomics in clinical practice, with a focus on improving the diagnosis and management of in-herited and complex genetic disorders. His work is centered on integrating modern genomic technologies into patient care to support precision medicine and enhance clinical outcomes.

Dr. Tabish is passionate about the field of genomics and continues to contribute to the growing understanding of genetic diseases through clinical practice and scientific engagement. He is committed to advancing genetic healthcare services and supporting the development of precision medicine approaches in the region.



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Dr. Aqeela Alhashim

Pediatric Neurologist & Neurogeneticist Consultant, National Neuroscience Institute, King Fahad Medical City, Riyadh, Saudi Arabia

Dr. Aqeela earned her MBBS degree from King Faisal University and completed her Pediatric Residency training in Riyadh, followed by the Saudi and Arab Boards in Pediatrics. She later obtained the Fellowship of the Royal College of Physicians and Surgeons of Canada (FRCPC) in Child Neurology and completed a Neurogenetics Fellowship in Clinical and Molecular

Genetics Research at The Hospital for Sick Children.

Dr. Alhashim has extensive expertise in pediatric neurology, neurogenetics, and rare neurological disorders. She is actively involved in clinical research, medical education, and international scientific collaborations, with multiple publications and presentations in the fields of neurogenetics and inherited neurological diseases.

She has also participated as a speaker and faculty member in numerous regional and international conferences focused on pediatric neurology and rare diseases.



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Dr. Bader Almuzaini

Clinical Scientist Consultant in Medical Genetics, Public Health Authority (PHA), Riyadh, Saudi Arabia.

Dr. Bader Almuzaini is a certified Clinical Laboratory Consultant and Research Scientist with more than twenty years of experience in diagnostic conventional cytogenetics, genomic and epigenomic research, including whole genome sequencing (WGS), transcriptome analysis, microarray technologies, and molecular diagnostics.

Dr. Almuzaini earned his PhD in Cell and Molecular Genetics from the Karolinska Institute and holds a Master's degree in Medical Genetics from the University of Glasgow.

His research focuses on identifying genetic and epigenetic signatures underlying cancer genetics and rare genetic disorders. He has led and co-investigated several funded research projects and has extensive scientific publications in genomics, molecular pathology, and precision medicine. In addition to his research and clinical work, Dr. Almuzaini is actively involved in teaching, mentorship, and national scientific committees related to laboratory medicine, genomics, and healthcare quality standards in Saudi Arabia.



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Dr. Hajar Alakeel

Medical and Molecular Genetics Consultant, Riyadh, Saudi Arabia.

Dr. Hajar Alakeel is a board-certified geneticist and genomic medicine consultant currently undertaking her second fellowship in Clinical Molecular Genetics and Genomics at King Faisal Specialist Hospital & Research Centre (KFSHRC), following the completion of her Clinical Genetics and Genomics Fellowship at King Abdullah Specialized Children's Hospital (NGHA). Prior to her subspecialty training, she completed her pediatric residency at NGHA and earned the Saudi Board of Pediatrics certification in 2021.

Throughout her training, Dr. Alakeel has been recognized for her clinical and academic excellence, receiving multiple distinctions, including the Best Resident Award. Her research has been presented at leading international conferences, including the American Society of Human Genetics (ASHG), the European Society of Human Genetics (ESHG), as well as regional MENA meetings.



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Dr. Hamad Althiyab

Medical Genetics Consultant, Genetics and Precision Medicine Department (GPM), King Abdullah Specialized Children Hospital (KASCH), King Abdulaziz Medical City (KAMC), Ministry of National Guard Health Affairs (MNG-HA), Riyadh, Saudi Arabia.

Dr. Hamad Althiyab earned his Bachelor of Medicine and Bachelor of Surgery (MBBS) from King Saud bin Abdulaziz University for Health Sciences (KSAU-HS).

He completed his Pediatrics Residency and obtained the Saudi Pediatric Board certification, followed by a Clinical Genetics and Metabolic Disorders Fellowship accredited by the Saudi Commission for Health Specialties (SCFHS). He later completed a Biochemical Genetics Fellowship at BC Children's Hospital, Canada.

Dr. Althiyab has contributed to several scientific publications and research projects in the fields of metabolic and genetic disorders, including leukodystrophies, neuronal ceroid lipofuscinosis, and rare neurodevelopmental syndromes.

He is actively involved in academic activities, committee work, and continuing medical education, with presentations delivered at regional and international conferences related to inborn errors of metabolism and pediatric genetics. His professional interests focus



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Dr. Muhammad Umair

Research Scientist, Team Leader, Functional Studies, Human Molecular Genetics & Rare Genetic Disorders Specialist, King Abdullah International Medical Research Centre (KAIMRC), King Abdulaziz Medical City (KAMC), Ministry of National Guard Health Affairs (MNG-HA), Riyadh, Saudi Arabia.

Dr. Umair research focuses on novel gene discovery, neurodevelopmental disorders, skeletal disorders, and translational genomics using advanced next-generation sequencing technologies.

Dr. Umair has published more than 200 peer-reviewed scientific articles and has contributed to the identification of 27 novel disease-associated genes. He also serves as an editor and reviewer for several international scientific journals. His work integrates clinical genomics, functional characterization, and precision medicine approaches to improve the diagnosis and understanding of rare human genetic disorders.



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Dr. Najim Ameziane

Senior Director Bioinformatics | Molecular Biologist with Self-Taught Bioinformatics Expertise

Najim Ameziane is a passionate molecular biologist and bioinformatics expert with extensive leadership experience in developing innovative methods and advanced tools for clinical molecular genetic testing. He has successfully led high-performing teams of bioinformaticians and data scientists, optimizing workflows for maximum efficiency and implementing forward-thinking strategies to support organizational growth and scientific advancement.

His expertise spans genomic research, drug development, and advanced sequencing technologies, including next-generation sequencing (NGS), whole genome sequencing (WGS), and RNA sequencing (RNA-seq). Through his multidisciplinary experience, he continues to contribute to the advancement of precision medicine and translational genomics.





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