



# ISIEM 2024

**TEAMING UP FOR TREATABLE IEMs**

**NATIONAL CONFERENCE OF INDIAN SOCIETY FOR INBORN ERRORS OF METABOLISM**

**IEMs-USHERING IN THE THERAPEUTIC ERA**

Date : 20<sup>th</sup> to 22<sup>nd</sup> September 2024

Venue : Rajasthan International Centre, Jhalana Doongri, Jaipur

*Scientific*  
**PROGRAMME**

[WWW.ISIEM2024.COM](http://WWW.ISIEM2024.COM)



Day 1 | 20<sup>th</sup> September 2024 | Friday

Registration Starts at 08:00 AM Onwards

### Workshop - 1 | Lab Diagnosis of IEMs

**Dr. Alpa Dherai, Dr. Anil Jalan, Dr. Usha Dave, Dr. Jayesh Sheth, Dr. Lokesh Kumar Agrawal & Dr. Chakshu Choudhary**

Time	Topic	Faculty
09:00 AM - 09:30 AM	Basic metabolite based approach to IEMs (GELAK)	Dr. Lokesh Kumar Agrawal
09:30 AM - 10:00 AM	The Tandem approach of TMS	Dr. Anil Jalan
10:00 AM - 10:30 AM	Organic acidurias - The GCMS profiling	Dr. Usha Dave
<b>10:30 AM - 10:45 AM</b>	<b>Tea Break</b>	
10:45 AM - 11:15 AM	Special Investigations in diagnosing IEMs	Dr. Alpa Dherai
11:15 AM - 11:45 AM	Enzyme Based Diagnosis	Dr. Jayesh Sheth
11:45 AM - 12:30 PM	Challenging Cases	Dr. Chakshu Choudhary

### Workshop - 2 | Dietary Management of IEMs

**Lead : Dr. Rani Singh (PhD,RD,LD), Professor Emory University, Department of Human Genetics**

**Genetic Metabolic Nutritionists (India) Mrs. Anuja Agarwala, Dr. Sheetal Mhamunkar, Mrs.Ranjini Parthasarathy, Mrs. Vaishali Madkaikar, Mrs.Safala Mahadik, Dr. Malathi Voora, Mrs. Surbhi Pareek & Mrs. Shipra Srivastava**

09:00 AM - 09:10 AM	Introduction of the workshop, Special guest (Dr. Rani Singh) and Indian team. Release of the workshop book Release of resource material	Dr. Rani Singh Dr. Priyanshu Mathur	
09:10 AM - 09:20 AM	Pretest	Mrs. Shipra Srivastava and Mrs.Surbhi Pareek	
09:20 AM - 09:40 AM	Keynote address-Importance of nutrition and diet in IEMs and - Overview - Nutrition Management in IEMs	<b>Speaker :</b> Dr. Rani Singh	<b>Chairpersons :</b> Dr. Seema Kapoor
<b>09:45 AM - 10:30 AM</b>	<b>Session - 1 Introduction to Genetic Metabolic Disorders</b>		
		<b>Speaker</b>	<b>Chairpersons</b>
15 Min	General metabolic pathways	Mrs. Vaishali Madkaikar	Dr. Rani Singh
15 Min	CHO metabolic disorders	Mrs. Ranjini Parthasarathy	
15 Min	Amino acid disorders + UCD	Mrs. Sheetal Mhamunkar	
<b>10:30 AM - 10:45 AM</b>	<b>Tea Break</b>		
<b>10:45 AM - 11:30 AM</b>	<b>Session I (Contd) : Introduction to Genetic Metabolic Disorders</b>		
		<b>Speaker</b>	<b>Chairpersons</b>
10 Min	Fat Metabolic disorders	Mrs.Safala Mahadik	Dr. Rani Singh
10 Min	Emergency regimen	Mrs. Anuja Agarwala	
10 Min	Use of indigenous foods	Dr. Malathi Voora	
10 Min	Q & A with audience & Expert comments by Chair	All Speakers & Chairperson	
<b>11:30 AM - 12:30 PM</b>	<b>Session 2 Case Studies and Quiz (Interactive Session)</b>		
<b>40 Min</b>	Case presentations by groups, followed by discussion/ Q & A	Mrs. Shipra Srivastava and Mrs. Surbhi Pareek	Dr. Rani Singh
<b>20 Min</b>	<b>Quiz</b>		

### Workshop - 3 | Newborn Screening (NBS) - Follow-up & Management

**Faculty - Dr. Seema Kapoor, Dr. Shankar V H, Dr. Shruti Bajaj, Dr. Kanika Singh  
Dr. Suvarna Magar & Dr. Swasti Pal**

Time	Topic	Faculty
09:00 AM - 09:30 AM	The NBS Program	Dr. Seema Kapoor
09:30 AM - 10:00 AM	Impact of NBS on Society	Dr. Kanika Singh
10:00 AM - 10:30 AM	Dealing with Screen Positive Result	Dr. Suvarna Magar

<b>10:30 AM - 10:45 AM</b>	<b>Tea Break</b>	
10:45 AM - 11:15 AM	Outcome of NBS Detected Cases	Dr. Shankar V H
11:15 AM - 11:45 AM	Newer Frontiers in NBS	Dr. Shruti Bajaj
11:45 AM - 12:30 PM	Challenging Cases	Dr. Swasti Pal
<b>12:30 PM - 01:30 PM</b>	<b>Lunch</b>	

### Venue : Main Auditorium, Ground Floor

### Workshop - 4 | Workshop Basics of IEMs : How to Recognize & Diagnose

#### Complimentary for all Attendees

01:30 PM - 02:00 PM	Inherited Metabolic Diseases from Past to Present	<b>Speaker :</b> Dr. Radha Rama Devi	<b>Chairpersons :</b> Dr. Rani H Singh Dr. Sandeep Mathur Dr. Madhu Raturi Dr. Kusum Devpura
02:00 PM - 02:35 PM	Small molecule metabolic disorders : Case Based Discussion	<b>Moderator :</b> Dr. Chaitanya Datar	<b>Panelist :</b> Dr. Madhulika Kabra Dr. Sunita Bijarnia Dr. Sujatha Jagadeesh Dr. Jaikishan Mittal Mrs. Vaishali Madkaikar
02:35 PM - 03:10 PM	Complex molecule metabolic disorders : Case Based Discussion	<b>Moderator :</b> Dr. Aradhana Dwivedi	<b>Panelist :</b> Dr. Uma Rama Swami Dr. Ratna Dua Puri Dr. Meenakshi Bhat Dr. Vivek Jain Dr. Sheetal Mhamunkar
03:10 PM - 03:45 PM	Energy Deficiency Disorders : Case Based Discussion	<b>Moderator :</b> Dr. Udaya Kotecha	<b>Panelist :</b> Dr. Neerja Gupta Dr. Mamta Muranjan Dr. Lokesh Saini Dr. Akash Sharma Mrs. Anuja Agarwala

**03:45 PM - 04:00 PM** Tea Break & Poster Viewing

## National Conference of ISIEM 2024

### Venue : Main Auditorium, Ground Floor

04:00 PM - 04:10 PM	Welcome address	Dr. Priyanshu Mathur	
04:10 PM - 04:20 PM	ISIEM Presidential Address	Dr. Seema Kapoor	
04:20 PM - 05:00 PM	Oration	<b>Speaker :</b> Dr. Uma Ramaswami	<b>Chairpersons :</b> Dr. Radha Rama Devi Dr. Sunita Bijarnia Dr. Jayesh Sheth Dr. Kuldeep Singh
05:00 PM - 05:30 PM	Keynote lecture - Biomarkers - Not everything worthwhile can be measured and not everything measured is worthwhile.	<b>Speaker :</b> Dr. Seema Kapoor	<b>Chairpersons :</b> Dr. Pankaj Agarwal (USA) Dr. Anil Jalan Dr. Alpa Dherai Dr. Kailash Meena

**05:30 PM - 06:00 PM** Inauguration Ceremony

### Gala Dinner



# Day 2 | 21<sup>st</sup> September 2024 | Saturday

Venue : Main Auditorium, Ground Floor

Time	Topic	Judges :
08:00 AM - 09:00 AM	Platform presentation - 6 (10 min each)	Dr. Inusha Panigrahi, Dr. Anil Jalan & Dr. Divya Agarwal

## Morning Sessions - IEMs with Established Treatments

09:00 AM - 09:30 AM	Nutritional management of IEMs with prototype as PKU	<b>Speaker :</b> Dr. Rani H Singh	<b>Chairpersons :</b> Dr. Neelam Mohan Dr. Deepak Shivpuri Dr. Jyoti Bagla Dr. Neeta Jindal
09:30 AM - 10:00 AM	Management of IEMs during acute crisis	<b>Speaker :</b> Dr. Uma Maheshwari	<b>Chairpersons :</b> Dr. Nagaram Dande Dr. Girisha K M Dr. R N Sehra Dr. Prashant Mitharwal
10:00 AM - 10:30 AM	Vitamin responsive epilepsies & epileptic encephalopathies	<b>Speaker :</b> Dr. Ajay Goenka and Dr. Varsha Vaidya	<b>Chairpersons :</b> Dr. Bhawana Sharma Dr. Dinesh Khandelwal Dr. Anurag Tomar Dr. Ramesh Choudhary
10:30 AM - 11:00 AM	Diagnosis and Management of Neurotransmitter Disorders	<b>Speaker :</b> Dr. Lokesh Lingappa	<b>Chairpersons :</b> Dr. Pradeep Meena Dr. Rupesh Masand Dr. Juhi Gupta Dr. Meenal Garg

**11:00 AM - 11:20 AM** Tea Break & Poster Viewing

## Sessions- Managing IEMs in Genomic Era

11:20 AM - 11:50 AM	Role of Genomics and other omics approaches in metabolic disorders	<b>Speaker :</b> Dr. Pankaj Agrawal (USA)	<b>Chairpersons :</b> Dr. Ashok Sharda Dr. B.M Raturi Dr. Devendra Sareen Dr. S P Sethi
11:50 AM - 12:20 PM	Diagnosing the undiagnosed in IEMs	<b>Speaker :</b> Dr. Ratna Dua Puri	<b>Chairpersons :</b> Dr. Kuldeep Singh Dr. Ashwini Kamdar Dr. Anupa Prasad Dr. Mithu Banerjee
12:20 PM - 12:50 PM	Making sense of VOUSes in metabolic Disorders	<b>Speaker :</b> Dr. Sheetal Sharda	<b>Chairpersons :</b> Dr. Rambabu Sharma Dr. Sanjeev Hooja Dr. Tarun Patni Dr. Mohit Vohra
12:50 PM - 01:20 PM	Management of IEMs in the Genomic Era- Small Molecule and Mitochondriopathies	<b>Speaker :</b> Dr. Chaitanya Datar	<b>Chairpersons :</b> Dr. Bhawana Dhingra Dr. Pramod Sharma Dr. Rakesh Jora Dr. Shyam Agrawal

**01:20 PM - 02:00 PM** Lunch & Poster Viewing

## Afternoon Sessions - Lysosomal Storage Disorders

02:00 PM - 02:30 PM	ERT in Gaucher Disease patients : Treatment outcome and monitoring	<b>Speaker :</b> Dr. Uma Ramaswami	<b>Chairpersons :</b> Dr. Dhananjay Mangal Dr. Kapil Sharma Dr. Deependra Garg Dr. Sunil Dutt Sharma
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02:30 PM - 03:00 PM	Genotype phenotype correlation in Gaucher disease patients	<b>Speaker :</b> Dr. Jayesh Sheth	<b>Chairpersons :</b> Dr. Kapil Garg Dr. Richa Choudhary Dr. J P Narayan Dr. Anshul Mittal
03:00 PM - 03:30 PM	Treatment challenges in MPS II patients	<b>Speaker :</b> Dr. Sujatha Jagadeesh	<b>Chairpersons :</b> Dr. Atul Shankar Dr. Siyaram Didel Dr. Amrita Mayanger Dr. Kamlesh Agrawal
03:30 PM - 04:00 PM	Panel Discussion Fabry - An adult disease : myth or reality.	<b>Moderator :</b> Dr. Kausik Mandal	<b>Panelist :</b> Dr. Meenakshi Bhat Dr. Arushi Gahlot Dr. Moni Tuteja Lt Col (Dr) Puneet Singh Dr. Jayarekha Dr. Ashka Prajapati Dr. Yashu Saini
<b>04:00 PM - 04:20 PM</b>	<b>Tea Break &amp; Poster Viewing</b>		
04:20 PM - 04:50 PM	Diagnosis & Management of Pompe Disease	<b>Speaker :</b> Dr. Sheela Nampootheri	<b>Chairpersons :</b> Dr. Madhu Chaudhary Dr. Rajesh Mathuria Dr. Lalit Bhariadiya Dr. Munish Kakkar
04:50 PM - 05:20 PM	Advances in Management of ASMD	<b>Speaker :</b> Dr. Neerja Gupta	<b>Chairpersons :</b> Dr. S D Sharma Dr. Anupam Chaturvedi Dr. Roop Sharma Dr. Natwar Parwal
05:20 PM - 06:00 PM	Introduction of NPRD - Dr. Madhulika Kabra Panel Discussion - Optimal use of available resources & NPRD	<b>Moderator :</b> Dr. Madhulika Kabra	<b>Panelist :</b> Dr. L. Swasticharan Dr. B. S. Charan Dr. Shankar Hariharan Dr. Kuldeep Singh Dr. Sanjeeva Dr. Inusha Panigrahi
06:00 PM - 06:30 PM	Debate - Diagnosis of Lysosomal Storage Disorders - Enzyme first Vs Genomics first <b>Moderator :</b> Dr. Divya Agarwal	<b>Panelist :</b> Dr. Beena Suresh Dr. Ravneet Kaur Dr. Sameer Bhatia Dr. Mayank Nilay	<b>Judges :</b> Dr. Seema Thakur Dr. Ratna Dua Puri Dr. Divya Agarwal
<b>06:30 PM - 07:30 PM</b>	<b>General Body Meeting of ISIEM</b>		



## Day 3 | 22<sup>ND</sup> September 2024 | Sunday

Time	Topic	Judges :	
08:00 AM - 09:00 AM	Platform presentation - 6 (10 min each)	Dr. Inusha Panigrahi, Dr. Anil Jalan & Dr. Divya Agarwal	

### Morning Sessions -Newer, Innovative Treatments & Prenatal Diagnosis

09:00 AM - 09:30 AM	Metabolic Liver Diseases	<b>Speaker :</b> Dr. Nishant Wadhwa	<b>Chairpersons :</b> Dr. Abhinav Sharma Dr. Ashish Agrawal Dr. Manish Mittal Dr. Sanjeev Gupta
09:30 AM - 10:00 AM	Newer Therapies in Leucodystrophies	<b>Speaker :</b> Dr. Sunita Bijarnia	<b>Chairpersons :</b> Dr. Renu suthar Dr. Rahul Gupta Dr. Meenakshi Lallar Dr. Varnit Shankar
10:00 AM - 10:30 AM	Liver Transplantation for IEMs	<b>Speaker :</b> Dr. Khalid Al Thihji	<b>Chairpersons :</b> Dr. Lakhnan Poswal Dr. Komal Uppal Dr. Md Asif Dr. Gajanand Tanwar
10:30 AM - 11:00 AM	Genetic Counselling in Prenatal Diagnosis of IEMs- Why & When?	<b>Speaker :</b> Dr. Usha Dave	<b>Chairpersons :</b> Dr. Sonia Meel Dr. Rekha Gupta Dr. Megha Sharma Dr. Aditi Bansal

**11:00 AM - 11:20 AM** Tea Break & Poster Viewing

### Sessions - IEMs - Special Situations

11:20 AM - 11:50 AM	Navigating the transition from paediatric to adult care of IEMs	<b>Speaker :</b> Dr. Mamta Muranjan	<b>Chairpersons :</b> Dr. Rajiv Bansal Dr. Pankaj Agrawal Dr. Rajkumar Goyal Dr. Vivek Gupta
11:50 AM - 12:20 PM	Role of Artificial Intelligence & telemedicine tools to improve patient care	<b>Speaker :</b> Dr. Seema Thakur	<b>Chairpersons :</b> Dr. Madhu Mathur Dr. Alok Tyagi Dr. Rajiv Jain Dr. Ambika Gupta
12:20 PM - 12:50 PM	Neuroimaging for IEMs	<b>Speaker :</b> Dr. Manish Parakh	<b>Chairpersons :</b> Dr. Sharad Sharma Dr. Lt Col Sachendra Badal Dr. Anshita Arora Dr. Abhishek Pandey
12:50 PM - 01:20 PM	Debate - Can Genomics replace NBS? <b>Moderator :</b> Dr. Alpa Dherai	<b>Panelist :</b> Dr. Meenal Agarwal Dr. Bhawana Agarwal Dr. Ranjana Mishra Dr. Sangeeta Khatta	<b>Judges :</b> Dr. Ratna Dua Puri Dr. Jayesh Seth Dr. J.P.Soni

**01:20 PM - 02:00 PM** Lunch

### Afternoon Session - Mixed Bag

02:00 PM - 02:30 PM	Role of Advocacy group & Patients Perspectives	<b>Moderator :</b> Dr. Dipanjana Datta	<b>Panelist :</b> Swartrenda Singha Representatives of advocacy group (RDIF, ORDI and MERD INDIA)
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02:30 PM - 03:00 PM	Panel Discussion - Quality control on laboratory testing	<b>Moderator :</b> Dr. Amit Gupta	<b>Panelist :</b> Dr. Rohit Cariappa Dr. Jyotsna Verma Dr. Prajnya Rangnath Dr. Aruna Gowdra
03:00 PM - 03:30 PM	Management of Alkaptonuria	<b>Speaker :</b> Dr. Sumita Danda	<b>Chairpersons :</b> Dr. Satyen Hemrajani Dr. Sunil Mahawar Dr. Virendra Mittal Dr. Mohit Poddar
03:30 PM - 04:00 PM	Porphyrias - Diagnosis & Management	<b>Speaker :</b> Dr. Anil Jalan	<b>Chairpersons :</b> Dr. Vaibhav Mathur Dr. Sawai Singh Lora Dr. Chandraprakash Gupta Dr. Sakshi Rajoria
04:00 PM - 04:30 PM	Quiz		<b>Dr. Suvarna Magar and Dr. Manisha Goyal</b>
04:30 PM - 05:00 PM	Vote of Thanks Prize Distribution & Valedictory	<b>Dr. Priyanshu Mathur Dr. Lokesh Kumar Agrawal</b>	



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