Message from the Chair, Organizing Committee
Dr Aysha Habib Khan, Professor,
Pathology and Laboratory Medicine

Rare diseases affects many aspects of an individual's life including their social, educational and employment opportunities and is an emerging global public health priority. When taken together, "rare" diseases are not so rare after all. According to an estimate around 300 million people, i.e. 4% of the world's population suffers from these conditions. Therefore public health policies at global and national level are needed to address this issue. Such a policy is becoming reality in many countries around the globe. However, non-availability of diagnostic facilities, delayed diagnosis, misdiagnosis, difficulty accessing appropriate information, difficulty accessing care, availability of experts help and poorly coordinated care are still the norm for rare disease patients and their families in Pakistan.

In this scenario, it is important for us as a nation to think that how can we form collaborations to facilitate resource development? As the resources for development are never enough, what market solutions can we create to unleash the potential of technology? What is the role of impact investing in the times of constrained development funds? In the era of tech boom, what can we do to make technology an integral part of the solutions and opportunities? How can we come together to create partnerships and synergies that will have a multiplier effect on the impact in the knowledge and skills in the area of r are disease diagnosis and management and how can we smartly and fully address the question in everyone's mind that what's in it for me? It is important to respect this question for the synergies we want to create.

In experience, no one organisation owns, nor can claim to own all the answers, but many can contribute when they consider it as a true strategic focus at the heart of healthcare. Together, we can find our ways to achieving all the outcomes we aspire to achieve and know are possible in rare disease diagnosis and management for patient safety.

Synergies and impact is about addressing these questions for our most valuable asset; newborns, neonates and infants, by bringing together partnerships and use of technology and data to drive the market solutions. In experience, every voice matters, and each of those individual voices contribute to an ocean of ripples that can positively impact countless lives.

The goal of this conference is to break silos and understand these issues and come up with concrete steps that can be implemented within tight timeframes after the conference. So here I invite you to come, let us reframe rare in Pakistan. Let's grow together and make impact, manifold! If brought together to collaborate with minimum vested interests and with a drive towards a common goal. The results can be enormous. That power of togetherness is hugely impactful.

I am thankful to all the guests who have gathered here upon our invitation. I must also thank our collaborating partners and contributors, who have helped make this event possible. Finally, I must congratulate the Organising Committee for their efforts.

Multidisciplinary Conference on Rare Genetic Diseases in Pakistan
Organised by Department of Pathology & Laboratory Medicine in collaboration with Division of Women & Child Health, the Aga Khan University (AKU), Karachi

Collaborators:
Department of Biological & Biomedical Science, AKU
Department of Obstetrics and Gynaecology, AKU

National Societies:
Pakistan Society of Chemical Pathologists (PSCP)
Pakistan Medical Association (PMA)
Pakistan Pediatric Association (PPA)
Pakistan Society of Hematology (PSH)
Pakistan Inherited Metabolic Diseases Network (Pak IMD-Net)

International:
International Federation of Clinical Chemistry & Laboratory Medicine (IFCC)

Institutes:
Ziauddin University, Karachi
National Institute of Child Health (NICH), Karachi
Dow University of Health Sciences (DUHS), Karachi
National Institute of Blood Disorders (NIBD), Karachi
Liaquat National Hospital, Karachi
The Indus Hospital, Karachi
The Children's Hospital and Institute of Child Health, Lahore
University of Iowa Health Care, USA
Auckland District Health Board, New Zealand
Meyer Children's Hospital, Florence, Italy

Patron:
Prof Mohammad Khurshid

Chief Guest
Prof Abdul Gaffar Billoo

Organizing Committee
Chair: Prof Aysha Habib Khan
Co-Chair: Dr Muhammad Sohail Salat
Members:
Prof Bushra Moiz
Dr Farooq Ghani
Dr Fyezah Jehan
Prof Imran Siddiqui
Dr Salman Kirmani
Dr Arit Prakash

Scientific Committee
Chair: Dr Lena Jafri
Co-Chair: Dr Bushra Afroze
Members:
Dr Fatima Kanani
Dr Ghulam Zainab
Dr Hafsa Majid
Dr Asghar Nasir
Dr Rehana Rehman
Dr Sahar Iqbal
Dr Sibtain Ahmed
Ms Shahnila Alidina

Workshop Committee:
Chair: Dr Hafsa Majid
Members:
Dr Shabnam Khowaja
Dr Waqas Uddin Khan
Dr Zahra Hoodbhoy
Dr Siraj Muneer
Ms Sarah Baber

Publication Committee
Chair: Dr Sibtain Ahmed
Members:
Ms Dhunmai Cowasjee
Ms Fabeha Pervez
Dr Howrah Ali
Dr Bilal Hashmi
Ms Mahek Anwar Ali

Congress Management Committee
Chair: Prof Imran Siddiqui
Co-Chair: Prof Adnan Mustafa Zuberi & Ms Shamsha Punjwani
Members:
Dr Bilal Hashmi
Dr Siraj Muneer
Mr Nasheed Irshad
Mr Karim Tejani
Ms Gulnar Zafar Ali
Mr Akber Madhwani
Ms Zohra Hajiani
Mr Asad Yaqoob
Mr Khawaja Hateem

Pre-Symposium Meetings
(Closed session by invitation only)
Wednesday, March 4, 2020
Venue: Hospital Office Building

8:30-9:30 am	**Aga Khan University Hospital Newborn Screening Advocacy Committee Meeting**
9:30-9:45 am	**Tea Break**
9:45-10:30 am	**Pak-IMD-Net Satellite Meeting**

****

Pre-Symposium Workshops
Thursday, March 5, 2020
Venue: Anatomy & Surgery Learning Studio & JHS Conference Room

9:00 am-12:00 pm	**Establishing Registries for Tracking Patient Outcomes** **Facilitators:** *Drs Zahra Hoodbhoy, Hafsa Majid, Adnan Jabbar, Mustafa Aslam, M Nadeem Anjum, Lena Jafri and Aysha Habib Khan*
9:00 am-12:00 pm	**Genetic counselling: How to do it right?** **Facilitators:** *Ms Fizza Akber, Drs Salman Kirmani, Siraj Muneer and Amy Calhoun*
2:00-5:00 pm	**Bioinformatics-Hands-on on IMD Genomics** **Facilitators:** *Dr Waqasuddin, Prof Shahid Mahmood Baig, Dr Sibtain Ahmed, Ms Kehkashan Imam and Ms Misha Ahmed*

****

Pre-Symposium Meetings
(Closed session by invitation only)
Friday, March 6, 2020
Venue: Anatomy & Surgery Learning Studio

9:00 am-12:30 pm	**Workshop on Essentials of Establishing a Newborn Screening Program** **Facilitators:** *Drs Hafsa Majid, Bushra Afroze, Dianne Webster, Aysha Habib Khan and Lena Jafri*

Pre-Conference Workshop on 'Establishing Registries for Tracking Patient Outcomes'

Thursday, March 5, 2020 | 9:00 am-12:00 pm | Anatomy & Surgery Learning Studio

Facilitators: Drs Zahra Hoodbhoy, Hafsa Majid, Adnan Jabbar, Mustafa Aslam, M Nadeem Anjum, Lena Jafri and Aysha Habib Khan

About the workshop: A patient registry is defined as "an organized system that uses an observational study method to collect uniform data to evaluate specified outcomes for a population defined by a particular disease, condition, or exposure, and that serves one or more predetermined scientific, clinical , or policy purposes". This half -day workshop will provide an insight in this fast-evolving field and will provide motivation for developing a clinical registry in their area of interest.
Target Audience: 20 (minimum 12). This half-day workshop will be specifically designed for clinicians, pathologist, post-doctoral fellows, and M. Phil. / Ph. D. students who plan to develop registry in their area of interest.

Programme

9:00-9:05 am	**Tilawat** **Welcome and icebreaking**
9:05-9:35 am	**Developing a Registry Identifying your registry's aim and objectives, Defining your registry dataset, Establishing registry data collection, Sustaining registry data collection, Sustaining a registry, Quality assurance, Resourcing & funding** Dr Zahra Hoodbhoy, Assistant Professor, Research Department of Paediatrics and Child Health, Aga Khan University
9:35-9:50 am	**Ethical Considerations while Developing a registry Governance, policies, ethics of registry data protection, sharing and transfer** Dr Mustafa Aslam, Senior Instructor Department of Pathology and Laboratory Medicine, Aga Khan University
9:50-10:20 am	**Working Examples of Registry Cancer Registry at AKUH: Software selection** Dr Adnan Jabbar, Associate Professor and Section Head Oncology Aga Khan University, Karachi, Pakistan **A Lysosomal Storage Disease Registry** Dr M Nadeem Anjum Assistant Professor The Children's Hospital, Lahore, Pakistan
10:20-10:30 am	**Bringing synergies and breaking silos: Way forward for developing laboratory consortia in IMD's** Dr Aysha Habib Khan, Professor, Department of Pathology and Laboratory Medicine Aga Khan University, Karachi, Pakistan
10:30-10:40 am	**Tea Break**
10:40-11:50 am	**Group Activity:** Dr Zahra Hoodbhoy, Lena Jafri & Hafsa Majid
11:50 am-12:00 pm	**Closing**

Pre-Conference Workshop on 'Genetic Counseling: How to do it right?'

Thursday, March 5, 2020 | 9:00 am – 12:00 pm | JHS Conference room

Facilitators: Ms Fizza Akber, Drs Salman Kirmani, Siraj Muneer and Amy Calhoun

About the workshop: The half-day workshop will provide an insight in this fast-evolving field of genetic counseling. Facilitators will talk about the communication process of genetic counseling. How to effectively deliver knowledge about the genetic aspects of illnesses with those who are at an increased risk or either having a heritable disorder or of passing it on to their unborn offspring. Participants will gain understanding of the inheritance pattern of illnesses and their recurrence risks; learn how to address the concerns of patients, their families, and their health care providers; and how best to supports patients and their families dealing with these illnesses.

Target Audience: This half-day workshop will be specifically designed for medical practitioners, clinicians, pathologist, post-doctoral fellows, M. Phil. / Ph. D. students, and corporate professionals working in the areas of genomics.

Requirements: All participants should bring their charged laptops (chargers, connectors, extensions)

Programme

9:00-9:15 am	**Tilawat** **Welcome & Ice Breaking Session**
9:15-9:30 am	**What is Genetic Counseling?** **Introduction to the evolving need for healthcare professionals trained in clinical genetics and genetic counseling in mainstream clinical practice.** Amy R.U.L. Calhoun, MD Clinical Associate Professor &Medical Director Iowa Newborn Screening Program, Division of Medical Genetics and Genomics, Stead Family Department of Pediatrics University of Iowa Health Care, Iowa City
9:30-9:45 am	**Inheritance patterns and Pedigrees** **Understanding and identifying inheritance patterns of genetic disorders and the impact on disease recurrence risk in families. Introduction and interpretation of familial disease history with the use of pedigrees** Dr Salman Kirmani, Associate Professor and Chair Department of Pediatrics and Child Health, Aga Khan University
9:45-10:30am	**Case studies on Making a Pedigree** Facilitators: Ms Fizza Akbar, Drs Siraj Muneer, Salman Kirmani
10:30-10:45 am	**Tea Break**
10:45-11:10 am	**Inside a Genetics Consultation** **Understanding the implications of a genetic test result and a genetic condition diagnosis for the patient and their families. Focusing on effective way to communicating the facts and showing empathy** Ms Fizza Akbar, Genetic Counselor Department of Pediatrics and Child Health, Aga Khan University, Karachi
11:10-11:50 am	**Role plays by participants followed by feedback by the facilitators** Facilitators: Ms Fizza Akbar, Drs Siraj Muneer, Salman Kirmani
11:50-12:00 pm	**Closing**

Pre-symposium Workshops on 'Bioinformatics hands-on on IMD genomics'

Thursday, March 5, 2020 | 2:00-5:00 pm | Anatomy & Surgery Learning Studio

Facilitators: Dr Waqasuddin, Prof Shahid Mahmood Baig, Dr Sibtain Ahmed, Ms Kehkashan Imam and Ms Misha Ahmed

About the workshop: High throughput, massively paralleled Next Generation DNA Sequencing (NGS) technology has revolutionized the field of genomics. Using NGS, an entire human genome can be sequenced within a few hours. This technology has tremendous applications in biomedicine, agriculture, and biotechnology. Correspondingly, the combination of NGS and Bioinformatics is indispensable to the interpretation and application of this biological data, specifically for clinical diagnostics and medical treatment in the area of inherited metabolic defects. This half-day workshop will provide an insight in this fast-evolving field and will provide an impetus for developing an understanding of NGS data interpretation related to biochemical genetics.

Target Audience: 20 (minimum 12). This half-day workshop will be specifically designed for medical practitioners, clinicians, pathologist, post-doctoral fellows, M. Phil. / Ph. D. students, and corporate professionals working in the areas of genomics and bioinformatics

Requirements: All participants should bring their charged laptops (chargers, connectors, extensions)

Programme

2:00 pm	**Tilawat**
2:05-2:15 pm	**Welcome & Ice Breaking Session** Dr Sibtain Ahmed
2:15-2:30 pm	**Introduction to the Next Generation Sequencing (NGS) for Inherited Metabolic Disorders (IMDs)** Prof Shahid Mahmood Baig
2:30 – 3:00 pm	**NGS Technology and Applications with Perspectives on IMDs** Dr Waqasuddin Khan
3:00-3:15 pm	**Tea Break**
3:15-3:45 pm	**How to interpret the VCF file – Interactive Case Study (Hands-on** Drs Waqasuddin Khan, Sibtain Ahmed, Ms Kehkashan Imam and Ms Misha Ahmed
3:45-4:45 pm	**Evaluation of the Genotype-Phenotype Relationship of Causal Variant (Hands-on)** Dr Waqasuddin Khan, Ms Kehkashan Imam and Ms Misha Ahmed
4:45 pm	**Closing**

Pre-Conference Workshop 'Essentials of establishing a Newborn Screening Programme'

Friday, March 6, 2020 | 9:00am – 12:30pm | Anatomy & Surgery Learning Studio

Facilitators: Drs Hafsa Majid, Bushra Afroze, Dianne Webster, Aysha Habib Khan and Lena Jafri

Introduction: This closed group symposia will connect the Pathologist, pediatricians and obstetricians from different institutes to understand the essential requirements of establishing Newborn screening program. The symposia will feature presentations on the technical and practical elements of setting up a laboratory to perform newborn screening; talks on experiences from different centers and challenges of implementing newborn screening programs.

Target Audience: Participation will be by invite only. This half-day workshop is specifically designed for healthcare personnel who want to establish newborn screening services at their institutes.

Programme

9:00-9:10 am	**Tilawat** **Welcome and icebreaking** Dr Aysha Habib Khan, Professor Department of Pathology and Laboratory Medicine Aga Khan University
9:10- 9:20 am	**Kahoot Quiz**
9:20-9:40 am	**Newborn Screening: It's a system and not a test.** **Wilson Jungner criteria, education to outcome assessments, components of a NBS program, sustainability of program** Dr Bushra Afroze**,** Associate Professor, Department of Pediatrics and Child Health, Aga Khan University
9:40-10:10 am	**Practical considerations for setting up a laboratory for newborn screening** **Applying Wilson Jungner criteria, selecting disorder, screening, confirmatory analytes and cutoffs, establishing follow-up services** Dianne Rosemary Webster, Director Newborn Metabolic Screening Programme LabPlus, Auckland City Hospital, New Zealand
10:10-10:30 am	**Laboratory requirements to establish complete Newborn screening** **Lab infrastructure requirement and instrumentation requirements, sample collection requirements, Quality control issues and reporting requirements, External Proficiency, Method Validation requirements** Dr Hafsa Majid, Senior Instructor, Department of Pathology and Laboratory Medicine, Aga Khan University
10:30-10:45 am	**Tea Break**
10:45 am-12:00 pm	**Group Activity:** Drs Dianne Webster, Aysha Habib Khan, Hafsa Majid and Bushra Afroze
12:00-12:10 pm	**Kahoot Quiz**
12:10 – 12:15 pm	**Closing**

Inaugural Session
Friday, March 6, 2020
Venue: AKU Auditorium

2:00-2:30 pm	**Registration**
2:30-2:35 pm	**Recitation of the Holy Quran**
2:35-2:40 pm	**National Anthem**
2:40-2:45 pm	**Introduction and Overview of Symposium** Dr Farooq Ghani Service Line Chief & Associate Professor, Department of Pathology & Laboratory Medicine, Aga Khan University
2:45-2:50 pm	**Welcome Address** Dr Adil H. Haider Dean, Medical College, Aga Khan University
2:50-4:00 pm	**Keynote Speeches** **Co-chairs: Prof Muhammad Khurshid and Prof Abdul Gaffar Billoo**
	State of the Genetic Diseases in Pakistan and Prevention Strategies Prof Shahid Mahmood Baig (PhD, Sitara i Imtiaz) Head of Human Molecular Genetics, Health Biotechnology Division, Deputy Chief Scientist, National Institute for Biotechnology and Genetic Engineering, Faisalabad
	Ethics of Newborn Screening: When it is Right 'NOT' to Know Dr Amy R.U.L. Calhoun, MD Clinical Associate Professor and Medical Director of Iowa Newborn Screening Program, Division of Medical Genetics and Genomics, Stead Family Department of Pediatrics, University of Iowa Health Care, USA
	Pecha Kucha: Reframing Rare in Pakistan Prof Aysha Habib Khan Department of Pathology & Laboratory Medicine, Aga Khan University
4:00-4:15 pm	**Deafening Silence -** MIME Show by Medical College Students
4:15 – 4:20 pm	**Address by Chief Guest** Prof Abdul Gaffar Billoo
4:20 – 4:25 pm	**Address by Patron** Prof Mohammad Khurshid
4:25-4:30 pm	**Remarks and Vote of Thanks** Prof Imran Siddiqui, Department of Pathology & Laboratory Medicine, Aga Khan University
4:30-5:00 pm	**'A Walk for Rare' Followed by Hi-Tea**

Symposiums Program
Saturday, March 7, 2020
Venue: AKU Auditorium

8:30-9:00 am	**Registration**
9:00-9:10 am	**Glimpses from the Past Conference on Rare Diseases, 2019**

Symposium I

9:00-10:15 am	**Outcome & Challenges in Management of Inherited Metabolic Disorders (IMDs)** **Chair:** Dr DS Akram **Co-chairs:** Dr Lumaan Sheikh **Moderator:** Dr Sidra Arshad **Metabolic Disorder Screening by Mass Spectrometry – What Are We Learning? – Video presentation** Prof Giancarlo La Marca Clinical Biochemistry and Clinical Molecular Biology, University of Florence, Italy, President Italian Society for Newborn Screening and Metabolic Diseases **Antenatal Presentation of Rare Diseases** Prof Shama Munim Department of Obstetrics and Gynaecology, Aga Khan University **Challenges and Outcome of Management of IMDs at Children Hospital, Lahore** Prof Huma Cheema, Department of Pediatric Gastroenterology-Hepatology & Nutrition, The Children's Hospital & Institute of Child Health, Lahore **Ten Years Outcome in Management of IMDs in Pakistan – *Challenges and Outcome*** Dr Bushra Afroze, Associate Professor & Consultant Metabolic Geneticist, Department of Pediatrics and Child Health, Aga Khan University **Q & A Session**
10:15-10:45 am	**Tea & Poster walk**

Symposium II

10:45-11:45 am	**Newborn Screening** **Chair:** Prof Huma Cheema **Co-chair:** Brig (R) Prof. Aamir Ijaz **Moderator:** Dr Ghulam Zainab **Newborn Screening: When to Decide to Discontinue Screening** Ms Dianne Webster, Director Newborn Metabolic Screening Program, Lead Clinical Scientist Antenatal Screening for Down syndrome and Other Conditions, Auckland District Health Board, New Zealand

Outcome of Newborn Screening in Punjab & Challenges Encountered
Dr Farkhunda Gafoor, Shalimar Medical & Dental College, Lahore

Outcome of a Short Term and Long-term Follow-up of Newborn Screening at Aga Khan University Hospital
Dr Hafsa Majid, Senior Instructor, Department of Pathology & Laboratory Medicine,
Aga Khan University
Dr Khadija Nuzhat Humayun, Associate Professor, Department of Paediatrics and Child Health, Aga Khan University

Experience sharing on NBS by "The ZB Foundation"
Dr Ayesha Raza, Program Manager, "The ZB Foundation"

Symposium III

11:45am-12.30 pm **Complex Clinical Cases: Challenges Faced by Experts Dealing with Rare Diseases**
Chair: Prof Shahid Pervez
Co-chair: Prof Shahid Baig
Moderator: Dr Rehana Rehman

Establishing a Genetics Service at Aga Khan University Hospital: Challenges and Way Forward
Dr Salman Kirmani
Associate Professor & Chair, Department of Paediatrics and Child Health, Aga Khan University

Rare Presentations of Osteoporosis
Prof Aysha Habib Khan
Department of Pathology & Laboratory Medicine, Aga Khan University
& Dr Salman Kirmani
Associate Professor & Chair, Department of Paediatrics and Child Health, Aga Khan University

Rare Diseases Presenting in Dermatology
Dr Sadia Masood
Assistant Professor, Section of Internal Medicine, Department of Medicine, Aga Khan University

Dr Saira Fatima
Associate Professor, Department of Pathology & Laboratory Medicine, Aga Khan University

Rare Disease Diagnosis in Radiology
Dr Muhammad Awais
Senior Instructor, Department of Radiology, Aga Khan University

12:30-1:30 pm **Lunch, Prayer & Networking**

Symposium IV

1:30-2:30 pm

Rare Hematological Disorders: Not so Rare!
Chair: Prof Parvez Ahmed
Co-chair: Prof Salman N Adil
Moderator: Dr Muhammad Shariq

Genetic Disorders of Hemoglobin; Not so Rare
Maj Gen (R) Dr Suhaib Ahmed
Riphah International University, Islamabad

Inherited Bleeding Disorders: Way Forward
Dr Anila Rashid
Assistant Professor, Department of Pathology & Laboratory Medicine,
Aga Khan University

Inherited Thrombophilia Testing: Choosing Right Test for Right Patient
Prof Bushra Moiz, Department of Pathology & Laboratory Medicine,
Aga Khan University

Q & A Session

Symposium V

2:30-3:15 pm

Laboratories, Quality and Ethics for Rare Diseases
Chair: Dr Farooq Ghani
Co-chair: Maj Gen (R) Dr Suhaib Ahmed
Moderator: Dr Fatima Khanani

Laboratories, Quality and Ethics for Rare Diseases: Where Are We in Pakistan?
Brig (R) Prof. Aamir Ijaz
Consultant Chemical Pathologist, Bahria International Hospital, Rawalpindi

Rare Cases Requiring Interpretation of Molecular Tests: Role of Clinicians
Dr Saima Siddiqui
Assistant Professor, National Institute of Blood Disorders (NIBD), Karachi

Good laboratory Practices for a Biochemical Genetics Laboratory
Dr Sibtain Ahmed
Senior Instructor, Department of Pathology & Laboratory Medicine,
Aga Khan University

Pecha Kucha: Reframing Rare in Pakistan: Pak IMD-Net
Dr Lena Jafri
Assistant Professor, Department of Pathology & Laboratory Medicine,
Aga Khan University

3:15-3:40 pm

Table Discussion: Policy Recommendations in Light of Today's Seminar by Rare Experts

Moderator:
Dr Salman Kirmani & Prof Aysha Habib Khan

3:40-4:00 pm

Closing Ceremony and Awards Distribution
Prof Afia Zafar
Department of Pathology & Laboratory Medicine, Aga Khan University

www.aku.edu/RareDiseaseConf

#AKURareDiseaseConf