

PAK-IMD-NET NAMA 2022-2023



The official newsletter of the Pakistan Inherited Metabolic Disease Network (Pak-IMD-Net)
A Working Group of Pakistan Society of Chemical Pathology (PSCP)
Strengthening diagnostics, education and research in the area of inherited metabolic disorders



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Message from the Chair

Dr. Lena Jafri

**Associate Professor, Pathology and Laboratory Medicine
Aga Khan University Hospital (AKUH), Karachi**



As the Chair and an expert in diagnostics of our working group on Inherited Metabolic Disorders (IMDs) in Pakistan, I am pleased to update you on our recent progress.

Our group has been working tirelessly to improve the diagnosis and management of IMDs in Pakistan, where these disorders remain underdiagnosed and undertreated. We recognize that timely and accurate diagnosis is critical for improving the outcomes of affected individuals, and our efforts have been focused on enhancing diagnostic capacity across the country.

To this end, we have been conducting workshops and training programs for healthcare professionals, including geneticists, pediatricians, and laboratory technicians, to raise awareness about the different types of IMDs, their clinical presentation, and the most appropriate diagnostic tests.

Furthermore, we have been advocating for the inclusion of IMDs in newborn screening programs in Pakistan, as early detection can help prevent severe symptoms and complications.

We have also been working on developing standardized protocols for the diagnosis of IMDs, including the appropriate selection of diagnostic tests and the interpretation of results, to ensure consistency and accuracy in the diagnosis of these complex disorders.

However, we still have a long way to go, and I would like to urge all members of our working group to continue their efforts towards achieving our goals. We must work together to promote the early diagnosis and appropriate management of IMDs in Pakistan, improve access to specialized care, and provide support to affected individuals and their families. Together, we can make a significant difference in the lives of those affected by IMDs in Pakistan.

Hopes for the future?

The PSCP working group is collaborating with other organizations and institutions to build capacity in the diagnosis and management of IMDs. They are also conducting research studies to better understand the epidemiology, genetics, and clinical features of these disorders in the Pakistani population.


With these efforts, we expect that the diagnosis and management of IMDs will improve in Pakistan, leading to better health outcomes for affected individuals and their families. Furthermore, new research in this area may also contribute to the development of novel therapies and interventions for IMDs worldwide.

Overall, the future of inherited metabolic disorder research in Pakistan looks promising, and with continued efforts and collaboration, we can hope to make significant progress in this field.

"Together, we can make a significant difference in the lives of those affected by IMDs in Pakistan."

Rare is Many - Rare is Strong - Rare is Proud- Rare Disease Day 2022

Rare Disease Day is an annual event held on the last day of February each year to raise awareness about rare diseases and the challenges faced by patients and their families. In 2022, Rare Disease Day was celebrated by Pak-IMD-Net by conducting a special webinar on February 28th.



آغا خان یونیورسٹی
THE AGA KHAN UNIVERSITY

Organised by Department of Pathology & Lab Medicine in collaboration with Department of Continuing Professional Education and Pakistan Inherited Metabolic Disorders Network (Pak-IMD-Net) A Working Group of Pakistan Society of Chemical Pathology (PSCP)

Registration and Information Nasheed Irshad
021 34861927
nasheed.irshad@aku.edu

Hybrid Webinar Series
Rare Links-in line with Rare Disease Day

illuminating Rare Disorders' Diagnostics through Newborn Screening in Pakistan

Monday, February 28, 2022 | 10:00 am to 12:00 pm | Anatomy & Surgery Learning Studio

The Rare Links webinar series aims to provide a platform where national and international experts can share knowledge and experience of newborn screening and diagnostics of inherited metabolic disorders in Pakistan.

Learning Objectives

By the end of the session participants should be able to understand

- Understand the clinical utility of newborn screening testing of rare inherited metabolic disorders
- Describe the tests for follow up testing of these rare disorders
- Interpret newborn screening tests and second tier tests through case discussions

Who can attend?

Pathologists, biochemical genetics technologists and scientists, clinicians, chemical pathology residents and trainees

Click [here](#) to register

Activity Director

Dr Lena Jafri

International Speakers

Dr Samir Munir
Paediatric Inherited Metabolic Medicine
Great Ormond Street Hospital for Children
London United Kingdom

Dr George Sayhoun
Director and Chief Scientific
Officer of Med Labs, Jordan

Local Speakers


Dr Aysha Habib Khan
Professor, Chemical Pathology
Aga Khan University

Dr Aamir Ijaz
Professor, Chemical Pathology
Mohi-ud-Din Islamic Medical College
Mirpur, Co-Chair Pak-IMD-Net

Dr Lena Jafri
Associate Professor, Chemical Pathology
& Chair Pak-IMD-Net
Aga Khan University

Dr Hafsa Majid
Assistant Professor, Chemical Pathology
Aga Khan University

Dr Sibtain Ahmed
Assistant Professor, Chemical Pathology
Aga Khan University



Activity Code: DNC-0011-2022
"Rare Links On Rare Disease Day"

The ACCME designates this live educational activity for a maximum of 2.00 credit hours of AMA PRA category 1 credit™. The faculty, staff and any individual connected to this activity certifies that this activity does not have any financial relationship with proprietary activities, products, marketing, or selling or distributing healthcare goods or services consulted or used at patients.

Rare Disease Day 2023

On Rare Disease Day 2023, an expanded newborn screening program was launched at AKUH, Karachi. The program will use advanced screening technologies to detect a wider range of rare diseases in newborns and work closely with healthcare providers and other stakeholders to ensure that affected infants receive the best possible care.

The program, led by the Department of Pathology and Laboratory Medicine and the Department of Paediatrics and Child Health, focuses on implementing a Newborn Screening (NBS) system.

Dr. Lena and Bushra Afroze presented the system, emphasizing that it is not a test but a comprehensive approach to identifying health issues in newborns. The program initially started with the main CPUs and expanded to include secondary hospitals. In 2022, the NBS system was implemented at Cap CEC and ERNDIM in Hyderabad, and in January 2021, it was implemented at the CAH main campus. Dr. Hafsa developed a virtual learning environment training program for the DBS collection system. Azeema and Farhat were recognized for their contributions to LCMS optimization during the COVID-19 pandemic.

Dr. Khadija Humayun discussed the time-consuming process of clinical follow-up and confirmatory testing. Despite the challenges, the effort was deemed worthwhile as it resulted in 27 children being saved from developmental delays. The families of these children also had an opportunity to discuss their well-being. Dr. Khadija also highlighted the issue of retrieval failure during this period and emphasized the importance of universal screening rather than just high-risk screening.

Dr. Sibtain shared the team's initial decision to use Indian cutoffs for the NBS program, which led to a high false positive rate. However, they later adopted the Oregon State Health Program, resulting in a significant decrease in false positives. The prevalence of the disorder in Oregon was 1 in 150, while in Pakistan, it was 1 in 8500. The team selected 12 disorders relevant to the Pakistani context, and the clinical team has been supportive throughout the program. However, the classical PKU algorithm was not included in the program due to its complexity in reaching a diagnosis.



Rare Disease Day 2023

Dean Adil Haider emphasized the importance of the NBS program for affected families. He stated that the care young children receive from everyone around them shapes them into great citizens who contribute to society.

Following the Dean's remarks, CEO Dr. Farhat Abbas shared his thoughts on the program. He commended the Tertiary Care Center for its commitment and presented a case study on AKU. Dr. Abbas praised the full-circle commitment of the staff, highlighting that the program is easy, inexpensive, and quick. He noted that society is split into those who cause destruction and those who care about the birth of every small child, as every newborn is the future. He encouraged everyone to do whatever they can to protect the future and acknowledged that these things take time to achieve. Dr. Abbas ended his remarks by urging everyone to feel proud of their contributions to the NBS program.

Dr. Aysha and Dr. Salman then emphasized that the success of the ABS program would depend on togetherness and the engagement of all stakeholders from the beginning. They stressed the importance of noncompetitive collaborations, which are rare in the South Asian region, especially in an expensive institution. However, they highlighted that the AKU has both academic and clinical credibility, which can support such collaborations.



ChemCon 2022

The Pakistan Society of Chemical Pathology (PSCP)-Karachi Chapter organized its 12th Annual Conference ChemCon 2022 with the theme of "Metabolomics: Revolutionizing Chemical Pathology" in Karachi, Pakistan, on November 25-26, 2022. The event was held under the auspices of the International Federation of Clinical Chemistry and Laboratory Medicine (IFCC).

The conference aimed to promote open science, particularly in metabolomics, and provided an opportunity to discuss the latest advancements, achievements, and concerns related to metabolomics in chemical pathology in Pakistan. The conference included a pre-conference workshop and two scientific sessions, which featured keynote talks by national and international experts, including Dr. Khosrow Adeli, President IFCC.

An innovative feature of the conference was "Reels on Metabolomics," where attendees submitted short videos highlighting their work and experiences related to metabolomics.

Held in Pearl Continental Hotel, Karachi, ChemCon 2022 brought together chemical pathology experts from various institutions in Pakistan to discuss the latest research and advancements in the field. The conference provided a platform for professionals to share their latest knowledge and network with peers. It facilitated networking opportunities, allowing attendees to connect with colleagues and potential collaborators in the field. These connections helped attendees build professional relationships and could lead to promising future collaborations, especially in metabolomics.

The competitive oral and poster presentations displayed novel scientific research and presented a great chance for learning. Overall, Chemcon 2022 was a beneficial event for chemical pathologists in Pakistan as it provided a platform for knowledge sharing, learning, and networking.



ChemCon 2022

Reels on Metabolomics from Pakistan

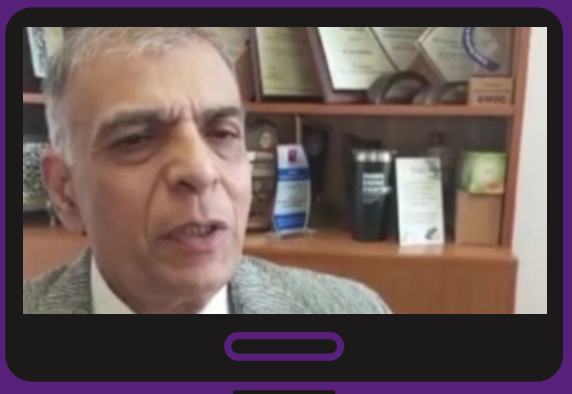
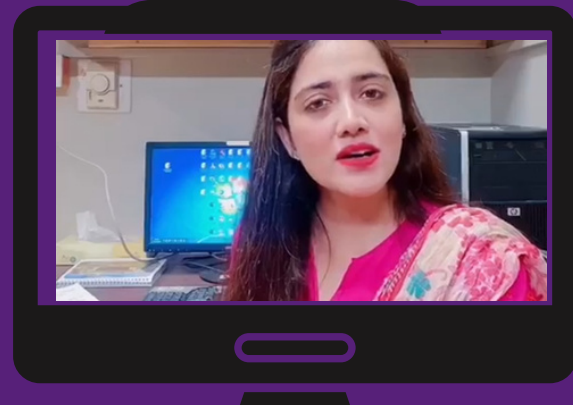
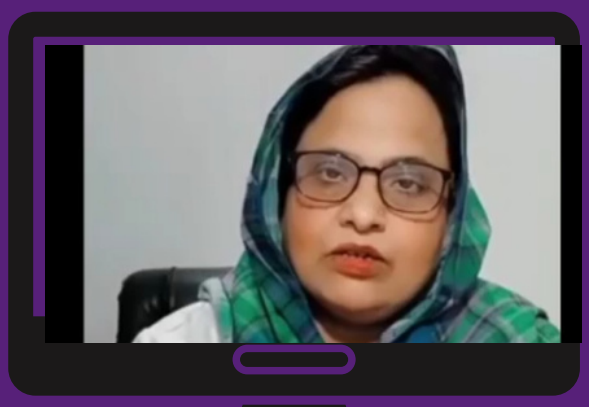
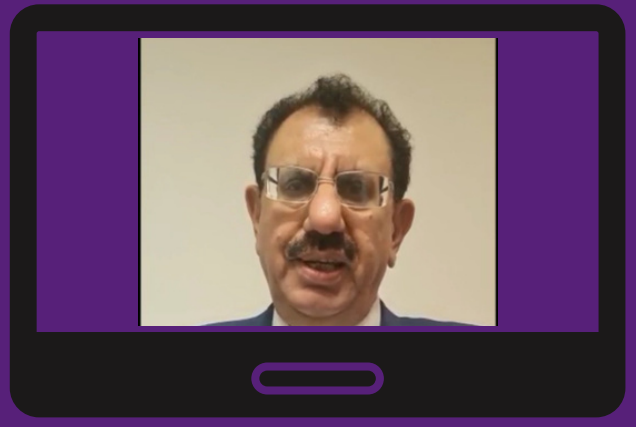
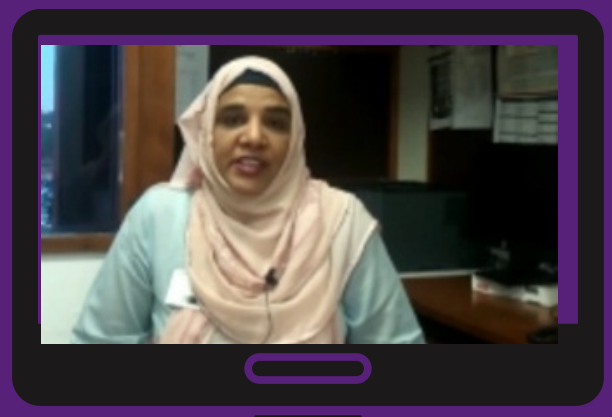
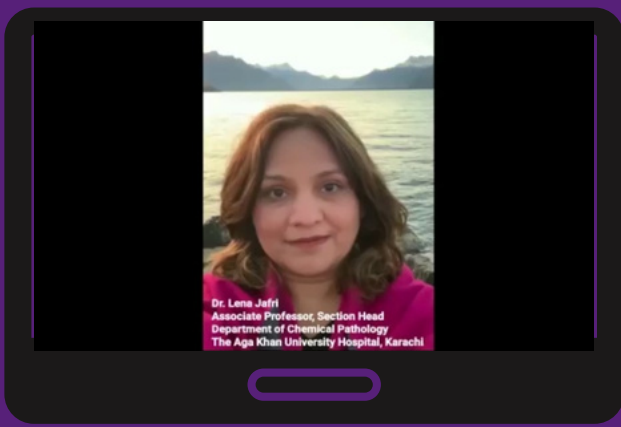
Dr Lena Jafri, Dr Hafsa Majid, and Dr Imran Siddiqui

The Metabolomics Reels project from Pakistan aimed to create buzz and raise awareness about metabolomics in the chemical pathology community and beyond. Short videos, or reels, were created and shared on social media platforms, showcasing experiences, achievements, and future related to metabolomics, while promoting its integration into chemical pathology laboratories and its impact on personalized medicine in Pakistan. This project was part of ChemCon 2022. The display of these videos on LCD screens during the conference and their subsequent sharing on the social media platforms of the Pakistan Society of Chemical Pathologists (PSCP) significantly increased their reach and visibility.

The purpose of these reels was threefold. Firstly, they aimed to raise awareness and generate interest in the field of metabolomics, fostering its recognition and importance among chemical pathology professionals in Pakistan. Secondly, the videos provided a platform for Pakistani labs to share their unique perspectives on metabolomics, showcasing their current advancements and contributions to the field. Lastly, the reels facilitated discussions on actionable plans for integrating metabolomics into the chemical pathology laboratories of Pakistan, thereby fostering collaboration, and driving future progress.

The guidelines provided for creating these reels ensured their effectiveness. Keeping the videos concise, with a maximum length of 30 seconds and consisting of 2-3 sentences, allowed for impactful and easily digestible content. The contents of the videos were diverse and aimed to engage the viewers with various aspects of metabolomics. Ideas for the content included sharing experiences and achievements related to metabolomics in chemical pathology, discussing how metabolomics can benefit clinicians in Pakistan, exploring the scope of metabolomics in personalized and precision medicine, highlighting training opportunities in metabolomics within Pakistan, discussing the integration of metabolomics into the Chemical Pathology curriculum, and emphasizing the value that metabolomics has added to the healthcare landscape of Pakistan.

Overall, the outcomes of this project were highly successful in promoting and highlighting the significance of metabolomics in chemical pathology. The reels effectively generated interest, fostered discussions, and laid the foundation for future advancements and collaborations in the field within Pakistan.



Contributors of Reels

Picture Gallery



Members of PAK-IMD-Net

Chair

Dr. Lena Jafri, Associate Professor, Department of Pathology and Laboratory Medicine, Aga Khan University Hospital (AKUH), Karachi

Co-Chair

Professor (Brig-Retd) Aamir Ijaz, Professor of Chemical Pathology, Mohiuddin Islamic Medical College, Mirpur, AJK

Members

Dr. Aysha Habib Khan, Professor, Department of Pathology and Laboratory Medicine, AKUH, Karachi

Dr. Hafsa Majid, Assistant Professor, Department of Pathology and Laboratory Medicine, AKUH, Karachi

Dr. Sibtain Ahmed, Assistant Professor, Department of Pathology and Laboratory Medicine, AKUH, Karachi

Dr. Khadija Humayun, Associate Professor, Department of Paediatrics and Child Health, AKUH, Karachi

Dr. Madiha Beg, Assistant Professor, Department of Medicine, AKUH, Karachi

Dr. Siraj Muneer, Staff Pathologist, AKUH, Karachi

Dr. Zaibunissa Khan, Resident Chemical Pathology, AKUH, Karachi

Azeema Jamil, Senior Technologist, BioGenetics Laboratory, AKUH, Karachi

Nasir Ali Khan, Senior Technologist, BioGenetics Laboratory, AKUH, Karachi

Iffat Arman, Senior Technologist, BioGenetics Laboratory, AKUH, Karachi

Hira Naeem, Technologist, BioGenetics Laboratory, AKUH, Karachi

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Dr. Bilal Hashmi, Staff Pathologist, AKUH, Karachi

Dr. Noreen Sherazi, Assistant Professor, National Institute of Cardiovascular Diseases, Karachi

Dr. Fatima Kanani, Section Head of Chemical Pathology, The Indus Hospital, Karachi

Dr. Shabnam Dildar, Consultant Chemical Pathology, National Institute of Blood Diseases, Karachi

Dr. Farheen Aslam

Dr. Usman Ali, Assistant Professor, Chemical Pathology, Ziauddin Hospital, Karachi

Dr. Erum Salim, Assistant Professor, Chemical Pathology, Ziauddin Hospital, Karachi

Dr. Howrah Ali, Assistant Professor, Chemical Pathology, Liaquat National Hospital, Karachi

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Lt Col Dr. Ayesha Hafeez, Chemical Pathologist, Armed Forces Institute of Pathology, Rawalpindi

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Maj Saima Bashir, Department of Chemical Pathology, Combined Military Hospital, Quetta

Col Zujaja Haroon, Department of Chemical Pathology, Combined Military Hospital, Multan

Corporate Member: Mr. Muhammad Abdullah, Nutricia

Announcement from PSCP Executive Council

This is to announce that Dr Hafsa Majid is nominated as Chair and Dr Usman Munir as Co-Chair of Pak-IMD-Net for next 3 years effective from June 19th 2023. The council would like to thank Dr Lena Jafri and Dr Aamir Ijaz (Founding members/Chair and Cochair respectively) for their contributions towards the working group

