



Wadia Hospitals



## BAI JERBAI WADIA HOSPITAL FOR CHILDREN

ORGANISES  
1ST NATIONAL CONFERENCE  
ON **RARE DISEASES**  
IN CHILDREN



BAI JERBAI WADIA HOSPITAL FOR CHILDREN  
1st National Conference  
on Rare Diseases in  
Children



MUMBAI

1st and 2nd  
March 2025

# Cure & Care for the Rare!

Raising Awareness,  
Advancing Solutions



[www.wadiahospitals.org](http://www.wadiahospitals.org)



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# BAI JERBAI WADIA HOSPITAL FOR CHILDREN



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## 1ST NATIONAL CONFERENCE ON RARE DISEASES IN CHILDREN

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BAI JERBAI WADIA HOSPITAL FOR CHILDREN  
1st National Conference  
on Rare Diseases in  
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MUMBAI



1st and 2nd  
March 2025



## SCIENTIFIC PROGRAM



## SCIENTIFIC PROGRAM - 1st MARCH 2025

Time	Topic	Faculty	Chairperson
0730-0900hrs	REGISTRATION AND BREAKFAST		
0900-1300hrs	<b>WORKSHOPS</b> <b>UNBOXING RARE DISEASE - KISSA GENES KA</b> Optimizing role of genetics in care of rare diseases <b>DASTANE DISORDERS</b> Case based clinical approach to common 'rare' disorders <b>BACHH KE REHNA RE 'BABA'</b> Role of fetal medicine in early detection and prevention <b>KAR HAR MAIDAN FATEH</b> Roadmap to pediatric renal and liver transplant <b>HUM SAATH SAATH HAIN</b> Parent Support Group Session. Supported by ORDİ		
1100-1130hrs	<b>Opening Ceremony - Master of ceremony – Dr. Sumitra Venkatesh</b> <b>Guest of Honor, Special Guests, Dr Minnie Bodhanwala - CEO Wadia Hospitals</b>		
1130-1300hrs	<b>Workshop Continues</b>		
1300-1345hrs	<b>Poster Inauguration by Dr. Smita Mahale</b> Judge: Dr. Smita Mahale, Dr Jaya Vyas, Dr. Girisha Katta, Dr. Bharat Agarwal, Dr. Geetanjali Sachdeva, Dr. Vandana Bansal, Dr. Pradnya Bendre, Dr. B S Charan <b>Followed by LUNCH</b>		

## SESSION I

Time	Topic	Faculty	Chairperson
1345-1405hrs	Rare Metabolic & Genetic Disorders from A to Z	Dr. Rashid Merchant	Dr. Bharat Agarwal Dr. Pradnya Bendre
1405-1425hrs	Rare Disease in Children - Defining and measuring the unmet needs (Zoom Interaction)	Dr Swasticharan L	Dr. Nehal Shah Dr. Praveen Gokhale Dr. B S Charan

## SESSION II

Time	Topic	Faculty	Chairperson
1425-1510hrs	<b>Mystery diagnosis – Clinical Conundrum</b> <b>Encephalopathy</b> Moderator - Dr. Vrushab Gawali Panelists - Dr. Vrajesh Udani, Dr. Girisha Katta, Dr. Shilpa Kulkarni <b>Rickets, Growth and Short stature</b> Moderator - Dr. Rajesh Joshi Panelists - Dr. Alpana Ohri, Dr. Sudha Rao, Dr. Sangeeta Mudaliar <b>Immuno-hematological disorder</b> Moderators- Dr. Akshaya Chougule, Dr. Ritika Khurana Panelists - Dr. Vijaya Gowri, Dr. Purva Kanvinde, Dr. Umair Bargir		

Dr. Bela Verma  
Dr. Prema Vartakavi  
Dr. Mangai Sinha





Dr. S.M.Merchant Auditorium, Bai Jerbai Wadia Hospital for Children, Mumbai

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## SCIENTIFIC PROGRAM - 1st MARCH 2025

Time	Topic	Faculty	Chairperson
1510-1530hrs	Mystery diagnosis - look in the right place	Dr. Uma Ali	Dr. Archana Kher Dr. Bela Verma
1530-1550hrs	Inborn Errors of Immunity	Dr. Mukesh Desai	Dr. Mahesh Balsekar Dr. Manisha Madkaikar
1550-1610hrs	TEA BREAK		
SESSION III			
Time	Topic	Faculty	Chairperson
1610-1630hrs	Melodrama of mitochondria -Case based discussion	Dr. Maya Thomas	Dr. Fazal Nabi Dr. Chhaya Valvi
SESSION IV -Panel discussion			
Time	Topic	Faculty	Chairperson
1630-1715hrs	Prevention is better than cure- Fetal perspective Moderators- Dr. Purnima Satoskar, Dr. Vandana Bansal Panelists: Dr. Suma Sundararaju, Dr. Seema Thakur, Dr. Pradnya Bendre, Dr. Trupti Nadkarni, Dr. Rachna Dalmiya, Dr. Sumitra Venkatesh		Dr. Vaidehi Dande Dr. Jeetendra Gavhane
1715hrs onwards	POSTER VIEWING		
1730-1800hrs	Felicitation Ceremony - Master of ceremony – Dr. Sumitra Venkatesh		
1900-2300hrs	CULTURAL EVENING AND BANQUET VENUE: US CLUB, COLABA		





Dr. S.M.Merchant Auditorium, Bai Jerbai Wadia Hospital for Children, Mumbai

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## SCIENTIFIC PROGRAM - 2nd MARCH 2025

Time	Topic	Faculty	Chairperson
720-800hrs	BREAKFAST & REGISTRATION		
730-0820hrs	Free Paper - Parallel Sessions Hall I - Clinical Seminar Hall (2nd Floor)		Dr. Shilpa Kulkarni Dr. Lekha Parikh
	Hall II - Library Seminar Hall		Dr. Sangeeta Mudaliar Dr. Sonal Rakshpaul
SESSION I			
Time	Topic	Faculty	Chairperson
820-850hrs	Care for the Rare- Working together for a sustainable future	Dr. Madhulika Kabra	Dr. Rajalakshmi Nair Dr. Varsha Phadke
850-910hrs	Acute presentation of rare diseases- Early diagnosis in transforming outcomes	Dr. Sankar V H	Dr. Ramaprasad V Dr. Rekha Solomon
SESSION II - Panel Discussion- Case based approach to diagnosis			
Time	Topic	Faculty	Chairperson
910-1005hrs	Storage disorder Moderator - Dr. Ami Shah Experts - Dr. Ashwin Dalal, Dr. Jayesh Seth, Dr. Madhulika Kabra, Dr Manjori Mitra		Dr. Sanjeevini Masavkar Dr. Shakuntala Prabhu Dr. Parmanand Andankar Dr. Vivek Bhat
	Cardiomyopathy Moderator - Dr. Shreepal Jain Experts- Dr. Ashwin Dalal, Dr. Snehal Kulkarni, Dr. Jayesh Seth		
	Refractory acidosis Moderator - Dr. Lakshmi Shobavat Experts- Dr. Madhulika Kabra, Dr. Rekha Solomon, Dr. Jayesh Seth, Dr. Alpana Ohri		
	Grasp Or Gasp Moderator - Dr. Parmarth Chandane Experts - Dr. Seema Kapoor, Dr. Manisha Juvekar, Dr. Sagar Warankar, Dr. Parag Karkera, Dr. Balagopal Kurup		
1005-1025hrs	Key note address by Dr. B S Charan The Policy Landscape on Rare Diseases		Dr. Sudha Rao Dr. Pradnya Bendre
1025-1035hrs	TEA BREAK		
SESSION III - Panel Discussion			
Time	Topic	Faculty	Chairperson
1035-1105hrs	Access to rare disease expertise - Move towards establishing disease specific centre Moderators - Dr. Prashant Hiwarkar, Dr. Sudha Rao Experts- Dr. B.S. Charan, Dr. N. Ambadekar, Dr. Seema Kapoor, Dr. Madhulika Kabra, Mr. Prasanna Shirol, Dr. Rajalakshmi Nair		Dr. Dhruv Mamtora Dr. Pradnya Bendre
SESSION IV			
Time	Topic	Faculty	Chairperson
1105-1200hrs	Oral Paper presentation Judge: Dr. Manisha Madkaikar, Dr. Archana Kher, Dr. Madhulika Kabra, Dr. Shakuntala Prabhu, Dr. Sandesh Parelkar		
1200-1230hrs	Role of Genetics in diagnosis - Do we always get an answer	Dr. Ashwin Dalal	Dr. Sudhir Sane Dr. C T Deshmukh
1230-1300hrs	Rare disease screening protocols in India	Dr. Seema Kapoor	Dr. Harvinder Palaha Dr. Simmi Ratan







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**SCIENTIFIC PROGRAM - 2nd MARCH 2025****SESSION V - Panel Discussion- Case based approach to diagnosis**

Time	Topic	Faculty	Chairperson
1300-1330hrs	<b>Rare anemia</b> Moderator- Dr. Sangeeta Mudaliar Experts - Dr. Archana Swami, Dr. Bharat Agarwal, Dr. Seema Kapoor		Dr. Bhavna Dhingra Dr. Rajesh Joshi
	<b>Skeletal dysplasia</b> Moderator - Dr. Kshitija Patil Experts - Dr. Girisha Katta, Dr. Anju Seth, Dr. Archana Kher, Dr Sandeep Vaidya		

1330-1420hrs **LUNCH - MEET AND GREET****SESSION VI - Defying odds - new age management options**

Time	Topic	Faculty	Chairperson
1420-1440hrs	<b>Genetics and cancer therapeutics</b>	Dr. Soumya Srivastava	Dr. Sangeeta Mudaliar Dr. Archana Swami
1440-1500hrs	<b>Precision medicine in Neurology</b>	Dr. Anaita Hegde	Dr. Uma Ali Dr. Surabhi Rathi Dr. Shilpa Kulkarni

**SESSION VII - Debate**

Time	Topic	Faculty	Chairperson
1500-1530hrs	<b>Therapy of Spinal muscular Atrophy (SMA)-</b> Is a step forward --Dr. Anne Agnes Mathew Is a step too far - Dr. Neelu Desai		Dr. Upendra Kinjawadekar Dr. Vijay Yewale

**SESSION VIII - Does transplant provide cure - Case based discussion**

Time	Topic	Faculty	Chairperson
1530-1610hrs	<b>Immunodeficiency and Bone marrow transplant</b> Moderators: Dr. Ambreen Pandrowala, Dr. Vaishnavi Iyengar Experts - Dr. Mukesh Desai, Dr. Prashant Hiwarkar		Dr. Navin Khattry Dr. Bharat Agarwal
1610-1650hrs	<b>Hepatic conditions and Liver transplant</b> Moderators - Dr. Shivangi Tatarbe, Dr. Arka Banerjee Experts - Dr. Girish Gupte, Dr. Vikram Raut, Dr. Pradnya Bendre, TBA		Dr. Sandesh Parelkar Dr. Paras Kothari

**SESSION IX**

Time	Topic	Faculty	Moderator
1650-1720hrs	<b>Priorities and perspectives of Cure and Care of Children with Rare Diseases - Vision at Bai Jerbai Wadia Hospital for Children</b> Dr. Sudha Rao, Dr. Pradnya Bendre, Dr. Shilpa Kulkarni, Dr. Sangeeta Mudaliar, Dr. Ami Shah, Dr. Purnima Satoskar, Dr. Dhruv Mamtara		Dr. Minal Shah Dr. Suma Sundararaju Dr. Mrudula Khedekar

**SESSION X**

Time	Topic	Faculty	Chairperson
1720-1740hrs	<b>QUIZ</b> Dr. Gayatri Munghate, Dr. Samantha Kumar, Dr. Deepti Malhotra, Dr. Saraswati Kini		
1740-1750hrs	<b>VALEDICTORY</b>		
1750hrs onwards	<b>HIGH TEA</b>		



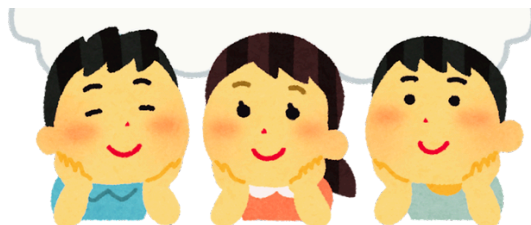


## WORKSHOP I - 1st MARCH 2025

## UNBOXING RARE DISEASE - KISSA GENES KA

Optimising role of genetics in care of rare diseases

Time	Topic	Faculty
0800-0900hrs	REGISTRATION AND BREAKFAST	
SESSION I		
900-915hrs	Past, present, future of cytogenetics	Dr. Prochi Madon
915-930hrs	Fundamentals and Basics of Molecular genetics including NGS	Dr. Girisha Katta
930-1030hrs	WORK STATIONS <i>Delegates are divided into two groups and exchange after 30 minutes</i>	
Station I	Workstation on cytogenetics	Dr. Ami Shah Dr. Kshitija Patil
Station II	Workstation on molecular genetics	Dr. Jaya Vyas Mr. Lalit Samant Ms. Shweta Sakpal
1030-1100hrs	TEA BREAK	
SESSION II		
Time	Topic	Faculty
1100-1120hrs	Insights into biochemical and metabolic conditions	Dr. Alpa Dherai
1120-1140hrs	Introduction to bioinformatics (focus on VUS)	Dr. Ramaprasad V
1140-1240hrs	WORK STATIONS <i>Delegates are divided into two groups and exchange after 30 minutes</i>	
Station I	Workstation on biochemical and metabolic conditions	Dr. Ami Shah Dr. Kshitija Patil
Station II	Workstation on bioinformatics	Dr. Jaya Vyas Mr. Lalit Samant Ms. Shweta Sakpal
SESSION III		
Time	Topic	Faculty
1240-1300hrs	Genetic counselling - a bridge and bonding with rare disease families	Dr. Hema Purandare
1300-1310hrs	VALEDICTORY	





## WORKSHOP II - 1st MARCH 2025

**DASTANE DISORDERS**

Case based clinical approach to common 'rare' disorders

Time	Topic	Faculty
0800-0900hrs	REGISTRATION AND BREAKFAST	
SESSION I - APPROACH TO CLINICAL CASE SCENARIOS		
Time	Topic	Faculty
900-930hrs	Tiny brain and tricky challenges	Dr. Shilpa Kulkarni Dr. Payal Shah
930-1000hrs	Cytopenias and Bleeding disorders	Dr. Sangeeta Mudaliar Dr. Purva Kanvinde Dr. Ritika Khurana
1000-1030hrs	Tubular disorders and Glomerulopathy	Dr. Alpana Ohri Dr. Amish Udani
1030-1100hrs	Metabolic emergencies	Dr. Rekha Solomon Dr. Lakshmi Shobavat Dr. Isha Bhagat
1100-1115hrs	TEA BREAK	
SESSION II - Unraveling the Enigma: Investigative Approaches to Rare Diseases		
Time	Topic	Faculty
1115-1130hrs	Decoding Skeletal Dysplasia : case-based approach	Dr. Girisha Katta
1130-1215hrs	Diagnostic Odyssey	Dr. Foram Gala Dr. Jayesh Seth Dr. Girisha Katta
SESSION III - THERAPEUTIC CHALLENGES AND DILEMMAS		
Time	Topic	Faculty
1215-1230hrs	Dietetic Arithmetics of Rare Disorders	Dr. Sheetal Mhamunkar
1230-1245hrs	Common remedies of uncommon disorders	Dr. Shilpa Kulkarni
1245-1300hrs	Beyond the Norm: Targeting Rare Disease Therapy	Dr. Alpana Ohri
1300-1310hrs	VALEDICTORY	







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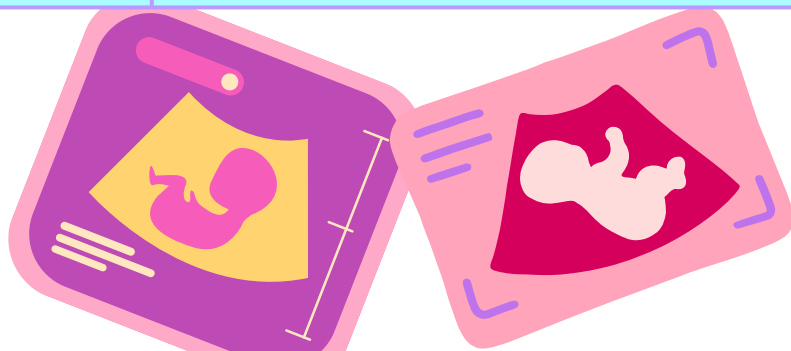
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## WORKSHOP III - 1st MARCH 2025

**BACHH KE REHNA RE 'BABA'**

Role of fetal medicine in early detection and prevention

Time	Topic	Faculty
0800-0900hrs	REGISTRATION AND BREAKFAST	
SESSION I		
Time	Topic	Faculty
900-920hrs	Red flags in the first trimester for genetic abnormalities	Dr. Manisha Beck
920-940hrs	Tell-tale ultrasonographic signs of genetic syndromes in the 2nd and third trimester	Dr. Rajas Chaubal
940-1000hrs	Screening for aneuploidies: which method is best	Dr. Chinmayee Ratha
1000-1020hrs	Hemoglobinopathies: how not to miss a case	Dr. Veronica Arora
1020-1030hrs	Q & A	
1030-1040hrs	TEA BREAK	
SESSION II		
Time	Topic	Faculty
1040-1100hrs	Which genetic test when: ask the expert-case scenarios	Dr. Ami Shah Dr. Purnima Satoskar
1100-1120hrs	Genetics of neurological disorders	Dr. Shruti Bajaj
1120-1140hrs	Cardiac anomalies: detection, counselling and management	Dr. Shreepal Jain
1140-1200hrs	Cranio-Facial Dysmorphism and its implications	Dr. Chander Lulla
SESSION III		
Time	Topic	Faculty
1200-1245hrs	Panel discussion -Phenotype to genotype and its management Moderator: Dr. Vandana Bansal Panelists: Dr. Chinmayee Ratha, Dr. Manisha Beck, Dr. Veronica Arora, Dr. Geetha Balsarkar, Dr. Praful Shanbhag, Dr. Forum Shah, Dr. Kshitij Sheth, Dr. Shilpa Agrawal, Dr. Pooja Bandekar, Dr. Payal Lakhani	
1245-1300hrs	Fetal Autopsy : A window of opportunity	Dr. Seema Thakur
1300-1310hrs	VALEDICTORY	





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## WORKSHOP IV - 1st MARCH 2025

## KAR HAR MAIDAN FATEH

Roadmap to pediatric renal and liver transplant - Co-ordinator: Dr Aditi Rangnekar

Time	Topic	Faculty
0800-0900hrs	REGISTRATION AND BREAKFAST	
SESSION I		
Time	Topic	Faculty
900-910hrs	Introduction and Overview	Dr. Gayatri Munghate
910-930hrs	Basics of Transplant medicine	Dr. Pradnya Bendre Dr. Uma Ali
SESSION II - RENAL TRANSPLANT		
930-945hrs	Indications and role of renal transplantation in rare diseases	Dr. Parag Karkera Dr. Uma Ali
945-1000hrs	Preparing the prospective kidney transplant recipient	Dr. Uma Ali Dr. Chintan Shah
1000-1010hrs	Donor work-up	Dr. Rishab Jain Dr. Chintan Shah
1010-1030hrs	Imaging of donor and recipient	Dr. Saroj Chavan Dr. Parul Garde
1030-1100hrs	Surgical overview (Living and deceased donor transplants)	Dr. H R Pathak Dr. Pradeep Vyvhare
1100-1115hrs	TEA BREAK	
SESSION II - LIVER TRANSPLANT		
Time	Topic	Faculty
1115-1130hrs	Indications and role of liver transplantation in rare diseases	Dr. Ira Shah Dr. Vikram Raut
1130-1150hrs	Preparing the prospective liver transplant recipient	Dr. Shivangi Tatarbe Dr. Vishruta Karkera
1150-1200hrs	Donor work -up	Dr. Arka Banerjee Dr. Gayatri Munghate
1200-1220hrs	Imaging of donor and recipient	Dr. Parul Garde Dr. Saroj Chavan
1220-1300hrs	Surgical overview (Living and deceased donor transplants)	Dr. Vikram Raut Dr. Taher Ahmed
1300-1310hrs	VALEDICTORY	

1310 to 1400hrs  
Hands on opportunities available post  
workshop for interested surgical specialists.

Faculty:  
Dr. Vikram Raut, Dr. B.R. Panda, Dr. P.S.  
Bendre, Dr. Parag Karkera, Dr. Gayatri  
Munghate, Dr. Abhishek Kshirsagar





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## WORKSHOP V - 1st MARCH 2025

## HUM HUM SAATH SAATH HAI

Parent Support Group Session - Play Therapy Park, Bai Jerbai Wadia Hospital For Children

Time	Topic	Faculty
0800-0900hrs	REGISTRATION AND BREAKFAST	
SESSION I		
Time	Topic	Faculty
900-910hrs	Introductory Ice Breaker Game	Dr. Veronique Dinand Ms. Anuradha Karegar Ms. Joanna Pinto Mr. Keith D'Souza
910-915hrs	Setting the tone for the Workshop	
915-920hrs	Performance 1	
920-1000hrs	Small Group Art Based activity to express feelings	
1000-1005hrs	Performance 2	
1000-1100hrs	Sharing of Journey with Parents and Children on the rare diseases	
1100-1115hrs	TEA BREAK	
SESSION II		
Time	Topic	Faculty
1115-1145hrs	Talk on Rare Diseases	Mr. Prasanna Shirol
1145-1215hrs	Q&A session for audience	Dr. Veronique Dinand Ms. Anuradha Karegar Ms. Joanna Pinto Mr. Keith D'Souza
1215-1220hrs	Performance 3	
1220-1235hrs	Witnessing and showcasing talents of children with rare diseases	
1235-1300hrs	Interactive Sessions	
1300-1310hrs	VALEDICTORY	

