RARE DISEASES INDIA FOUNDATION

NATIONAL SYMPOSIUM ON GENETIC DISEASES

# 5th NATIONAL SYMPOSIUM ON

# GENETIC DISEASES

**Theme: The Journey Beyond Diagnosis**

# NSGDCON 2026, New Delhi

**24th-25th Oct, 2026, Delhi-NCR**

**Venue: Ashoka Convention Hall, Manekshaw Centre Auditorium, Delhi Cantonment, New Delhi**

# Scientific Program



CONFERENCE PROGRAMME

# 5th National Symposium on Genetic Diseases

## Theme: The Patient Journey

Integrating Clinical Care and Lived Experience

"The Journey Beyond Diagnosis — From Discovery to Lifelong Care"

**Ashoka Hall, Manekshaw Auditorium, New Delhi · 24–25 October 2026**

Keynote · Clinical session · Panel · Debate · Talk · Quiz · Sponsored session · Inauguration / Oration · Break

DAY 1 · 24 OCTOBER 2026

## The Journey Beyond Diagnosis

The journey begins

**08:15–08:45** · 30 min
**Registration and coffee**

**08:45–08:50** CEREMONY · 5 min
**Welcome and opening remarks**
Theme introduction: "The Journey Beyond Diagnosis — From Discovery to Lifelong Care"

**08:50–09:30** TALK · 40 min · incl. 5 min Q&A
**Abstract presentations**
Selected abstracts — **Dr Annie Thomas, Dr Niral Desai, Dr Umang Arora, Dr Rajesh Bhatulla, Dr Vanshikha Arora, Dr Nadir**

*Judges: Dr Sumita Danda · Dr Kanika Singh · Moderator: Mridula Menon*

**09:30–09:45** TALK · 15 min · incl. 5 min Q&A
**AI to Help Reduce Delays and Bottlenecks in Care Pathways**
**Dr Milan Choksey**

**09:50–10:20** CLINICAL SESSION · 30 min · incl. 5 min Q&A
**Impact of Genetic Diagnosis**
What does the diagnosis really give to a family? — **Dr Sheetal Sharda**
Eye disorders in genetics: what does diagnosis change? — **Dr Shailaja Tibrewal**

*Chairs: Dr Neerja Gupta, Dr Seema Thakur, Dr Seema Kapoor*

**10:20–10:50** CLINICAL SESSION · 30 min · incl. 5 min Q&A
**Changing Management After Diagnosis**
Living with alkaptonuria: nitisinone, surveillance and long-term care — **Prof Sumita Danda**
Changing the course of atypical HUS — **Dr Aditi Sinha**

*Chairs: Lt Col Puneet Singh, Dr Bhawna Dhingra*

**10:50–11:10** KEYNOTE · 20 min · incl. 5 min Q&A
**Keynote 2: Beyond Bleeds — Transforming the Haemophilia Journey**
**Dr Shubha Phadke**

*Chairs: Dr Sumita Danda, Dr Ratna Devi*

**11:10–11:55** CLINICAL SESSION · 45 min · incl. 5 min Q&A
**One Diagnosis, Many Lives**
From RAS-MAPK to real life: translating diagnosis into real life — **Dr Inusha Panigrahi**
Stopping oxalate at its source: the new era of hyperoxaluria treatment — **Dr Sumita Danda**
The adult NF1: tumours, transformation and targeted treatment, including discussion on transition of care from paediatric to adult services — **Dr Shankar VH**

*Chairs: Dr Rohit Gupta, Dr Shubha Phadke, Dr Ankita Srivastava · Moderator: Dr Pooja Motwani*

**11:55–12:25** CLINICAL SESSION · 30 min · incl. 5 min Q&A
**The Geneticist as a Longitudinal Physician**
22q11.2 microdeletion syndrome: journey and jeopardies (prenatal perspective) — **Dr Seema Thakur**
Metabolomics: from pathways to therapeutics — **Dr Chaitanya Datar**

*Chairs: Dr Vivek, Dr Sunita Bijarnia Mahay, Dr Deepti Saxena, Dr (Col) Gunjan Rai*

**12:25–12:40** TALK · 15 min
**Casting the Podcaster — Dr Nachiket Bhatia**
By **Dr Veronica Arora**

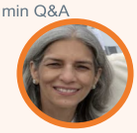
*Cases are anonymised and consent obtained; images and genomic data are protected; discussions are educational and not individual medical advice.*

**12:40–13:00** CEREMONY · 20 min

### Inauguration by Chief Guest and Lamp Lighting

**13:45–14:30** CEREMONY · 45 min · incl. 5 min Q&A

### IC Verma Oration: "Charting the Changing Landscape of Genetics"



**Prof Ratna Dua Puri**

*Chairs: Brig Ranjit Ghuliani, Prof Narinder Mehra, Dr BS Charan, Dr Jagdish Chandra*

**15:45–17:00** CLINICAL SESSION · 1 h 15 min · incl. 5 min Q&A

### The LSD Journey Beyond Diagnosis, From Survival to Quality of Life

Defining meaningful outcomes in LSDs — **Dr Ashok Vellodi**

Optimising treatment outcome in LSDs in resource-limited settings — **Dr Madhulika Kabra**

Case-based patient journeys

1. Sponsored session: TAKEDA — Navigating the Gaucher disease journey — **Dr Mamta Muranjan**
2. Niemann-Pick disease: beyond diagnosis to emerging horizons — **Dr Neerja Gupta**
3. Debate: ERT in MPS I and II — transforming outcomes or falling short of expectations?

Moderator: **Dr Neerja Gupta · Participants: Dr Shailesh Pandey, Dr Manisha Goyal, Dr Bhawna Dhingra, Dr Sangeeta Khatter**

*Chairs: Prof Ratna Dua Puri, Dr Madhulika Kabra*

**17:30–18:00** CLINICAL SESSION · 30 min · incl. 5 min Q&A

### Neuromuscular & Cognitive Genetics

SMA after diagnosis: from treatment initiation to lifelong care — **Prof Sheffali Gulati**

Recognising treatable genetic and metabolic conditions presenting with autism — **Dr Praveen Suman**

*Chairs: Dr Madhulika Kabra, Lt Col Abhishek Pandey · Moderators: Dr Sayli, Dr Niladri*

**19:00–19:30** PANEL · 30 min · incl. 5 min Q&A

### Optimising Pathways to Clinical Trials in Rare Disorders in India

Moderator: **Dr Hemlata Wadhwani Bhatia**

Experts: **Dr Shubha Phadke, Dr Inusha Panigrahi**

Panelists: **Dr Manjeet Mehta, Dr Shailesh Pandey, Dr Saswati Mukhopadhyay, Dr Dipanjana Datta, Dr Nitika**

**20:00–20:10** SPONSORED SESSION · 10 min

### Information Box

NPRD in 2026: The evolving landscape of treatment and access — **Dr Mayuri Yeole (10 min talk + 5 min discussion)**

*Chairs: Dr Anita Rajhoria, Dr Rohit Gupta, Dr Ashutosh Gupta, Dr Vikrant Sood*

**13:15–13:45** · 30 min

### Lunch

**14:30–15:45** CLINICAL SESSION · 1 h 15 min · incl. 5 min Q&A

### Metabolic Corner

A race against time: beyond diagnosis in ALD — **Dr Sunita Bijarnia Mahay**

Wilson's disease: can we cure it? — **Dr Binukumar BK**

Clinical pearls on managing IEMs — **Moderators: Dr Sunita Bijarnia Mahay, Dr Sayli Bidkar · Panelists: Dr Chaitanya Datar, Dr Pankaj Garg, Dr Divya, Anuja, Shipra**

*Chairs: Dr Shankar VH, Dr Neerja Gupta, Dr Dinesh Goel, Dr Nimmi Kansal, Dr Seema Kapoor*

**17:00–17:15** KEYNOTE · 15 min

### From Diagnosis to Disease Pathways — Rethinking Continuity

**Dr Seema Kapoor**

*Chairs: Dr Madhulika Kabra, Dr Ashutosh Gupta, Dr Mamtha Gowda*

**17:15–17:30** · 15 min

### Tea

**18:00–19:00** CLINICAL SESSION · 1 h · incl. 5 min Q&A

### Diagnosis Beyond Diagnosis

Mobile Elements: Jumping Genes and the Diseases They Leave Behind — **Dr Neha Garg**

Transferrin Isoforms: Reading the Sugar Code Behind the Diagnosis — **Dr Alpa J Dherai**

Beyond the Blood Count: Intricacies of Thalassaemia Diagnosis — **Dr Jyoti Kotwal**

*Chairs: Dr Nimmi Kansal, Dr (Col) Samir, Dr Samarth Kulshrestha, Dr Deepti Gupta, Col Shivani Sangwan*

**19:30–20:00** PANEL · 30 min · incl. 5 min Q&A

### The Genetic Counsellor's Perspective

Role of genetic counsellors in breaking bad news, communicating uncertainty and avoiding over-interpretation — Mridula Menon, Deepti Chowdhary, Nirali, Dr Mamtha Gowda

*Chairs: Dr Gayatri, Dr Manisha Goyal, Dr Hemlata Wadhwani Bhatia · Moderators: Dr Sheeba Farooqui, Dr Vishakha Agarwal · Lead: Neeraja Reddy (Genetic Counsellor)*

**20:10–22:10** · 2 h

### Gala Dinner

DAY 2 · 25 OCTOBER 2026

# Rising to Challenges

**08:00–09:05** CLINICAL SESSION · 1 h 5 min · incl. 5 min Q&A

### Case Series Presentations — Longitudinal Data That Will Surprise You

**Dr Aman, Dr Mahrukh, Dr Ami Shah, Dr Haseena, Dr Dipanjana Datta**

*Chairs: Dr Rekha Gupta, Dr Aradhana Dwivedi, Neeraja Reddy (Genetic Counsellor) · Moderator: Dr Sarah*

**09:05–09:25** DEBATE · 20 min · incl. 5 min Q&A

### From Diagnosis to Action

When treatment exists, should genetic testing come before symptoms? — **Dr Divya Agarwal, Dr Sameer Bhatia, Dr Kshitija, Dr Anup Rawool**

*Moderators: Dr Gayatri, Dr Parminder Kaur*

**09:25–10:20** TALK · 55 min · incl. 5 min Q&A

### The Case That Changed Me: four clinical stories

Turning points, lessons and changes in practice — **Dr Ravneet, Dr Chakshu, Dr Parminder, Dr Anup Rawool**

Genomics from a fetal perspective — **Dr Sumitra Bachani**

Chairs for this talk: Dr Shailesh Pandey, Dr Seema Thakur, Dr Divya Agarwal, Dr Mala Srivastava, Dr Reema Bhat, Dr Mamtha Gowda

*Chairs: Dr Manjeet Mehta, Dr Mahrukh Hameed, Dr Priyanshu*

**10:20–10:40** TALK · 20 min · incl. 5 min Q&A

### Impact of Genetic Testing in Epilepsies

**Dr Mayank Nilay**

### Cancer Update

**Dr Somya Srivastava**

*Experts: Surg Cdr Benjith Paul K, Dr Divya Agarwal, Dr Sameer Bhatia*

**10:40–11:00** DEBATE · 20 min · incl. 5 min Q&A

### Debate: The right to genetic treatment should be as fundamental as the right to genetic diagnosis

**Dr Ashka, Dr Meenakshi Lallar, Dr Kiruthika, Dr Daniel**

*Moderators: Dr Swasti, Dr Roopardarshini*

**11:00–11:15** TALK · 15 min · incl. 5 min Q&A

### Integrated Care Pathways for Rare Diseases

**Dr Ashok Vellodi**

*Chairs: Dr Sheela Nampoothiri, Dr Shankar VH, Dr Pradeep Sharma*

**11:15–11:55** PANEL · 40 min · incl. 5 min Q&A

### Beyond the Clinic — Building the Patient Journey

Themes: palliative care — it isn't always about the end of life; building a multidisciplinary service; from early intervention to inclusive education — an integrated system of care for children with neurodevelopmental disorders

Moderators: **Prof Ratna Dua Puri, Dr Ruparna**

Panelists: **Dr Archana Iyengar, Dr Anjan Bhattacharya, Megha Goenka, Dr Chakshu**

**11:55–12:30** PANEL · 35 min · incl. 5 min Q&A

### Panel: Inequity and Resilience

Panelists: **Prof Mamta Muranjan, Dr Rekha Gupta, Dr Shailesh Pandey, Dr Gagan, Dr Priyanshu Mathur**

*Moderators: Dr Madhulika Kabra, Dr Meenakshi Lallar*

**12:30–13:10** PANEL · 40 min · incl. 5 min Q&A

### The Uncomfortable Conversation

**Dr Sarah, Dr Sayli, Dr Suvarna**

*Moderator: Dr Shifalli Gupta · Chairs: Dr Manisha Goyal, Dr Ami Shah, Dr Pradeep Sharma*

**13:10–13:40** · 30 min

### Lunch



**13:40–14:00** SPONSORED SESSION · 20 min

### MPS II: A Quest for Newer Treatments

**Dr Priyanshu Mathur**

*Chairs: Dr Sangeeta Khatter, Dr Sumitra Bachani, Dr Seema Thakur, Dr Nimmi Kansal*

**14:00–14:45** DEBATE · 45 min · incl. 5 min Q&A

### Genetics Courtroom — You Be the Judge

Difficult clinical decisions presented as courtroom-style cases — **Dr Rhea, Dr Devi Saranya, Dr Somya Srivastava, Dr Pallavi Sinha**

*Judges: Dr Shubha Phadke, Dr Nimmi Kansal, Dr Megha Goenka · Moderator: Dr Shifalli Gupta*

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**14:45–15:05** KEYNOTE · 20 min · incl. 5 min Q&A

**Keynote 3: My Follow-Up: the story doesn't end with the report**

Surveillance, treatment, changing clinical needs, family counselling and long-term support

Fibulin-4 aortopathy: a single-centre experience of 21 years — **Dr Sheela Nampoothiri**

*Chairs: Dr Anjan Bhattacharya, Dr Ashutosh Gupta, Dr Inusha Panigrahi · Moderator: Dr Ambika Gupta*

**15:05–15:20** · 15 min

**Tea**

**15:20–15:40** KEYNOTE · 20 min · incl. 5 min Q&A

**Keynote 4: DMD — Management Opportunities: Very Near Yet Very Far**

**Dr Ann Agnes, Mr Praveen Singla**

*Chairs: Dr Shubha Phadke, Dr Shankar VH, Mrs Gayatri Chahal, Dr Ratna Devi · Moderator: Dr Devi Saranya*

**15:40–16:25** CLINICAL SESSION · 45 min · incl. 5 min Q&A

**Genetics Unfiltered — When the Gene Opens the Door to Discovery**

Therapeutic windows, disease-modifying treatment, monitoring and access — **Dr Pallavi, Dr Kshitija, Dr Ambika Gupta, Dr Mayuri**

*Chairs: Dr Divya Agarwal, Dr Evanka Madan, Dr Prashant Seth*

**16:25–16:55** TALK · 30 min · incl. 5 min Q&A

**What I Wish I Knew — Lessons from the Middle of the Journey**

**Dr Amit Gupta, Dr Gayatri, Dr Vibha Jain**

*Chairs: Dr Suvarna, Dr Megha Goenka, Dr Ashka, Dr Sarah · Moderator: Dr Daniel*

**16:55–17:35** TALK · 40 min · incl. 5 min Q&A

**The Next Generation — Four Voices from the Clinic**

Challenges, errors, future lessons — **Dr Janmeeta, Dr Swasti, Dr Vishakha Agarwal, Dr Pooja Motwani**

*Chairs: Dr Manjeet Mehta, Dr Inusha Panigrahi, Dr Prashant Seth*

**17:35–17:50** QUIZ · 15 min

**Quiz**

*Quiz masters: Dr Roopardarshini, Dr Sheeba Farooqui · Judges: Dr Ravneet, Dr Priyanshu, Dr Anjan Bhattacharya*

**17:50–18:05** CEREMONY · 15 min

**Valedictory Function**

**Lifelong care — the journey continues**

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## Conference Secretariat

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