



PAK-IMD-NAMA 2020-2021



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

INSIDE:

Messages from the Chair and Co-Chair

RARE-Link Webinar Series

**Highlights from Rare Disease Day 2020 and 2021 Conferences
and Workshops**

Research Projects

Members' Details

FROM THE CHAIR

Dr. Lena Jafri

**Associate Professor, Pathology and Laboratory Medicine
Aga Khan University, Karachi**



The first year of Pak-IMD-Net has been wonderfully fulfilling. We charged into the year 2020 with two goals. Firstly, to hold educational workshops, training courses and meetings to enhance the knowledge of HCPs regarding screening, diagnosis and prevention of Inherited Metabolic Disorders (IMDs). Secondly, to promote research in the area of IMDs in Pakistan through establishing partnerships with funding bodies. Amid COVID-19 restrictions, we turned to the screen to host our popular 'Rare Links' webinar series completely online. Our national and international members collaborated readily to educate and learn about IMDs via Zoom, proving that when the going gets tough, we get tougher! Year 2021 was celebrated as the **year of metabolomics** by Pak-IMD-Net. We realize that the advances in the techniques of analytical chemistry and metabolomics are the source of the rapid evolution of a new omics era. The introduction of metabolomics in Pakistan for IMDs pertains to two important platforms: analytical techniques, (gas chromatography mass spectrometry and liquid chromatography tandem mass spectrometry) and multivariate data analysis software (Collaborative Laboratory Integrated Reports -in collaboration with Mayo Clinic, USA). Our educational meetings in 2021 revolved around these two omics domains. Pak-IMD-Net members and the chemical pathology fraternity will keep on learning and translating metabolomics related educational and research activities into clinical practice for the benefit of patients with IMDs in Pakistan.

During 2021, in several meetings we discoursed on the powerful technology of tandem mass spectrometry (LC-MS/MS) that can be used for screening of multiple IMDs using a dried blood spot.

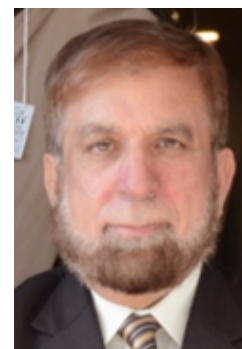
The 'Dried Blood Spot Metabolic Profile' when introduced on LC-MS/MS at AKU lead to an active discussion amongst Pak-IMD-Net members on need of clinical pathways and algorithms for clinicians. The new test 'Dried Blood Spot Metabolic Profile' will provide quantification of multiple amino acids, multiple acylcarnitines and succinylacetone, and is intended as a screening method for the early detection of disorders of amino acid, fatty acid metabolism and organic acidemias in newborns.

A central problem is the large number and variety of IMDs and the poor recognition of these disorders that make it challenging for the pediatric neurologist and/or pediatricians to decide on the initial evaluation with the consequent delay in diagnosis and timely treatment.

Therefore, it is necessary for the clinical labs to provide diagnostic algorithms to clinicians based on local burden of IMDs, practicalities and resources.

FROM THE CO-CHAIR

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



Pak-IMD-Net since its inception in 2019, is striving hard to create awareness and opportunities for the screening and diagnosis of IMDs in Pakistan. During the years 2020 and 2021 the world was fighting an intense battle against COVID-19 pandemic, which is still far from end. In spite of the pandemic the activities of our network continued, mostly virtual though.

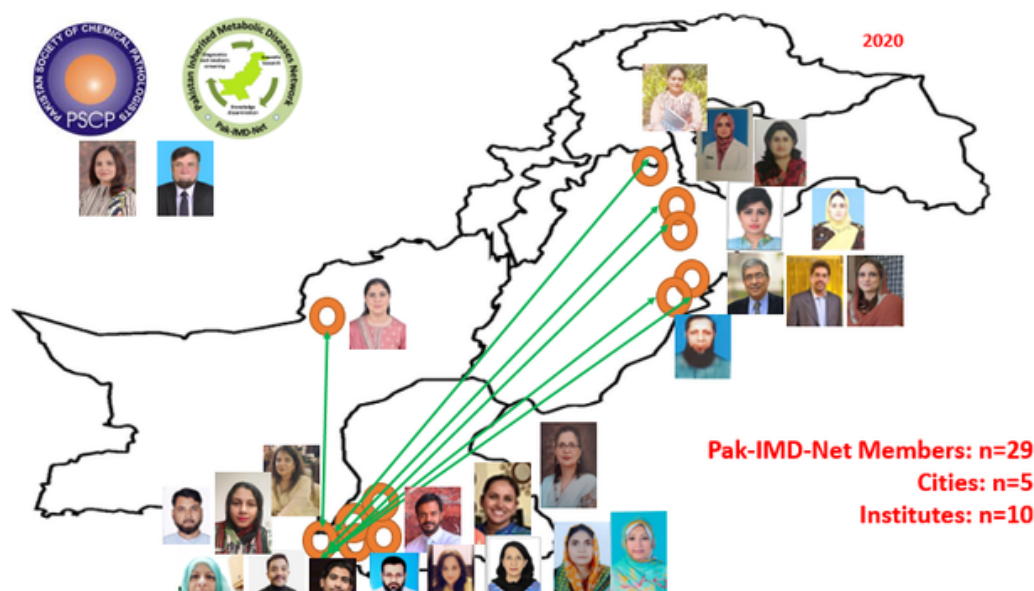
Use of tandem mass spectrometry (LC-MS/MS) as a powerful platform for Newborn Screening (NBS) is most outstanding. Parallel to the use of metabolomics for NBS, equipment for hormonal disorders like Congenital Hypothyroidism (CH) and Congenital Adrenal Hyperplasia as well as disorders like Cystic Fibrosis and sickle cell anaemia has also been installed at Armed Forces Institute of Pathology Rawalpindi.

Efforts of **ZB Foundation** in establishing NBS services in Pakistan cannot be ignored. This NGO is probably the only organization which is offering free of cost testing for CH. Shorter hospital stays of mothers after deliveries is one of the major hurdles in specimen collection of the newborn for NBS of CH at 3rd day of life. To overcome this problem a study has been started at a very busy hospital of Peshawar to establish TSH cut-off values for first 12 hours of life. This study is being done in collaboration with ZB Foundation.

To create awareness about IMDs and NBS a book-chapter has been added in an under publication Neonatology Textbook and 2nd edition of PSCP Handbook on IMDs (2016) is also underway.

Presently, a close collaboration amongst Pediatricians, Neonatologists, Chemical Pathologists and other stake holders is the need of the hour. A common platform like a web portal can provide an excellent method of communication. Similarly, societies of parents of affected children to discuss the problems of their suffering kids are also required and Pak-IMD-Net will facilitate these societies in near future inShAllah.

OUR NETWORK GREW



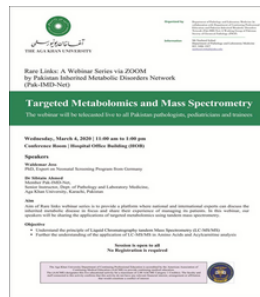
RARE-LINK WEBINAR SERIES

Amid physical COVID-19 restrictions, we moved our popular RARE-Link lecture series to the screen, allowing national and international pathology experts to discuss inherited metabolic diseases in focus and share their experiences in patient management freely via Zoom.

The webinars were attended by an array of pathologists, pediatricians, biochemical genetics technologists, and pathology residents across Pakistan.

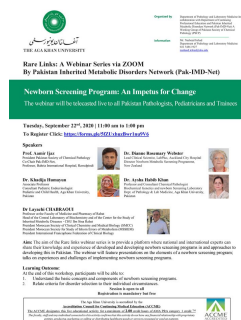
Targeted Metabolomics and Mass Spectrometry – Mar 4, 2020

This webinar saw the speakers, **Dr. Sibtain Ahmed**, and **Waldemar Jess, PhD** and expert on neonatal screening from Germany who discussed the applications of targeted metabolomics using tandem mass spectrometry. The speakers detailed the principle of **liquid chromatography mass spectrometry**, and its application in **amino acids** and **acylcarnitine analysis**. The webinar was telecasted live to all Pakistani pathologists, pediatricians, and technologists, and more than 50 participants attended.



Newborn Screening Program: An Impetus for Change' – Sep 2020

The webinar gathered a decorated panel of national and international experts on NBS, namely Dr. Dianne Webster, Prof. Layachi Chabraoui, Prof. Aamir Ijaz, Dr. Khadija Humayun, and Dr. Lena Jafri to discuss various aspects of the newborn screening program in Pakistan. The focus of the workshop was on understanding the basic



concepts and components of a complete newborn screening program, and relating criteria for disorder selection to their individual circumstances. The session was open for all Pakistan pathologists, pediatricians, neonatologists, public health experts and policy makers. Following an icebreaking session by the Chair, Co-chair Dr. Aamir Ijaz took over the proceedings and described the requirements to build a NBS laboratory. Dr. Aysha Habib Khan shared a Consensus Report from Conference on NBS in Pakistan and Dr. Hafsa shared success stories of the NBS program in AKUH, the panelists approved Policy Recommendations for starting a Nationwide NBS program. It was a remarkably successful event, with more than 70 participants from 3 different countries, 17 different cities, and 34 different institutes of Pakistan. The participants were from different areas of medicine including Pediatrics, Laboratory Medicine, Gynecology and Obstetrics, nursing, Family Medicine, Internal Medicine, and Public Health sectors.

Laboratory Diagnosis of IMDs – Feb 2, 2021

This CME activity is focused at diagnosis of inherited metabolic disorders. Amino Acidopathies and organic acidurias are rare inherited metabolic disorders (IMDs) due to absolute or



relative deficiency of specific enzyme or coenzyme.

With Dr. Lena Jafri and Dr. Sibtain Ahmed as speakers, this session emphasized upon the clinical utility of first line and specialized biochemical genetic tests available for the timely screening and diagnosis of IMDs, which in turn helps in saving critical time to initiate timely disease-specific treatment and effective genetic counselling.

Good Lab Practices for Establishing Newborn Screening Program' – June 15, 2021

The CME based webinar stressed on the clinical utility of newborn screening testing for IMDs. Guest speaker Dr. George Sayhoun, Director and Chief Scientific Officer of MedLabs discussed the challenges faced by clinical laboratories in newborn screening; while the AKUH faculty shared their experiences during initiation of the program at their clinical laboratory. Pathologists, biochemical genetics technologists and scientists, and chemical pathology residents learned how to describe the tests for follow-up testing of various IMDs and how to interpret newborn screening reports when presented to them. Case discussion on dried blood spot metabolomic data were done. This metabolomic data panel included DBS-acylcarnitines, DBS-amino acids and DBS-succinylacetone concentrations.

Newborn Screening Program Strategic Planning Retreat' – July 2021

The intense retreat saw Pak-IMD-Net committee from AKUH brainstorm key drivers and barriers to NBS in Pakistan and discuss concerns with SWOT analysis. Each member shared their visions for the future of NBS in the country and actively prioritized strategies for its implementation. After a brief tea break, the members resumed action planning for said strategies, and together with Participants from Lahore and Quetta joined the hybrid event via Zoom, they decided the resources required for expansion of NBS in the region.

Organized by Department of Pathology & Lab Medicine, collaboration with Pak-IMD-Net, a working group of Pakistan Society of Clinical Pathology
Registration and Information: National Medical Centre, AKUH, Department of Pathology & Lab Medicine, Contact: 011-346-1921, e-mail: pathlab@medakuh.edu.pk

Rare Inlets & Webinar Series By Pakistan Inherited Metabolic Disorders Network (Pak-IMD-Net)

Good Lab Practices for Establishing Newborn Screening Program

Tuesday, June 15, 2021 | Virtually Via Zoom | 11:00 am-1:00 pm

 Dr. Lena Jafri Assistant Director, MedLabs, National Medical Centre, Department of Pathology & Laboratory Medicine, Aga Khan University	 Dr. George Sayhoun Director and Chief Scientific Officer, MedLabs, National Medical Centre, Department of Pathology & Laboratory Medicine, Aga Khan University	 Dr. Sibtain Ahmed Assistant Professor, National Medical Centre, Department of Pathology & Laboratory Medicine, Aga Khan University
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Aim: Aim of Rare Inlets webinar series is to provide a platform where national and international experts can discuss about the inherited metabolic disease to focus and share their experiences.

Learning Objectives: At the end of this CME session, participants will be able to

- Understand the clinical utility of newborn screening testing of inherited metabolic disorders.
- Describe the tests for follow up testing of various inherited metabolic disorders.
- Interpret newborn screening reports for various inherited metabolic disorders.

Who should attend? Pathologists, Biochemical Genetics, Technologist & Scientists, Chemical Pathology Residents and Trainers

Register now!! Limited Space, No Registration Fee. However, registration is mandatory due to limited seats. Zoom details will be shared with Registered participants only.

For further information please contact:
National Inbred | Department of Pathology & Lab Medicine | 011-346-1921 | medlab@medakuh.edu.pk

RESEARCH

Surveys

Two surveys on newborn screening were planned and conducted after ethical review approval. The first was conducted on mothers residing in Pakistan titled "Awareness & Attitude about Neonatal Screening of Mothers in Pakistan (n=208). The second survey was done on pathologists in Pakistan titled "Survey of Pathologists for Better Understanding of Current Practices & Need of NBS in Pakistan (n=49)". Findings of both surveys will be shared with readers in near future.

Grants

Dr. Hafsa Majid received a Scholarship of Teaching and Learning (SoTL) Grant of 3129 USD for the project titled 'Curriculum Development using a Virtual community of practice (CoP) for postgraduate trainees to enhance the Knowledge of Inherited Metabolic Disorders (IMDs)' by Teaching and Learning Network.



Dr. Aysha Habib Khan was awarded a grant by the Society for the Study of Inborn Errors of Metabolism (SSIEM), London to support an event for patients and families with inherited metabolic defects. The hybrid event was organized on 1st March 2021 at the occasion of world Rare Disease Day to launch Ek-Sath! E-portal.

IFCC's Taskforce on Global Newborn Screening Implementations

Dr. Aysha Habib Khan represents Pakistan Society of Chemical Pathologists in the Academic Council Of SSIEM and serves as a member of IFCC's Taskforce on Global Newborn Screening Implementations in low resource settings. During her first term in office, Dr. Khan has facilitated a global survey on newborn screening need assessment in low middle income countries.

Learn more about the Task Force here: <https://www.ifcc.org/executive-board-and-council/eb-task-forces/task-force-on-global-newborn-screening/>

International Neonatal Screening Day
28 June 2021

[Click here to learn more](#)



The Task Force NBS members during their inaugural meeting. Prof Adeli, IFCC President participated into the meeting. Dr U. Ceglarek, not portrayed here, was in attendance as well.

WORKSHOPS

Newborn Screening in Pakistan: Bridging the Gap, Educate and Enlighten – April 13, 2021

Organized by the Aga Khan University, an arm of Pak-IMD-Net. This giant workshop invited experts, public and private institutions and policymakers from all over Pakistan to bridge the gap between their understanding about Newborn Screening and to develop a workforce for its successful implementation. From Sindh, experts from multiple departments at AKUH, Indus Hospital, DoctHERs, and members of the Sindh Provincial Assembly attended. From Punjab, faculty of Shalimar Medical College, Fatima Memorial Hospital, and representatives of ZB Foundation attended physically, while other participants joined via Zoom.

The agenda included discussion of the essential components of NBS according to the criteria of the American College of Medical Genetics, detailed by Dr. Lena Jafri. For a disease to be included in the NBS program, it must be at As Congenital Hypothyroidism (CHT) is the most common inherited metabolic disorder and fulfils Wilson and Jungner classic screening criteria, it can be centered in national pilot NBS program as a prototype.

Ms. Rabia Azfar Nizami, Member of the Sindh Provincial Assembly, briefed the participants about the legislative framework involved for implementing the NBS program. Currently there are 3 legislations for NBS in Pakistan (2013, 2014 and 2019) which need to be elaborated for guidelines to be implemented in the public and private sector. Subject matter specialists are not part of the legislation process, but can be called for amendments in the law. Recently experts from AKUH have been consulted to fill the gaps in NBS legislations. A call was raised to form a multifaceted body to work on NBS framework together.

According to the data shared by the various institutes and organizations in the forum, the incidence of CHT in Pakistan is approximately 1:1300 live births, higher than rates found in the West (1:4000 live births).

After passing of the Sindh Newborn Screening Act of 2019, National Institute of Child Health (NICH) has been performing NBS for in-patients and referred patients from the province since 2020. AKUH in Karachi has screened 96% of its patients for CHT since 2019, and since 2020, NBS for Congenital Adrenal Hyperplasia (CAH) is also being performed.

NBS is not performed on a public level in Punjab, however, several hospitals and organizations perform it privately. Fatima Memorial Hospital has the oldest NBS program in the region, having conducted NBS for CHT since 2007. ZB Foundation is a welfare-run non-profit organization working on advocacy and institutionalization of NBS in Pakistan since 2016. Inmol Institute performs NBS for CHT free of cost at 8 different private hospitals in Lahore. As the NBS bill for Punjab is yet to be passed, no public sector hospitals are included in the program.

There are several ethical, economical and clinical issues in NBS that require clear understanding before implementation of an NBS program. To address the challenges that come up in follow-up for confirmatory tests, representatives from Naya Jeevan explained how the monitoring of positively screened patients could be supported by storing the data in an EMR system, which could in the future be integrated into patients' identity cards.

Furthermore, the need to build NBS specific screening and diagnostic laboratories is essential as they require micro-techniques according to CLSI and CAP guidelines. Economic evaluations can assess cost effectiveness and cost utility analyses for evidence based decision making in healthcare resource allocation for NBS. All hospitals, institutes, and organizations should collect patient data about NBS to contribute towards building a nationwide database registry, the template for which was suggested to be shared by AKUH.

At Dr. Aysha Habib Khan's suggestion to develop a rare disease/NBS consortium, it was mutually decided to establish a task force on NBS to decide end goals, identify stakeholders, build structure on partnerships, and establish a solid project management structure and timebound projects. The taskforce should comprise of national chemical pathologists, hematologists, pediatricians and obstetricians.

Flipped Style Workshop on Developing Education and Awareness Guides for Families for Newborn screening - April 27, 2021

Under the directorship of Dr. Lena Jafri and Dr. Hafsa Majid this workshop was conducted with the objectives to develop guides and educational material for mothers/ parents coming to antenatal clinics, to develop standardized message /email for informing positive newborn screening results to families and to develop story board for video for education of NBS Program. Participants included Pediatricians, Obstetricians, Biochemical Geneticists, ENT specialists, Hematologists, Nurse managers, Chemical Pathologists and Biochemical Genetics Scientists. Participants worked in 4 groups to develop the flyers and educational materials. Then a Gallery Walk was conducted to review and improve each other's group outcome. The workshop ended once each group presented their educational material related to Congenital hypothyroidism, Congenital adrenal hyperplasia and newborn screening of Hearing.

This was a fruitful multi-workshop.



A Round Table Discussion: Reviewing Markers, Biochemical Pathways & Algorithms for Expanded Newborn Screening in Pakistan’ – November 18, 2021

After a short welcome by the Chair of Pak-IMD-Net the learning objectives of the meeting were outlined. Dr. Sibtain Ahmed identified the common interests in the diverse participating pathologists, metabolic physicians, biochemical genetics technologists, scientists and chemical pathology residents present in the meeting.



Dr. Ahmed explained the group that the validation of dried blood spot acylcarnitine, amino acids and succinyl acetone was successfully completed on LCMS/MS. The group was gathered to identify the limitations, challenges and solutions for initiating expanded newborn screening. The participants were split into two teams and directed to list down eight commonly encountered inherited metabolic disorders (IMDs) in our country based on urine organic acid testing, plasma amino acids testing and urinary succinyl acetone lab data. Next, the two tables battled head to head to describe the second tier and confirmatory tests for follow up testing of various IMDs, and review pathways and algorithms of the identified disorders. At the end of the activity, both teams presented their discussion points and the outcomes of the activity were shared with the entire hall.

RARE DISEASE DAY 2020 REFRAMING RARE IN PAKISTAN BREAKING SILOS AND BRINGING SYNERGIES

**Organized by Pak-IMD-Net in collaboration with the Department of Pathology & Laboratory Medicine and Division of Women & Child Health, Aga Khan University
March 4-7, 2020**

This multidisciplinary 4-day conference gathered experts from around the world to break dialogue on (not so) rare genetic diseases under the auspices of International Federation of Clinical Chemistry & Laboratory Medicine (IFCC)

International experts from USA, Italy and New Zealand addressed the symposia and facilitated workshops to train the local work force for establishment of Newborn Screening in Pakistan.

Many national and international institutes participated actively in the conference, including the following:

Ziauddin University, National Institute of Child Health (NICH), Dow University of Health Sciences (DUHS), National Institute of Blood Disorders (NIBD), Liaquat National Hospital, 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

The following National Societies were part of the conference:

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)



Day 1 and 2; Pre-symposium workshops of the Conference - March 4 and 5, 2020

These workshops were held as a preamble to the symposia on day 3 onward at JHS Auditorium, Aga Khan University Hospital. Participants included medical professionals, M. Phil and Ph.D students, clinicians and allied professionals with an area of interest in the workshops.

Workshop on Bioinformatics hands-on on IMD genomics

Facilitated by Dr. Waqasuddin, Prof Shahid Mahmood Baig, Dr. Sibtain Ahmed, Ms. Kehkashan Imam and Ms. Misha Ahmed, this half-day workshop provided insights into the fast-evolving field of Bio-informatics and gave an impetus for developing an understanding of Next Generation DNA Sequencing (NGS) technology and data interpretation related to biochemical genetics. High throughput, massively paralleled NGS technology has revolutionized the field of genomics, and has tremendous applications in biomedicine, agriculture, and biotechnology. Using NGS, an entire human genome can be sequenced within a few hours. 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.

Workshop on Establishing Registry for Rare Diseases

Facilitated by Drs. Zahra Hoodbhoy, Hafsa Majid, Adnan Jabbar, Mustafa Aslam, M Nadeem Anjum, Lena Jafri and Aysha Habib Khan, this half-day workshop was specifically designed for clinicians, pathologists, post-doctoral fellows, and M. Phil. / Ph. D. students who planned to develop registry in their area of interest. The agenda involved a thorough walkthrough on developing a registry. Identifying your registry's aim and objectives, defining your registry dataset, establishing and sustaining a registry data collection, quality assurance, and resourcing and funding were some of the aspects discussed by Dr. Hoodbhoy, while Dr. Aslam elaborated the ethical considerations of developing a registry. He explained that the process requires adequate knowledge of governance, policies, ethics of registry data protection, and sharing and transfer of data. As prototypes for software selection, Dr. Jabbar shared working examples of the Cancer Registry at AKUH , while Dr. Anjum shared information about lysosomal storage disease registries. The workshop ended with a talk on how to break the silos in developing laboratory consortia in inherited metabolic diseases.



Workshop on Genetic Counseling: How to do it right?

Targeted to medical practitioners, clinicians, pathologists, post-doctoral fellows, M. Phil. / Ph. D. students, and corporate professionals working in the areas of genomics, this half-day workshop provided valuable insights in the fast-evolving field of genetic counseling. Facilitators briefed the audience about how to effectively deliver knowledge about the genetic aspects of illnesses with those who are at an increased risk of either having a heritable disorder or of passing it on to their unborn offspring. Participants gained understanding of the inheritance pattern of illnesses and their recurrence risks; learned how to address the concerns of patients, their families, and their health care providers; and how best to support patients and their families dealing with these illnesses. Dr. Amy R.U.L. Calhoun, the Clinical Associate Professor & Medical Director of Iowa Newborn Screening Program briefed the participants to the concepts of genetic counselling, emphasizing the evolving need for healthcare professionals trained in clinical genetics and genetic counseling in mainstream clinical practice. Dr Salman Kirmani presented the inheritance patterns and pedigrees in understanding and identifying inheritance patterns of genetic disorders, and the impact on disease recurrence risk in families. He also taught the audience how to interpret familial disease history with the use of pedigrees. Case studies were shared by all facilitators. Ms. Fizza Akbar focused on effective ways to communicate the facts and showing empathy to the affected patients and their families, followed by role plays by participants.

Workshop on Essentials of Establishing a Newborn Screening Program

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

This closed group symposium connected pathologists, pediatricians and obstetricians from different institutes to understand the essential requirements of establishing a Newborn Screening (NBS) program. Featuring presentations on the technical and practical elements of setting up a laboratory to perform newborn screening, and talks on experiences from different centers and challenges of implementing newborn screening programs, this half-day workshop was specifically designed for healthcare personnel wishing to establish newborn screening services at their institutes.

The workshop was opened by a talk on the essential components of the NBS program, then bringing our international collaborator Dr. Dianne Rosemary Webster to the forum. The director of the Newborn Metabolic Screening Programme at LabPlus, Auckland City Hospital taught the participants practical considerations of newborn screening- applying Wilson Jungner criteria, selecting disorders, screening, confirmatory analytes and cutoffs, and establishing follow-up services. The AKUH faculty expanded on the laboratory's requirements for infrastructure, instrumentation, and sample collection. Quality control issues and reporting requirements, external proficiency, and method validation requirements were also discussed.



Day 3 of the Conference, Inaugural Session March 6, 2020



After keynote speeches by the Chair and Co-Chair, the speakers took the floor to deliver the talks of the day.

State of the Genetic Diseases in Pakistan and Prevention Strategies by Professor Shahid Mahmood Baig (PhD, Sitara-e-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 by 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 by Professor Aysha Habib Khan, Department of Pathology and Laboratory Medicine, Aga Khan University Hospital (AKUH)

The session also had students from the AKU Medical College performing a mime show titled **Deafening Silence** to symbolize the neglect displayed around rare diseases. Esteemed Chief Guest Professor Abdul Gaffar Billoo and Patron Prof Mohammad Khurshid then addressed the audience and shared their thoughts about the event. After a brief closing speech by Professor Imran Siddiqui, AKUH, the participants exited the auditorium to the courtyard for a **Walk for Rare** in solidarity with patients and families affected by IMDs.



Conference Symposium

March 7, 2020; Venue: AKU Auditorium

After watching glimpses from the past conference on Rare Diseases, 2019, the symposium program officially began.



Symposium I - Outcome & Challenges in Management of Inherited Metabolic Disorders

The first symposium was chaired by Dr. DS Akram and Dr. Lumaan Sheikh and moderated by Dr. Sidra Arshad. Dr. Giancarlo La Marca, Professor, Clinical Biochemistry and Clinical Molecular Biology, University of Florence, and President of the Italian Society for Newborn Screening, joined virtually to give a video presentation on the learning outcomes in metabolic disorder screening by mass spectrometry. Obstetrician and Professor Shama Munim from AKUH discussed the detection of rare diseases when presented antenatally. Expanding on challenges related to NBS, Professor Huma Cheema from Children's Hospital, Lahore, shared the experiences of management of IMDs at her institution. Dr Bushra Afroze from AKUH then closed the session by taking a glimpse into the future and envisioning the ten year outcome in management of IMDs in Pakistan, opening the floor for a participant led Q & A session.

Symposium II - Newborn Screening

This symposium was chaired by Professor Huma Cheema and Brig (R) Professor Aamir Ijaz and moderated by Dr. Ghulam Zainab. The program commenced with a talk by Ms. Dianne Webster, who discussed the complex conditions involved in discontinuing screening programs. She shared the Auckland District Health Board's encounters of performing antenatal screening for Down syndrome and other conditions, This was followed by presentations of local programs detailing their experiences, Dr. Farkhunda Ghafoor shared challenges of running the NBS program in Shalimar Medical & Dental College, Punjab. Drs. Hafsa Majid and Khadija Nuzhat Humayun followed from AKUH, sharing the outcomes of short-term and long-term follow-ups of previously screened patients at the clinical laboratory and Department of Paediatrics and Child Health at AKUH. Last of the speakers was Dr. Ayesha Raza, program manager at "The ZB Foundation", a non-profit organization dedicated to performing NBS free of cost for the public.

Symposium III- Complex Clinical Cases: Challenges Faced by Experts Dealing with Rare Diseases

Chaired by Professor Shahid Pervez and Professor Shahid Baig, and moderated by Dr. Rehana Rehman, this symposium gathered various AKUH faculty on the podium to share rare cases from their respective departments. In his opening presentation, Dr. Salman Kirmani from the Department of Paediatrics and Child Health, AKUH remarked on his experience establishing a genetics service at the hospital. He shared his future visions and dreams for the program, sharing the mic with Professor Dr. Aysha Habib Khan from the Department of Pathology & Laboratory Medicine, who shed light on the rare presentations of osteoporosis. Dr. Sadia Masood from the Department of Medicine shared rare case studies presenting in her Dermatology clinics, while Dr. Saira Fatima shared cases from the Section of Histopathology, The session was closed by Dr. Muhammad Awais' talk on radiological diagnosis of rare diseases.

Symposium IV- Rare Hematological Disorders: Not so Rare!

This symposium was chaired by Prof Parvez Ahmed and Prof Salman N Adil, and moderated by Dr. Muhammad Shariq. Maj Gen (R) Dr Suhaib Ahmed from Riphah International University, Islamabad opened the symposium with a riveting presentation on the commonly occurring "rare" genetic hemoglobin disorders. Dr. Anila Rashid from the Section of Hematology at AKUH focused her talk on visualizing the way forward in managing inherited bleeding disorders. Prof Dr. Bushra Moiz closed the session by sharing insights on diagnosis and testing of Inherited Thrombophilia, and the crucial decisions that go into deciding the right tests for the right patients. An active Q & A session ensued.

Symposium V- Laboratories, Quality and Ethics for Rare Diseases

Chaired by Dr. Farooq Ghani and Maj Gen (R) Dr Suhaib Ahmed, and moderated by Dr. Fatima Khanani, this was the last symposium in the program. Prof Aamir Ijaz shared his perspective on the current state of laboratories, quality and ethics for rare disease diagnosis and management in Pakistan. Dr. Saima Siddiqui from National Institute of Blood Disorders (NIBD) discussed the crucial role of clinicians in interpretation of molecular tests in rare disease diagnosis. This was followed by the AKUH faculty of Pathology and Laboratory Medicine presenting multiple talks on laboratory practices for Biochemical Genetics Laboratory, reframing the concept of rare disease in Pakistan to raise public awareness.

Next was a table discussion on policy recommendations by the rare disease experts in attendance, moderated by Dr. Salman Kirmani & Prof Dr. Aysha Habib Khan, and the symposia officially ended with the Closing Ceremony and Awards Distribution by Prof Dr. Afia Zafar, Pathology and Laboratory Medicine, AKUH.

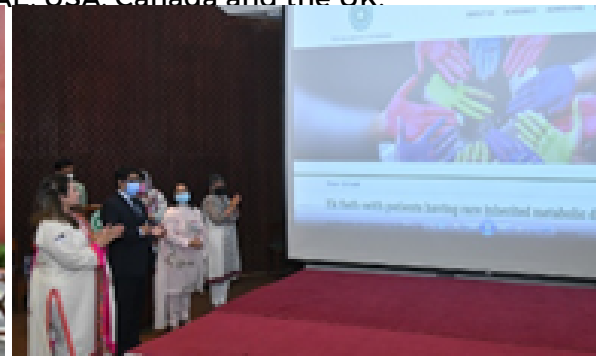


RARE DISEASE DAY 2021 EK-SATH!

March 1, 2021

For Pakistani families affected by IMDs, there is a paucity of appropriate health services and support groups, making it difficult for them to find accessible information and support for their life-altering conditions.

Ek-Sath! is a collaborative effort between Pak-IMD-Net, PSCP and AKUH, created to provide an infrastructure for support of patients and families with rare IMDs in Pakistan and help in dealing with challenges in their diagnosis & management. This was supported by Society for the Study of Inborn Errors of Metabolism (SSIEM) London. The Ek-Sath e-portal was launched on March 1st, 2021. Due to COVID-19 restrictions, the event was attended by a few selected patients and their families physically while over 100 viewers were in virtual attendance from Pakistan, Afghanistan, UAE, USA, Canada and the UK.

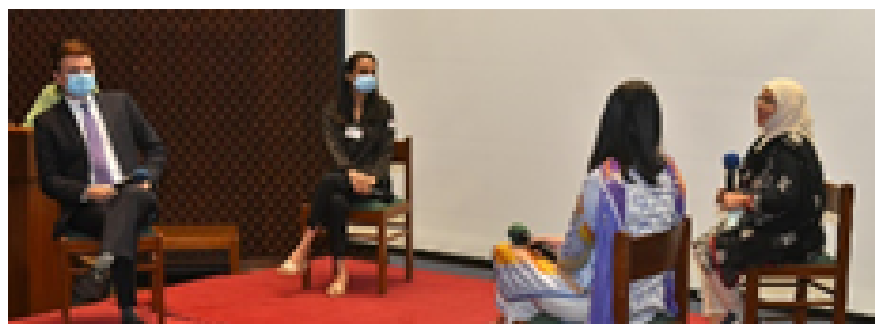


Highlights of the Event

After launch of Ek-Sath web portal by Mrs. Reema Ismail, wife of Governor Sindh, a storytelling session was held for patients and professionals to address challenges, diagnosis and better management of patients with IMDs.

A riveting puppet show was organized by Thespianz Theatre to promote awareness on rare diseases and send positive vibes to the patients and families.

School-hospital partnerships and research collaborations were discussed by the professionals in attendance.



The types of activities proposed for the parents and families on the e-portal of Pak-IMD Net include:

- Awareness & education on IMDs through flyers, brochures and newsletter.
- Events organization on IMDs
- Family Support Group Sessions
- Rare Stories by Rare Patients and Families
- Invites to join in for research
- Blogs

For more information visit <https://www.aku.edu/ek-saath/Pages/home.aspx>

Publications, Talks, and Presentations on Ek-SATH Event:

1. https://www.aku.edu/news/Pages/News_Details.aspx?fbclid=IwAR3QNJwod Q4Y11K3hZTt-eNdYl0cdwHH0IG5EsCinqVlnznUkurBEFuuG4&nid=NEWS-002461
2. <https://runwaypakistan.com/puppet-show-held-at-aku-to-create-awareness-on-rare-diseases/>
3. <https://images.dawn.com/news/1186655>
4. <https://www.dailyadvent.com/news/f568e442a91f9e04b54a06flead3bea6-Thespianz-Theatre-organises-puppet-show-to-educate-people-on-rarediseases>
5. <https://www.facebook.com/watch/?v=723531471679809>
6. https://m.facebook.com/story.php?story_fbid=10165051423540245&id=644415244&sfnsn=scwspmo

Health First Show on DAWN News

February 28, 2021

To raise awareness of rare inherited metabolic disorders and newborn screening in Pakistan, Dr. Lena Jafri along with Dr. Mohsina Ibrahim from National Institute of Child Health (NICH) appeared on the Health First Show on Dawn News on World Rare Disease Day and discussed the importance of newborn screening of salvageable diseases like congenital hypothyroidism and congenital adrenal hyperplasia.



Watch the full interview at <https://www.youtube.com/watch?v=620gpCOJSm8>

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, Mohi Uddin Islamic Medical College, Mirpur, AJK

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Dr. Aysha Habib Khan, Professor, Department of Pathology and Laboratory Medicine, AKUH, Karachi
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Dr. Sibtain Ahmed, Senior Instructor, Department of Pathology and Laboratory Medicine, AKUH, Karachi
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Dr. Madiha Beg, Assistant Professor, Department of Medicine, AKUH, Karachi
Dr. Siraj Muneer, Resident Chemical Pathology, 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
Farhat Jahan, Technologist, BioGenetics Laboratory, AKUH, Karachi
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Dr. Shabnam Dildar, Consultant Chemical Pathology, National Institute of Blood Diseases, Karachi
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
Dr. Sheharbano Imran, Consultant Chemical Pathology, OMI Hospital, Karachi
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Maj Dr. Safia Fatima, Chemical Pathologist, Armed Forces Institute of Pathology, Rawalpindi
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Dr. Nusrat Alavi, Associate Professor, Chemical Pathology, Rahbar Medical and Dental College, Lahore
Professor Dilawar Khan, Department of Chemical Pathology, Shalamar Medical & Dental College, Lahore
Dr. Masood Afzal, Demonstrator, Department of Pathology, Shalamar Medical & Dental College, Lahore
Dr. Tayyaba Rashid, Demonstrator, Department of Pathology, Shalamar Medical & Dental College, Lahore
Dr. Khushbakht Adnan, Consultant, Balochistan Institute of Child Health, Quetta, BICHQ
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

CALL FOR NEW MEMBERS



In the past two years, the Pak-IMD-Network has grown to 32 active and associate members. We are a Community of Practice. Community of Practice is a group of people who "share a concern or a passion for something they do and learn how to do it better as they interact regularly". Our mission and vision is capacity building of laboratorians to develop diagnostics of newborn screening and IMDs in Pakistan and to improve diagnostics and care of patients with IMDs. A prevailing memory of 2021 will be the fact that science, and more specifically open science, delivered solutions for one of the biggest challenges of our time.

Since Pak-IMD-Net was founded in 2019, we have stood for open science and our mission remains unchanged. Throughout 2020 and 2021, thanks to the enormous efforts of everyone who is part of our wonderful community we reached several new and important milestones in the history of laboratory science in Pakistan.

Dear PSCP members, feel free to contact us if you are involved and interested in IMD testing and research. And as always, identify and invite new members to contribute to our cause.

Hoping for another bright year ahead!

Register to be a member of Pak-IMD-Net here: <https://forms.gle/48WJfJpwbQKocUn4A>

