Knowledge Partner

# THE 2nd RARE GENETIC DISEASES RESEARCH SUMMIT (REDRESS)

23rd - 24th November 2023

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# THE 2nd RARE GENETIC DISEASES RESEARCH SUMMIT (REDRESS - 2023)

*INAUGURAL SESSION*

**Dr. RAKESH MISHRA**

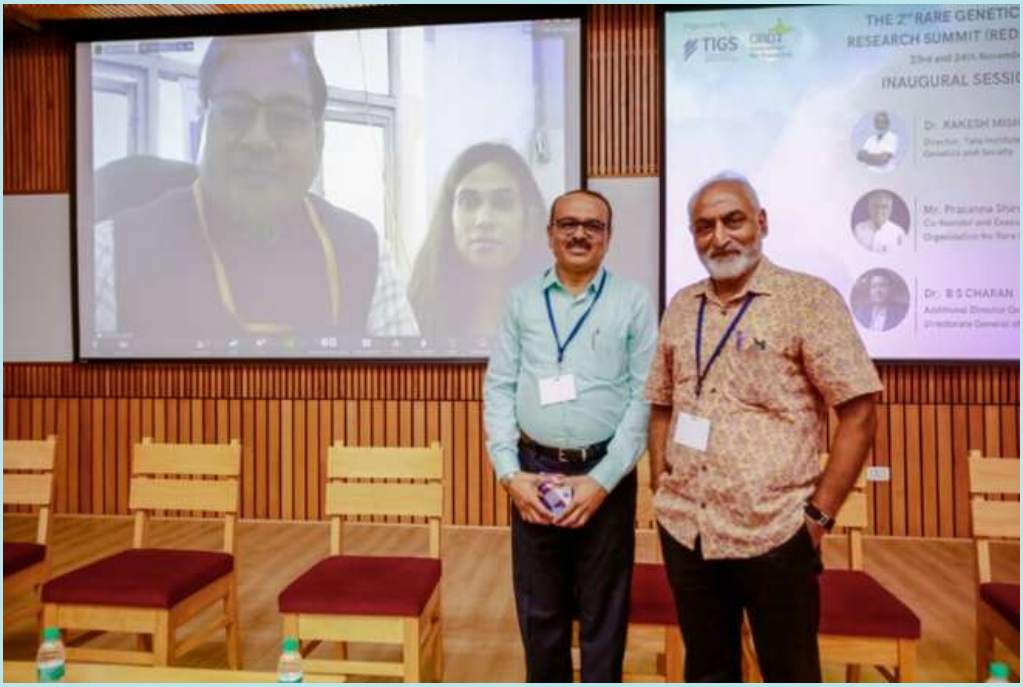
*Director, Tata Institute for Genetics and Society*

**Mr. PRASANNA SHIROL**

*Co-founder and Executive Director, Organization for Rare Diseases India*

**Dr. B S CHARAN**

*Additional Director General, Directorate General of Health Services, Government of India*




*From left: Dr. B.S Charan (online), Mr. Prasanna Shirol, Dr. Rakesh Mishra*

# *Rare Genetic Diseases Research Summit (REDRESS) 2023:* Unveiling the Power of Multi Stakeholder Engagement

In the complex realm of rare genetic diseases (RGDs), millions in India navigate a terrain of hardship and isolation. The Rare Genetic Diseases Research Summit (REDRESS) 2023 hosted by the Tata Institute for Genetics and Society (TIGS) and the Organization for Rare Diseases India (ORDI), along with the Indian Council for Medical Research (ICMR) as a knowledge partner, was our second national summit. This unique platform serves as a powerful catalyst for a multi-pronged approach in uniting stakeholders to accelerate diagnostic and therapeutic solutions for RGDs in India using cutting-edge science and technology.

Building upon the success of its inaugural year, REDRESS 2023, held on November 23rd and 24th, aimed to achieve a great impact. Over 50 diverse voices echo from the halls of government ministries to the labs of groundbreaking researchers resonated throughout the summit. Dignitaries, renowned clinicians, passionate patient advocates, and innovative industry pioneers joined forces to present the contemporary trends in their fields, brainstorm, exchange ideas and share perspectives to accelerate RGD research and care in a country as dense and diverse as India

The collective focus traversed the RGD spectrum, delving into the intricacies of basic research, cutting-edge MedTech advancements, and the complexities of running clinical trials and navigating regulatory policies. REDRESS 2023 spotlighted the **vital roles of diagnostics, policy research, and industry investment in propelling progress forward in developing cost-effective therapeutics.**

The opening session set the stage, with powerful voices leading the charge. Dr. Rakesh Mishra, Director of TIGS championed the vital need for a homegrown approach, emphasizing the importance of societal acceptance in tackling RGDs and the need to bring our collective resources and expertise to the table. Mr. Prasanna Shirol of ORDI, speaking from the heart, painted a poignant picture of the hope and challenges faced by RGD patients and their families. He highlighted how this hope is now moving towards a strong research focus, coupled with policy advocacy for acceptance. Dr. BS Charan, representing the government, acknowledged the struggles faced by the community while showcasing concrete steps, such as the National Policy for Rare Diseases and the expansion of Centres of Excellence, to address these challenges head-on. He highlighted several steps to improve access and bring these patients closer to available solutions.

However, REDRESS 2023 did not merely preach from a podium. ***The agenda vibrated with the pulse of collaboration***. Researchers exchanged crucial insights across various RGDs, forging bonds that promise to bridge research gaps and accelerate the development of life-changing diagnostics and treatments. The need for policy reforms, ensuring easier access to healthcare and research opportunities for patients, resonated throughout the halls, finding passionate advocates in patient groups and policymakers alike. Empowering patients to participate in research and decision-making processes was another key theme, recognizing their indispensable role in shaping the future of RGD research.

The impact of REDRESS transcends the confines of the two-day event. The discussions, collaborations, and action points set in motion the pathways to accelerate solutions in the RGD field. With each REDRESS summit, the hope flickers brighter, promising a future where rare diseases are no longer rare in terms of awareness, resources, and, most importantly, hope.

**"The need for policy reforms, ensuring easier access to healthcare and research opportunities for patients resonated throughout the halls, finding passionate advocates in patient groups and policymakers alike"**

# *SESSION 1*
# *RGD LANDSCAPE AND DIAGNOSTICS IN INDIA*

**SESSION CHAIR**

**Prof. B.K THELMA**

*University of Delhi*

**KEYNOTE SPEAKER**

**Dr. MEENAKSHI BHAT**

*Centre for Human Genetics, Bengaluru*

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**Dr. RATNA DUA PURI**

*Institute of Medical Genetics & Genomics, Sir Ganga Ram Hospital, New Delhi*

**Dr. SUBASREE RAMAKRISHNAN**

*National Institute of Mental Health and Neurosciences*

**Dr. ASHWIN DALAL**

*DBT- Centre for DNA Fingerprinting and Diagnostics, Hyderabad*

**Dr. AMLIN SHUKLA**

*National Registry of Rare and Other Inherited Disorders, ICMR, New Delhi*

**Dr. SWARKAR SHARMA**

*Centre for Molecular Biology, Central University of Jammu, J&K*

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## PATIENT ADVOCACY GROUPS

**Mr. VIKAS BHATIA**

*MERD India Foundation*

# *Rare Genetic Diseases Research Summit (REDRESS) 2023:*
# Session 1 : RGD Landscape and Diagnostics in India

The first REDRESS session, **"RGD Landscape and Diagnostics in India,"** chaired by Prof. B K Thelma of Delhi University, featured clinicians, researchers, ICMR officials, and patient advocates. Dr. Meenakshi Bhat from the Centre for Human Genetics (CHG) delivered the keynote. Speakers included experts like Dr. Ratna Dua Puri from Sir Ganga Ram Hospital in New Delhi, Dr. Ashwin Dalal from the Centre for DNA Fingerprinting and Diagnostics in Hyderabad, Dr. Swarkar Sharma from the Central University of Jammu, Dr. Subasree Ramakrishnan from NIMHANS in Bengaluru, and Dr. Amlin Shukla from the National Registry of Rare and Other Inherited Disorders-ICMR New Delhi, addressing RGD challenges. Mr. Vikas Bhatia, representing a patient advocacy group, shared a moving personal journey. This diverse session highlighted diagnostic advancements and patient-centric perspectives, showcasing India's progress in tackling rare genetic diseases.

***Shining a Light on Rare Diseases: Karnataka's Leadership and the Fight for Diagnosis, Treatment, and Support:***

Dr. Bhat emphasized that approx. 400 rare diseases prevalent and reported in India. While 3 million people in Karnataka are affected by rare diseases, she commended Karnataka state for being at the forefront, consistently funding rare disease therapy since 2016. The CHG evaluated 30,000 patients since 2006, maintaining a comprehensive data repository. Notably, 44 patients with LSDs (lysosomal storage disorders) received enzyme replacement therapy (ERT). She shared the challenging journey of a young Pompe disease patient, highlighting the struggles in obtaining an accurate diagnosis, treatment, and subsequent investigations. Dr. Bhat stressed the multi-systemic impact of rare diseases, placing a significant financial burden on families for supportive care. She drew attention to India's revised clinical trial rules by the Central Drugs Standard Control Organization (CDSCO) and underscored the pressing need for public awareness and a stronger societal support system for children grappling with rare diseases.

**"The introduction of dried blood spots and Next-Generation Sequencing (NGS) has transformed the screening and diagnosis of several established genetic syndromes as well as led to investigations around unexplained intellectual disabilities emphasizing the importance of diverse diagnostic strategies."**

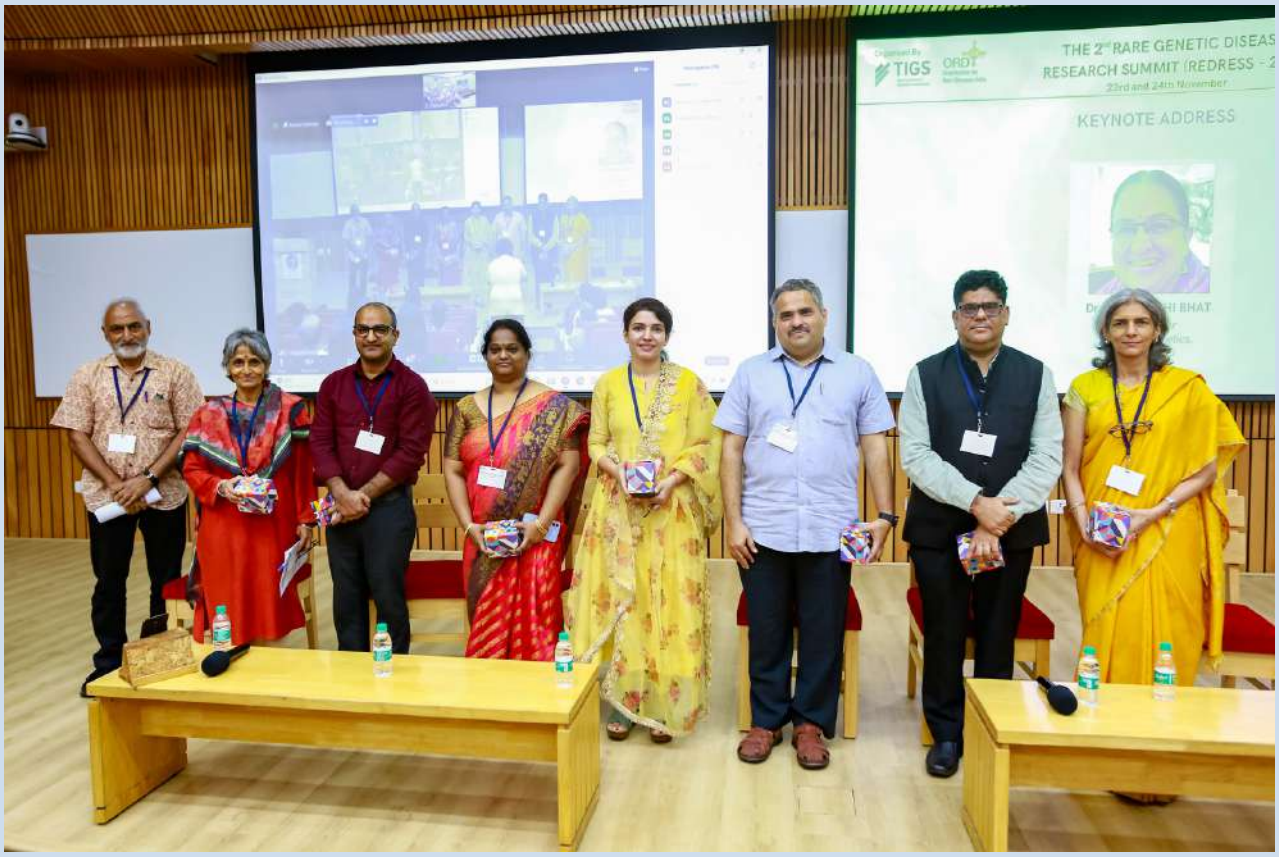
Dr. Ratna Dua Puri gave a retrospective overview of the RGD diagnostics of the last two decades, particularly focusing on Thalassemia and Hemoglobinopathies in India as among the first genetic conditions to be investigated widely. The early work involved characterizing disorders with traditional techniques like enzyme assays which itself was difficult to perform. However, the introduction of dried blood spots revolutionized enzymatic and molecular diagnoses in remote areas. Later, NGS (Next-Generation Sequencing) ushered into a transformative era that resolved cohort studies related to unexplained intellectual disabilities, which she demonstrated through compelling case studies. She also highlighted that variant of uncertain significance is the greatest challenge for definite diagnosis. The importance of using varying diagnostic strategies for identical disorders was also highlighted in her talk. The presentation acknowledged the ICMR-funded Indian Undiagnosed Disease Program (UDP), comprising four genetic centers aimed at refining diagnostic practices. She concluded by emphasizing not only the diagnostic odysseys for rare diseases but also the necessity for informed management and the application of transformative therapies in patient care.

***Addressing Rare Genetic Diseases in India: Insights and Initiatives by Dr. Ashwin Dalal:***
Dr. Ashwin Dalal highlighted India's substantial contribution to the RGD burden due to its vast population. He underscored the absence of a specific rare disease definition due to inadequate epidemiological data. Traditional diagnostics have shifted towards cytogenetics and NGS, yet treatment cost remains a challenge. Dr. Ashwin emphasized determining RGD prevalence and carrier frequency using technologies like exome sequencing. He spoke about various country-level initiatives like India's Rare Disease Policy under which 12 centres of excellence have been established across the country. The policy categorizes RGD into three groups, granting eligible patients financial aid and offering up to 50 lakhs for select patients but remains a temporary solution. The Rare Disease Portal archives patient data for crowdfunding, while the National Health Mission prioritizes birth defect screening for a comprehensive preventive strategy. The pediatric rare genetic disorder mission spans 16 centers, focusing on early diagnosis, genetic counseling, and functional assessment of novel genes using models like Drosophila, Zebrafish, and cell lines.

***Unravelling Genetic Diversity in Jammu and Kashmir: Insights from Dr. Swarkar's Exploration:***
Dr. Swarkar highlighted the diverse genetic makeup among ethnic groups in different areas of Jammu and Kashmir (J&K). The region's diverse populations possess unique genetic traits contributing to specific genetic disorders within these communities. The geographical complexity, combined with frequent endogamous marriages, can increase the occurrence of certain genetic disorders due to limited genetic diversity. Understanding these variations is vital for identifying and managing prevalent genetic disorders in the J&K region. Tracing ancestry plays a pivotal role in comprehending these communities. Dr. Swarkar cited instances from various villages: Dhadkai, known for prevalent deaf and mute residents, Arai with a rare hand disfigurement condition, and Pralkot, a village with a high incidence of deaf and mute individuals. He emphasized the significance of 'Project JK DNA' in gathering crucial clinical data to advance research and comprehension of rare genetic disorders in the area.

***Unravelling Frontotemporal Dementia: Insights:***
Dr.Subasree Ramakrishnan discussed her research on frontotemporal dementia (FTD). She highlighted FTD's complexity and variability within the population, discussing its heritable nature and use of genetic testing like the Goldman score. Next-generation sequencing (NGS) and whole exome sequencing (WES) were preferred for diagnosis, studying a cohort of 162 patients exhibiting FTD symptoms at an average age of 56. She emphasized four main genes associated with FTD mutations- MAPT, GRN, SQSTMI, and C9ORF72- acknowledging the importance of understanding these genetic factors for improved diagnosis and management in India

**"Tracing ancestry plays a pivotal role in comprehending these communities. Dr. Swarkar cited instances from various villages: Dhadkai, known for prevalent deaf and mute residents, Arai with a rare hand disfigurement condition, and Pralkot, a village with a high incidence of deaf and mute individuals".**




*From left: Dr. Rakesh Mishra, Prof B.K Thelma, Dr. Swarkar Sharma, Dr. Subashree Ramakrishnan, Dr. Amlin Shukla, Dr. Ashwin Dalal, Mr. Vikas Bhatia, Dr. Ratna Dua Puri*

*From left: Prof B.K Thelma and Dr. Meenakshi Bhat*

# *SESSION 2*
# *BASIC AND CLINICAL RESEARCH-1*

**SESSION CHAIR**

**Prof. ARVIND RAMANATHAN**

*DBT - Institute for Stem Cell Science and Regenerative Medicine*

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**Dr. ANJU SHUKLA**

*Kasturba Medical College, Manipal*

**Dr. SRINIVASARAO REPUDI**

*DBT - Institute for Stem Cell Science and Regenerative Medicine*

**Dr. RADHA RAMA DEVI**

*Rainbow Children's Hospital,Hyderabad*

**Dr. BINUKUMAR BK**

*CSIR - Institute of Genomics and Integrative Biology, New Delhi*

**Dr. RAMACHANDRAN SHAJI**

*Christian Medical College, Vellore*

**Dr. KARTHIK BHARADWAJ**

*CSIR -Centre for Cellular and Molecular Biology, Hyderabad*

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# *SESSION 3*
# *BASIC AND CLINICAL RESEARCH-2*

**SESSION CHAIR**

**Dr. RAVI MANJITHAYA**

*Jawaharlal Nehru Centre for Advanced Scientific Research*

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**Dr. PRATIBHA BHALLA**

*University of Texas Southwestern Medical Centre, USA*

**Ms. PARVATHY KRISHNAN**

*Krishnan Family Foundation*

**Dr. ALKA CHAUBEY**

*Bionano Genomics, Inc*

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# *Rare Genetic Diseases Research Summit (REDRESS)2023:*
## Session 2: Basic and Clinical Research- 1 & 2

The theme basic and clinical research had two editions I and II at the REDRESS 2023. The edition I was curated to highlight the contemporary development in basic, clinical and translational fronts of rare genetic disorders (RGD) in India whereas the edition II consisted of Indian speakers from abroad who shared their expertise on selected segments of RGD. The sessions began with clinical overviews by Dr Anju and Dr Radha Rama Devi followed by mechanistic and genomic research presentation by Dr. Shaji, Dr. Binukumar, Dr. Srinivas and Dr. Pratibha. The diagnostic modality on focus was optical genome mapping with two presentations – scientific aspects covered by Dr Alka and clinical utility by Dr. Karthik. The session ended with patient advocacy group presentation of Parvathy from Krishnan Family Foundation.

Dr Anju Shukla opened her talk with an overview of trends in rare genetic disorders in the past decade by quoting the milestones in diagnosis and discovery due to next generation sequencing, prevention enabled by detection techniques, prenatal and preimplantation testing alongside multidisciplinary care. The major challenges cited were a huge burden, lack of training - expertise and collaborative efforts in our country. Dr. Shukla discussed two RGD cohorts:

- ***Myelin abnormalities in central nervous system consisting of hypomyelination and demyelination. Currently, there are 406 monogenic and four mitochondrial disorders, and 410 genes associated with leukodystrophy. They are a spectrum disorder yet one third of them are not listed under leukodystrophy. She shared a collaborative Indian publication of leukodystrophy with 250 individuals from 226 families from 10 cities. The study reported 68% diagnostic yield and identified population specific variants for Aicardi Goutieres syndrome where 3 patients from south, western and north India had the same variant. She summarised different therapeutic strategies like hematopoietic stem cell transplant, steroidal therapy, supplements, along with emerging ones – RNA, gene, small molecules, and protein therapies.***
- ***Disorders of epilepsy – About 142 families enrolled with diagnostic yield of 52% of which 38 families benefitted with therapeutic implication. A novel gene association in KCNH5 was reported and the case was listed in GeneMatcher. This led to international collaboration with Northwestern university, Chicago and a joint publication. Another interesting finding was sequence variant in SNRPN, an imprinted gene which was only known to have methylation disturbances associated with Prader Willi Syndrome.***

Dr Shukla mentioned about **Center for Rare Disease Diagnosis, Research and Training, a DBT Wellcome India Alliance collaborative program to further RGD research in India.**

***Advancements in Metabolic Disorders and Newborn Screening: A Journey from the 1980s to the Present:***

Rama Devi gave a glimpse of her journey and the progress in metabolic disorders and newborn screening from 1980s till present. She discussed the diverse nature of RGD, with most of them life threatening and remaining undiagnosed. The estimated burden is 72- 96 million in India and the average time for diagnosis is 7 years. No effective treatment is available for many of the disorders and less than 5% have therapies. She emphasised on how finding the right diagnosis and treatment are crucial for improving quality of the life of patients. She discussed few case scenarios where early intervention has enabled near normal life to children with metabolic disorders. Research of several types including treatment, diagnosis and screening is the need of the hour to mitigate this disease burden. Dr Radha also presented the trends in therapeutics and treatments of inborn errors of metabolism that have supplements and treatment available in India including PKU diets, disorders that are expensive and inaccessible like ERT and how repurposing of drugs is the arena where the clinical and academic focus needs to be shifted.

***Advances in Disease Modeling and Drug Screening for Hematological Disorders: Insights***

Dr. R.V. Shaji presented about iPSC-based disease model for learning disease mechanism and drug screening for hematological disorders. **Using CRISPR-Cas9 based genome editing technique he described how multiple genes involved in Fanconi anemia and Diamond Blackfan anemia can be screened.** Further he also described how hematopoietic lineages derived from the disease iPSC lines can mimic the hematopoietic anomalies and bone marrow failure observed in the patients and are better than mouse models. Using this platform his group identified a drug molecule that improved the disease phenotype in Fanconi Anemia hematopoietic cells. Finally, he also described how base editing can be efficiently used for incorporating specific mutations in iPSCs, a technique, they have efficiently used to generate disease iPSC lines for DFA and congenital dyserythropoietic anemia

**"Restoring the Wwox gene in a mouse model with epileptic encephalopathy type 28 reverses neuron defects, promotes neuron survival, and improves myelination, offering potential therapeutic interventions for this condition."**

***Modeling Epileptic Encephalopathy Type 28 in Mice: Insights and Therapeutic Prospects***

Dr. Srinivasarao Repudi presented about modelling and studying development epileptic encephalopathy type 28 in mouse models. His study demonstrated how loss of Wwox, a tumor suppressor gene in mouse causes spontaneous seizures, ataxia, growth retardation, microcephaly, kyphosis of early onset and feeding difficulties. As the Wwox knockout mice dies in 3 weeks, using neuron specific knockout model, he demonstrated neuron associated defects such as defect in oligodendrocyte precursor cell differentiation. Further to develop therapeutic interventions, he showed that delivering the Wwox gene by AAV9 results in rescue of neuron defects, promotes neuron survival and improves myelination. RNA sequencing – electron microscopy showed myelination is highly regulated process. Neuronal WWOX deletion causes epilepsy. WWOX restoration in brain leads to reversal of growth retardation.

***Advancing Understanding and Diagnosis of Wilson Disease:***
Dr. Binukumar presented about Wilson Disease (WD) research program at IGIB. WD is a disorder of copper metabolism and primarily affects the liver and neurons The global prevalence is 1 in 10,000 to 30,000. ATP7B is the associated gene and carrier frequency is 1 in 90. He shared genetic spectrum of variants in WD which is about 4000 across the globe with major being variants of uncertain significance. Few of the essential databases and resources in the analysis, validation and reporting workflow are Clinvar, COSMIC, HGMD. LOVD, mutalyzer, ACMG and AMP guidelines . The IndiGen data for WD is a variant registry that hosts carrier frequency and prevalence in India. IndiGen reports 1 in 69 in India to be carrier and 1 in 18678 to be affected with WD which is second largest in the world. IGIB also hosts WilsonGen, a database for Wilson disease from the 128 processed samples of 238 received for analysis. Dr Binu summarised the workflow for variant prioritization and structural variants along with hotspots and structural variants of ATP7B in Indian WD patients. He presented an interesting algorithm for genetic burden testing of rare variants using Testing Rare variants using Public Data. Insilico models for molecular dynamic simulation of variants using alpha fold protein structures and functional assay with ATP7B knockout-HEK293T cells using CRISPR cas9 is developed for analysing VUS. At the diagnostic front, targeted, scalable and cost effective NGS based diagnosis should be implemented. Dr Binu proposed creating an ecosystem for clinical genomics in India, with WD as an example, wherein WilsonGen has variant database with AMP, ACMG curation, iCROWD has genomic research in WD with whole exome data, targeted variants to be developed by the diagnostic industry, functional validation of VUS and finally cell therapy for patient's iPSC model system and platforms for drug discovery of rare diseases.

***Understanding Thymus Hypoplasia in 22q11.2 Deletion Syndrome:*** Dr Pratibha Bhalla, opened session II by presenting her research on thymus hypoplasia in kids affected with 22q11.2 Deletion Syndrome (DS) also known as Di George syndrome covering aspects of disease mechanism and therapeutic strategies. DS is caused due to 1.5 Mb to 3Mb deletion on 22q11.2 leading to haploinsufficiency of 106 genes including TBX1. Affected individuals suffer from thymus hypoplasia, congenital heart defects, hypoparathyroidism, facial anomalies, and neurological disorders. The 22q11 region is prone to rearrangement and the aetiology is majorly de novo in occurrence. The prevalence is 1 in 2148.

Thymus is a gland involved in immunity and its size is determined by age, stress, DS and FOXN1 mutations. Small thymus causes low T cell count leading to recurrent infections. Most DS patients develop normal but reduced T cell subsets. Her lab developed mouse model of DS with hypoplastic thymus lobes. Fetal thymic organ culture revealed development abnormalities showed hypoplastic epithelial and mesenchymal cells. To identify functionally incompetent stromal cell population, reaggregate thymic organ culture was performed wherein normal mesenchymal could restore hypo plasticity of DS thymuses. Single cell RNA sequencing revealed overexpressed transcripts in mesenchymal population and elevated deposition of collagen in DS. Mouse models, Human DS thymuses have slight elevations in collagens thus revealing a potential therapeutic target. Minoxidil which is FDA approved for the treatment of high blood pressure and hair loss inhibits collagen crosslinking. In utero mouse models showed successful restoration. She concluded her talk by proposing these drugs to be repurposed for DS as compared to expensive In utero gene and stem cell therapy.

**"Repurposing FDA-approved drug Minoxidil, known for treating high blood pressure and hair loss, as a potential therapeutic strategy for addressing collagen-related issues in 22q11.2 Deletion Syndrome, providing a cost-effective alternative to in utero gene and stem cell therapy".**

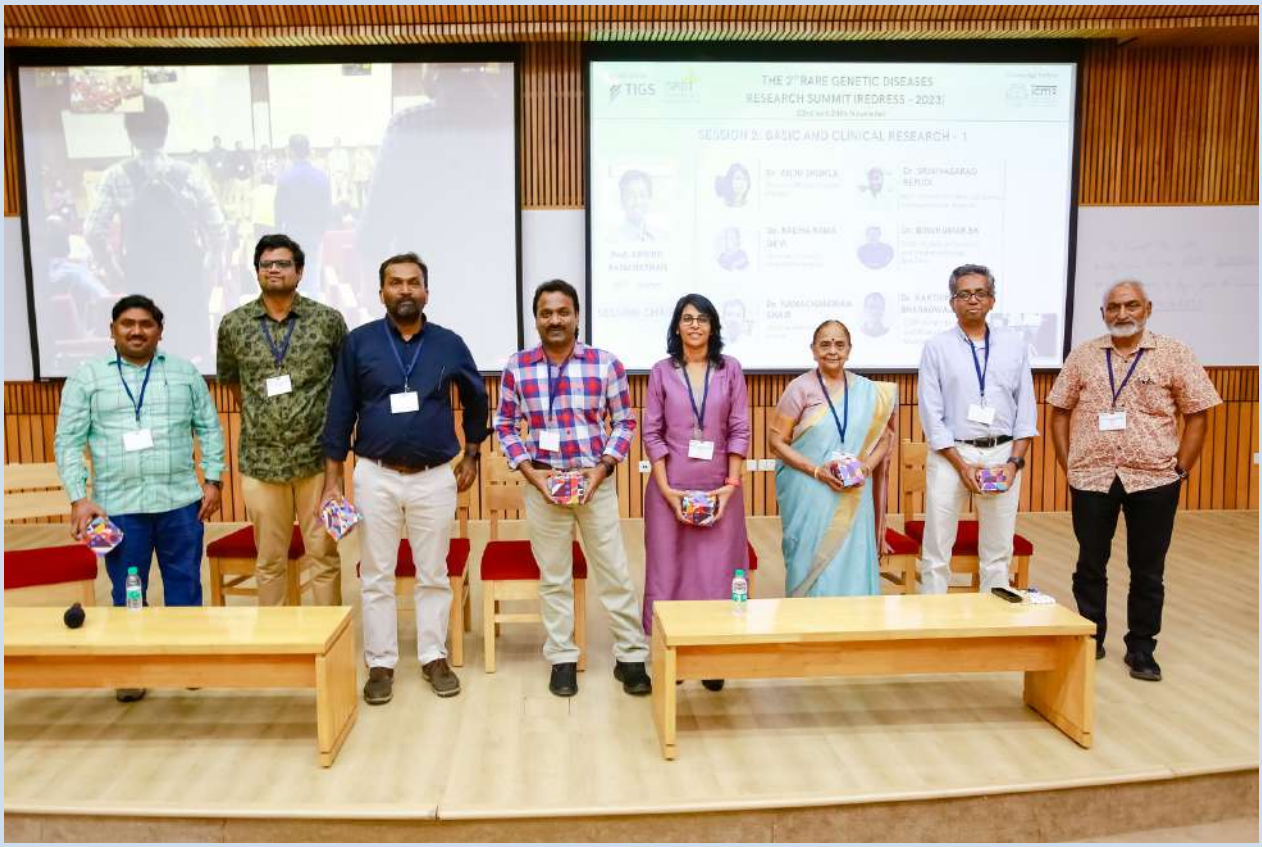
## Optical Genome Mapping

Dr Alka Chaubey from Bionano, USA explained the principles and work-flow of the contemporary technology of optical genome mapping (OGM). She illustrated the clinical utility of OGM in improving diagnostic yield especially in disorders with autism, developmental delays and dysmorphism. She gave a visual overview of evolution of cytogenetic techniques. OGM provides ~1000 times more bands in the form of labels and can detect chromosomal abnormalities as small as 500 bp. OGM can help detect variant classes like aneuploidy, deletion, duplication, translocation, inversion, absence of homozygosity, repeat expansion and contraction. She stated structural variants are important contributors to the mutational landscape of inherited disorders, including retinal diseases where OGM can diagnose 1/3rd of undetected cases. Other examples include reclassification of Duchenne muscular dystrophy variant of uncertain significance, atypical teratoid rhabdoid tumors in two silings with germline retrotransposon insertion in SMARCB1 and a novel fusion detection in B acute lymphoblastic leukemia. A study using OGM in South Carolina for neural tube defects reported diagnostic yield of 10%. She concluded her presentation on remarkable note of combining OGM and short read sequencing can unlock several diagnostic puzzles.

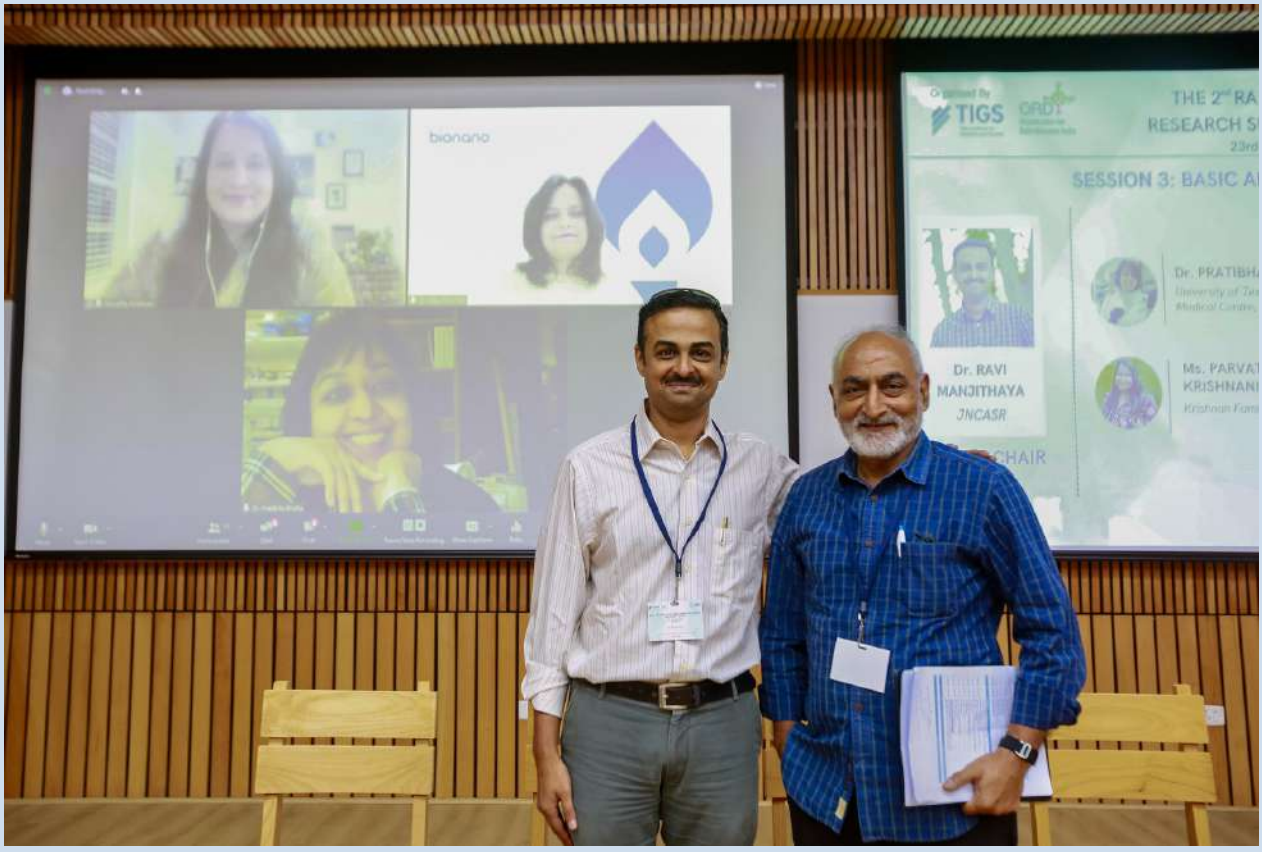
Dr Karthik in session had presented on clinical utility of OGM especially in diagnosis of facioscapulohumeral muscular dystrophy. He gave an overview of ease of sample preparation for OGM with just loading of ultra-high molecular weight labelled DNA on the chip for imaging. At the analysis front, digital molecules are captured, a consensus optical map is generated and compared with the reference. OGM is automated and performs with more than 95% sensitivity. For clinical reporting, denovo variants, ~500 bp is the resolution cut off whereas for somatic changes it is 5KB, and for Inversion it is~30KB. Beginning with FSHD, an autosomal dominant myopathy with 4-10/100,000 cases prevalence involving microsatellite contraction, from their cohort, 23 patients were confirmed with OGM. Another case of Hemophilia A where whole exome sequencing was negative, OGM helped in identifying inversion in intron 22. In a case with developmental delay, dysmorphism, a balanced translocation was identified in chromosome 12 showing ~16mb, fusion depicting copy number gain. Other examples included fragile X and disruption of AUTS2 gene among others. The challenges with OGM include lack of adequate reference data, inability to detect structural variants across acrocentric region and poorly labelled regions.

## Parent Advocacy Group

Parvathy from Krishnan Family Foundation, USA shared her family's journey with constitutional mismatch repair deficiency (CMMRD) with her son being diagnosed at a very young age with ampullary cancer, jejunal cancer, two incidences of small bowel cancer and rectal cancers. CMMRD is an autosomal recessive disorder caused due to two mutant alleles of EPCAM gene. Having a single affected allele is enough to cause hereditary cancer predisposition syndrome, to the family's dismay, both parents were heterozygous for the variant allele. This was followed by their daughter who in addition also inherited several other recessive alleles and affected with not only CMMRD but also Bardet Biedl syndrome, Factor VII deficiency and other debilitating conditions leading to her untimely passing away at the age of 4 years. Parvathy emphasised the need for genetic counselling and extended family screening in inherited conditions and shared how several family members including herself have undergone the cascade testing and are under periodic surveillance. The family has shown unusual resilience in the face of this medical complexity and are supporting several others who are similarly affected with family support, surveillance implementation, rare disease registry. They are also proactive in facilitating centralized repository for biosamples, are striving to implement policies to create ICD codes for CMMRD, coverage for surveillance protocols and spreading awareness about the disorder in several forums. Parvathy concluded that PAGs are forthcoming to participate in research studies to accelerate the diagnosis, treatment, and management of RGDs.




*From left: Dr. Srinivasarao Repudi, Dr. Karthik Bharadwaj, Dr. Ramachandran Shaji, Dr. BK Binukumar, Dr. Anju Shukla, Dr. Radha Rama Devi, Prof. Arvind Ramanathan, Dr. Rakesh Mishra*




*From left: Online: Upper panel- Parvathy Krishnan, Dr. Alka Chaubey, Lower Panel- Dr. Pratibha Bhalla. Dr. Ravi Manjithaya and Dr. Rakesh Mishra*

# *SESSION 4*
# *ENHANCING RGD THERAPY AND RESEARCH ENVIRONMENT*

**SESSION CHAIR**

**Prof. ANURANJAN ANAND**

*Jawaharlal Nehru Centre for Advanced Scientific Research, Bengaluru*

**DR. NABENDU SEKHAR CHATTERJEE**

*Indian Council for Medical Research, New Delhi*

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**Dr. RITA SARIN**

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**Dr. JOGIN DESAI**

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**Dr. AJAY SINGH**

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**Dr. SADHNA JOGLEKAR**

*Novartis India, Hyderabad*

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## PATIENT ADVOCACY GROUPS

**Ms. MOUMITA GHOSH**

*CureSMA foundation of India*

**Ms. KIRTI DA OZA**

*Sjogrens's India*

# Research, Innovation, and Hope for Genetic Diseases

A vibrant group of experts delved into the challenges and opportunities in developing effective therapies and diagnostics for Rare Genetic Diseases (RGD) at the session on "Enhancing RGD Therapy and Research Environment".

Dr. Taslim ignited the discussion, emphasizing the transformative power of innovation in delivering both therapeutics and diagnostics for RGDs. He stressed the crucial role of evidence-based research, urging its translation into tangible social impact. He also highlighted the need for robust investments in science and technology, nurturing a virtuous cycle of good science fueling further advancements.

Intellectual property rights expert Dr. Rita Sarin shed light on the financial hurdles hindering RGD drug development. She explained the lengthy, expensive process of creating a drug molecule, a major deterrent for pharmaceutical companies due to low returns. She delved into the intricacies of licensing and various patent types, emphasizing the healthcare industry's focus on primary patents due to limitations on secondary patents in India. Dr. Sarin also showcased inspiring examples of successful indigenous and generic medications, while proposing valuable solutions. These included integrating research pipelines and fostering collaborative research, paving the way for either industry-academia partnerships to drive innovation or affordable API (active pharmaceutical ingredient) supplies through collaborations between innovative firms and the Indian pharmaceutical sector.

***Unveiling the Challenges of Retinitis Pigmentosa: Insights and Recommendations:***

Dr. Jogin Desai brought the spotlight to retinitis pigmentosa, a rare congenital retinal disease. He highlighted the devastating consequences of undiagnosed cases due to insufficient data, stressing the societal costs of inaction. He advocated for integrating genetic insights into societal knowledge, laying the groundwork for a comprehensive understanding of these often-overlooked conditions. His concluding remarks focused on key areas for improvement, including:

- Establishing a robust regulatory framework and addressing cost concerns through a homegrown innovation ecosystem.
- Approving in vitro experiments for RGD research, potentially reducing dependence on animal models.
- Enabling GMP (Good Manufacturing Practices)-like production facilities.
- Developing both a validated safety model and a gene-agnostic model.

Dr. Ajay Singh broadened the lens, emphasizing the need for a diverse RGD ecosystem encompassing investors, NGOs, manufacturers, researchers, industry players, and patient advocacy groups. He underscored the fact that a staggering 80% of rare diseases have a genetic component, highlighting the collective responsibility to tackle this widespread challenge.

Dr. Sadhna Joglekar provided a global perspective, presenting a landscape study of advancements in rare disease drug research from 1950 to 2023. She underscored the significance of collaborative decision-making and incentivizing rare disease drug development. Recognizing the complexities in research planning and execution, she advocated for overcoming these hurdles to accelerate progress.

***Government Initiatives in Rare Genetic Diseases:*** Dr. Nabendu Chatterjee shed light on government initiatives through ICMR (Indian Council of Medical Research), outlining the various ways in which the government is supporting RGD research and addressing crucial gaps:

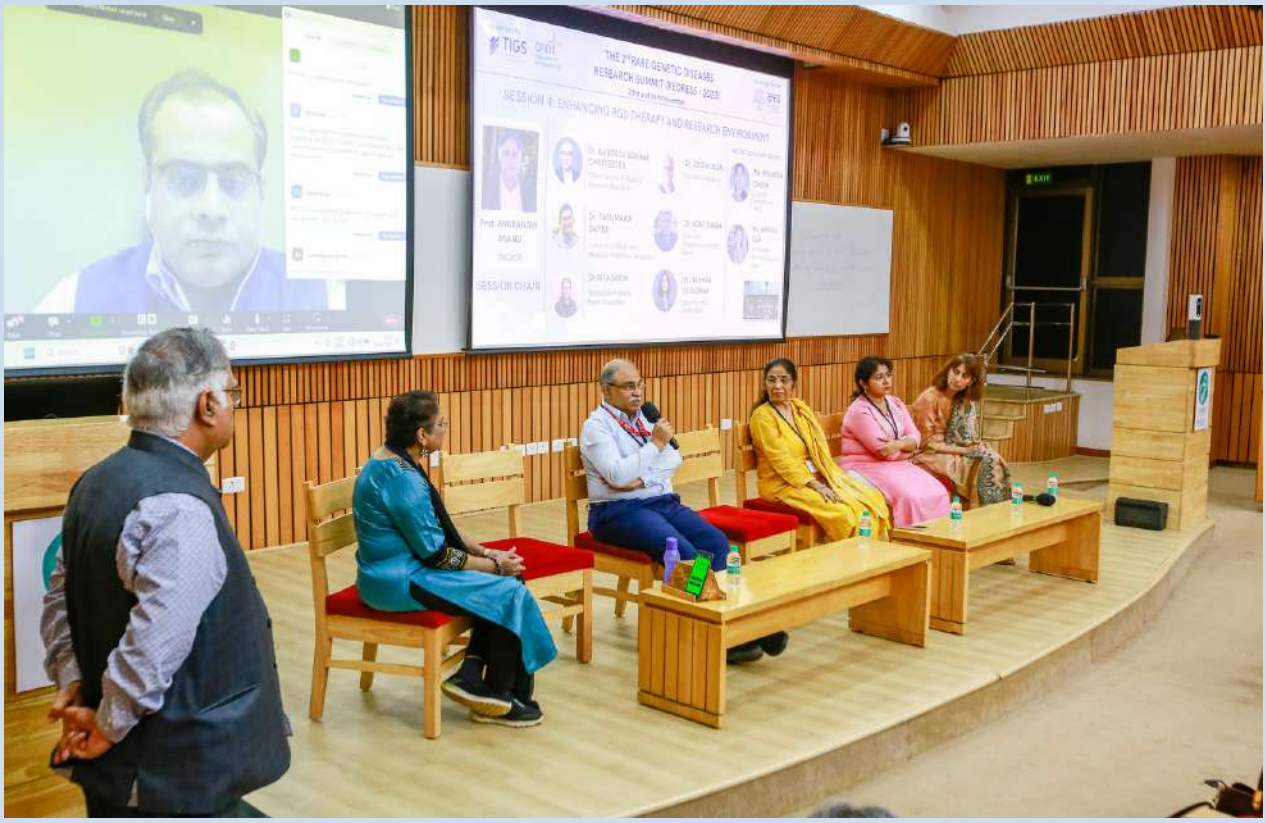
- Early diagnosis
- Epidemiological data collection
- Therapeutics availability and affordability
- Awareness and expertise development
- Dedicated research initiatives

To address the critical challenges of therapeutic availability and affordability, Dr. Chatterjee suggested increasing grants, fostering collaboration, and strengthening networking efforts. He further elaborated on existing government initiatives such as genomics projects, networking between patient advocacy groups and other stakeholders, therapeutics development for inherited rare diseases, and the National Apex Committee: National Consortium for Research and Development on Therapeutics in Rare Diseases (NCRDTRD). He concluded by outlining potential future directions, including prioritizing early diagnosis, developing novel or indigenous treatments, establishing a national registry, and characterizing rare diseases in the Indian context.

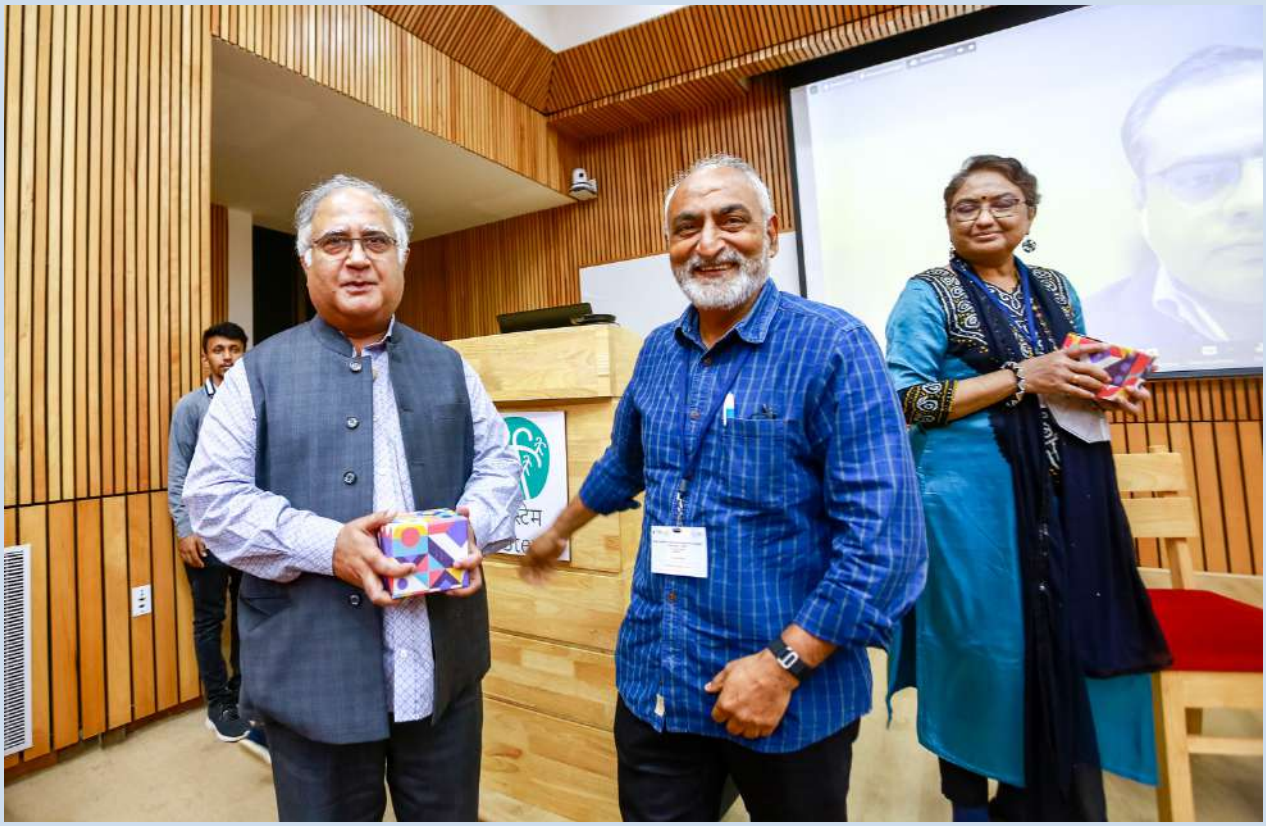
The latter part of the event resonated with personal stories from patient advocacy groups. Mrs. Moumita Ghosh of the Cure SMA Foundation of India shared the heart-wrenching journey of her daughter with SMA (Spinal Muscular Atrophy), highlighting the struggles in accessing proper care and treatment. She spoke passionately about the organization's mission and how they support individuals with SMA. Ms. Kritida Oza, living with Sjogren's disease, shared her experiences and the challenges faced by patients with this rheumatic condition for which no specific therapy exists. Her plea for equal opportunities and acknowledging patients' perspectives added a powerful human touch to the discussion.

**"Increased grants, collaboration, and networking efforts emphasizing existing government initiatives such as genomics projects, therapeutics development, and the National Consortium for Research and Development on Therapeutics in Rare Diseases (NCRDTRD) would help in addressing challenges in therapeutic availability and affordability"**

The event offered a potent mix of expert insights and lived experiences, illuminating the path towards a brighter future for those living with RGDs in India. By fostering collaboration, innovation, and a supportive ecosystem, we can ensure that no one is left behind in the quest for effective therapies and a better quality of life.




*From left: Prof. Anuranjan Anand, Ms. Kirti Da Oza, Dr. Nabendu Sekhar Chatterjee, Dr. Rita Sarin, Ms. Moumita Ghosh, Dr. Sadhana Joglekar Online: Dr. Ajay Singh*



*From left: Prof. Anuranjan Anand and Dr. Rakesh Mishra*

# *PANEL DISCUSSION: 1*
# *RARE GENETIC DISEASE POLICY AND INSURANCE*

**PANEL CHAIR**

**Mr. SAMIR SETHI**

*Indian Rett Syndrome Foundation*

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**Dr. VISWANATH PINGALI**

*Indian Institute of Management, Ahmedabad*

**Prof. SANJEEV JAIN**

*National Institute of Medical Health and Neurosciences, Bengaluru*

**Dr. NEERJA GUPTA**

*AIIMS, New Delhi*

**Dr. SHAKILA N**

*Department of Health and Family Welfare, Government of Karnataka*

**Mr. NABENDU DAS**

*Centogene, Noida*

# Rare Genetic Disease Policy and Insurance

The second Rare Genetic Disease Research Summit (REDRESS) 2023 spanned two consecutive days, the 23rd and 24th of November 2023 involving panel discussions on each day about different themes. The Day one panel discussion was themed "Rare genetic disease policy and insurance" and was chaired by Mr. Samir Sethi (Member, Indian Rett syndrome Foundation and Chairman of advocacy committee group, ORDI ) and distinguished panelists - Dr. Vishwanath Pingali (Indian Institute of Management, Ahmedabad), Dr. Neerja Gupta (All India Institute of Medical Sciences, Delhi), Prof. Sanjeev Jain (National Institute of Mental Health and Neurosciences, Bengaluru), Mr. Nabendu Das (Centogene, Greater Noida) and Dr. Shakila N (Department of health and family welfare, Government of Karnataka, Nodal Officer for Sickle Cell Mission, Karnataka). The session was initiated with the introduction of panellists followed by discussions.

***Evaluating India's Rare Disease Policy and Challenges in Implementation***

The rare disease policy, which is an important framework set up by the government, was made in 2017 and was updated in 2021. The discussion highlighted the need for understanding what the policy offers and how it can be improved further.

The government has established Centres of Excellence (CoEs) for the diagnosis and treatment of rare diseases. These CoEs, initially designed as tertiary care multi-speciality centres, present certain challenges. Sensitizing medical staff to rare diseases amidst patients with varied medical conditions is a primary concern. The heterogeneous nature of rare diseases makes it challenging to determine patient numbers accurately. Consequently, patients often navigate multiple wards, leading to time constraints for both patients and clinicians. Additionally, challenges include medication procurement, paperwork, delays in drug delivery, and dosage adjustments based on weight.

***Shaping the Future: Standardization, Education, and Government Involvement in Rare Genetic Disease Management***

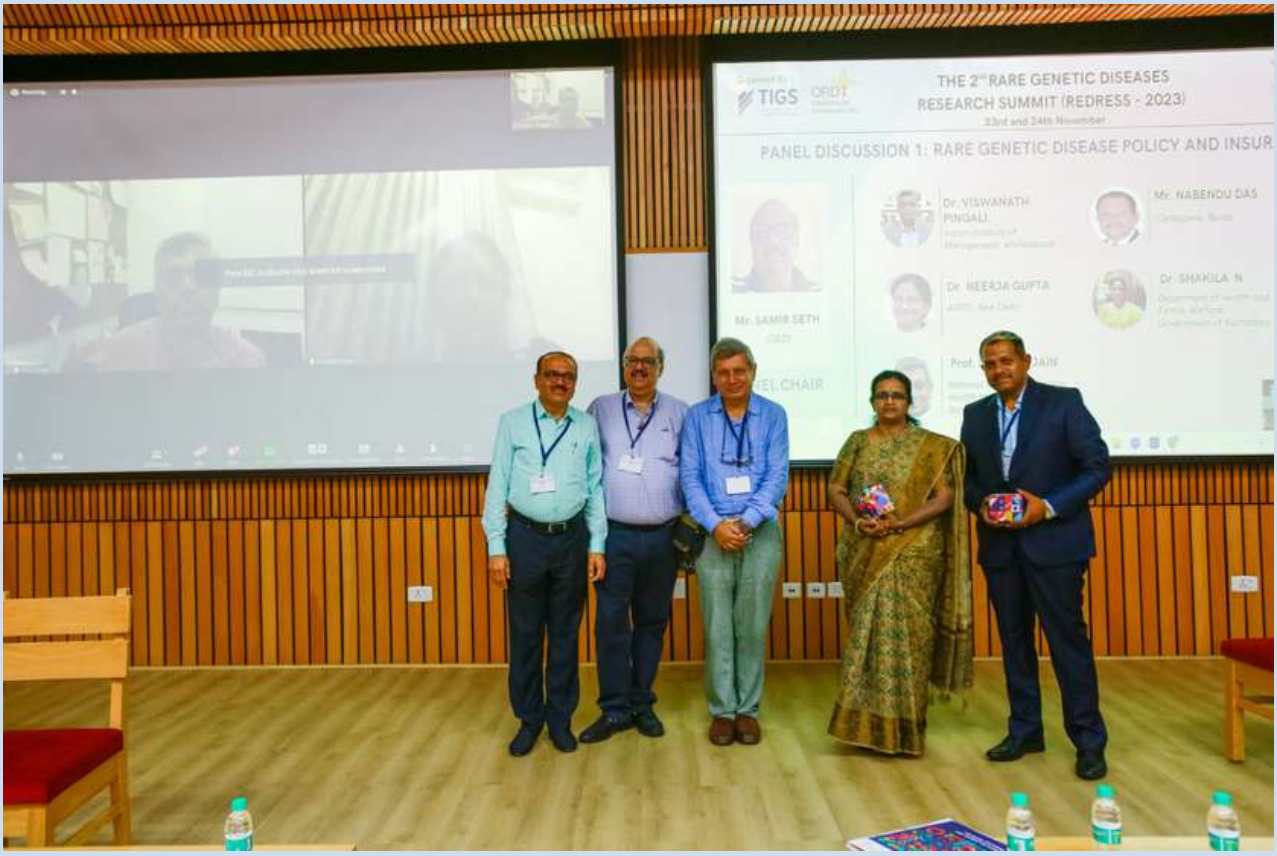
The discussion also highlighted the importance of assortment and standardization of rare diseases definition and classification, which, due to lack of epidemiological data, is not clear in the existing rare disease policy. It was suggested that there should be a universal consensus on the parameters that classify a certain genetic condition as rare. To attract recognition and support, setting up a standard is necessary. The CoEs maintain the registry of the rare disease patients that can be used to get the prevalence data necessary for research and development.

The proposition to incorporate RGDs into the academic curriculum, complementing the existing courses accessible on the IAMG (Indian Academy of Medical Genetics) website, was advocated by the panellists. Also, establishing a well-structured Standard Operating Procedure (SOP) can be brought forward which is crucial for diagnosing patients, particularly those residing in remote locations, to prevent unnecessary travel. One effective measure is the doorstep collection of samples, minimizing the need for patients to commute.

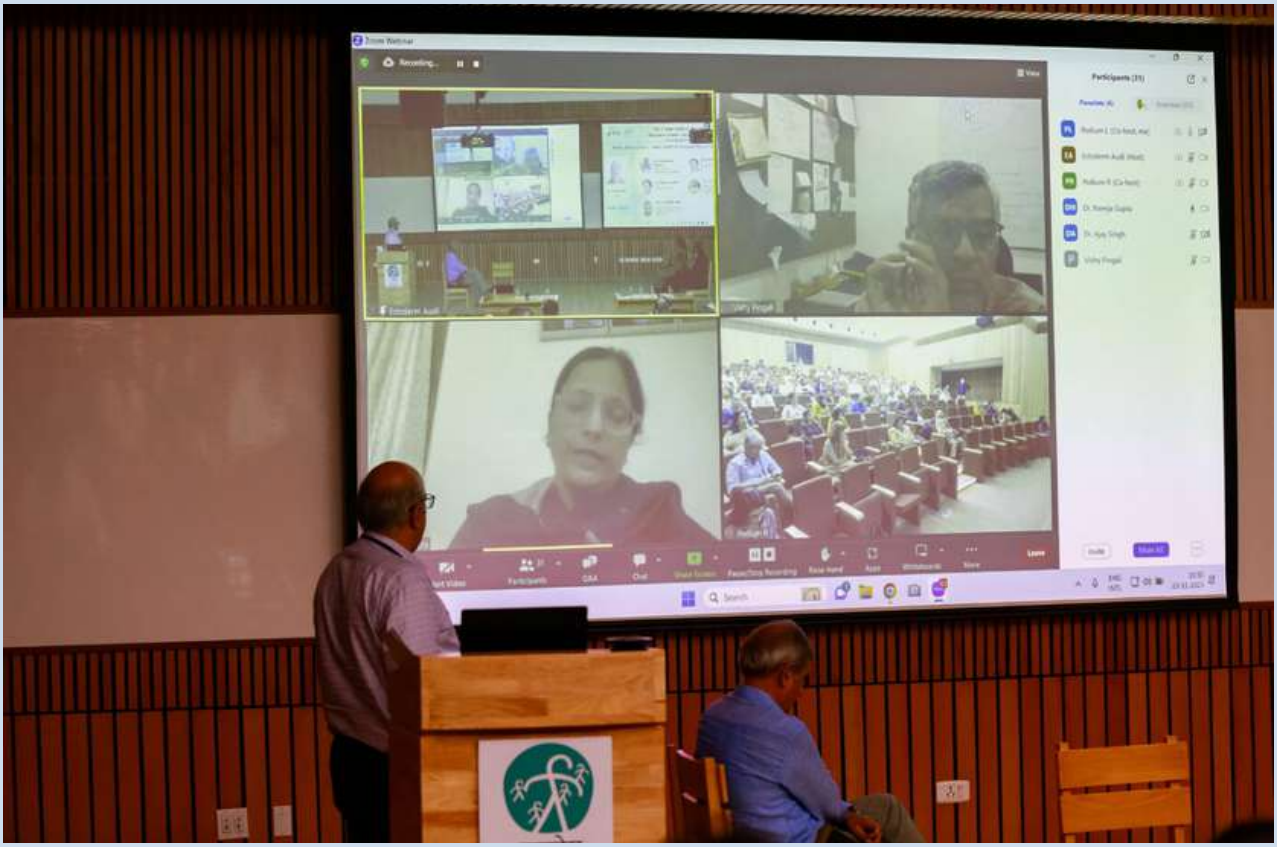
The panel deliberated on the pivotal role of the government in the management and treatment of Rare Genetic Diseases (RGDs). Emphasizing the necessity of establishing unequivocal policies, the discussion underscored the government's responsibility to subsidize pharmaceuticals dedicated to the treatment of rare genetic conditions. A crucial observation underscores the potential of Government laboratories to motivate private industries to engage in the development of therapeutics for rare genetic diseases. The need to address awareness of RGDs among people can be a milestone in dealing with respite care and disease management apart from therapeutics and diagnostics. With the RGD patient-friendly infrastructure, the focus should be on the primary concern of the patients.

With the democratisation of knowledge after COVID-19, it's easier to create awareness of the therapeutics and discovery of potential drugs for RGDs. If more companies in India are established to tackle drug discovery, there will be an increase in treatment, in other words, the treatment won't be rare and hence the costs will be brought down without compromising the quality, as witnessed in the past 4-5 years. Supportive care should be the prime focus of the government, and the pavements and infrastructures should be RGD patient friendly. To avoid patient inconvenience, tele-calling facilities should be particularly formed for people suffering from RGD. Once the RGD prevalence data is clear, it can pave the way for the interests of startup builders in diagnostics and therapeutics.

**"There is a necessity for a universal consensus on rare disease classification, advocating for standardized definitions. The importance of incorporating Rare Genetic Diseases (RGDs) into academic curricula and establishing a well-structured Standard Operating Procedure (SOP) is highlighted, with an emphasis on government responsibility in subsidizing pharmaceuticals and fostering public awareness for effective disease management"**




*From left: Mr. Prasanna Shirol, Mr. Samir Sethi, Prof. Sanjeev Jain, Dr. Shakila N, Mr. Nabendu Das*



*Mr. Samir Sethi*
*Online: Dr. Vishwanath Pingali and Dr. Neerja Gupta*

# *PANEL DISCUSSION: 2*
# *INDIAN ORPHAN DRUG MARKET:OPPORTUNITIES*

**PANEL CHAIR**

**Prof. K. THANGARAJ**

*CSIR - CCMB*

**DR. CHARU GAUTAM**

*IQVIA, Ahmedabad*

**MR. PARAMESHWARAN SITARAM**

*Sanofi Specialty Care, New Delhi*

**Dr. ANIL KUKREJA**

*AstraZeneca, Bengaluru*

**Dr. MONIKA PAHUJA**

*Indian Council of Medical Research, Headquarters,New Delhi*

**Ms. SHARMILA MULPUR**

*National Insurance Company, Bengaluru*

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## PATIENT ADVOCACY GROUPS

**Mr. SAMIR SETHI**

*Indian Rett Syndrome Foundation*

# Indian Orphan Drug Market: Opportunities

With a similar commitment towards RGD support, the theme of the REDRESS second day panel discussion was " Indian Orphan Drug Market: Opportunities". The session was chaired by Dr. Thangaraj (CCMB, Hyderabad) and prominent panellists Dr. Charu Gautam (IQVIA, Ahmedabad), Dr. Anil Kukreja (AstraZeneca, Bengaluru), Ms. Sharmila Mulpur (National Insurance Company, Bengaluru), Mr. Parameshwaran Sitaram (Sanofi), Dr. Monika Pahuja ( Indian Council of Medical Research, Headquarters, New Delhi), Mr. Samir Sethi ( Member, Indian Rett Syndrome Foundation), Mr. Sri Harsha (Indian Prader Willi Syndrome association).

***Policy Perspectives on Rare Genetic Disease Pharmaceuticals: Addressing Coverage Gaps in Insurance***

One of the primary objectives of the panel session was to offer definitive policies and assume accountability for subsidizing pharmaceuticals targeting rare genetic diseases. Globally, the orphan drug market was estimated to be 140,000 million USD in 2020, and 450,000 million USD in 2030. The one-time treatment costs for rare diseases range from several lakhs to crores, in INR.

Given the emphasis on insurance expenditures during discussions, it was proposed that these expenses be structured to encompass all facets related to rare genetic diseases. Currently, some of the insurance policies cover the costs of clinical trials. However, since RGDs have a genetic origin and are considered as 'pre-existing' conditions, several challenges are faced by the patients to cover their treatment costs under insurance. However, a newborn with a genetic disorder is covered under insurance, post 90 days of birth, if the parents are covered under maternity policy. Currently, rheumatic and dental treatments are not covered under insurance policies in India. It was suggested to include these as the costs of treatment are exorbitant. There were comprehensive suggestions for addressing the needs of RGD patients by approaching insurance coverage for outpatient care. For instance, some patients with SMA need lifelong respiratory care, similarly, individuals with Gaucher's disease need to cover expenses for blood transfusions etc. These coverages should be brought under the umbrella of insurance for RGDs.

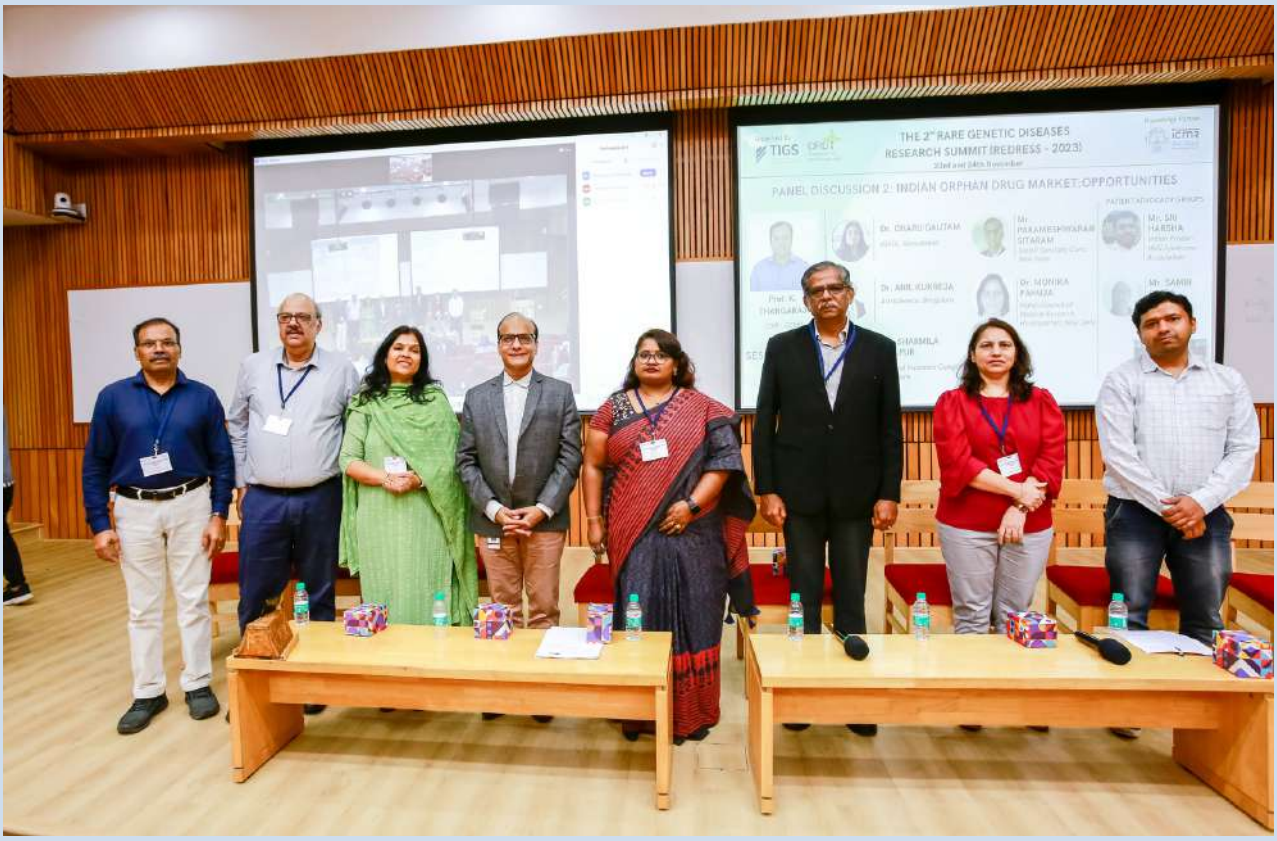
Another important highlight of the discussion was centred around clinical trials in India for orphan drugs. The panel focused on the important regulatory requirements for biosimilar trials in India. Every life is precious and clinical trials should be formulated in India similar to the new Western policies of flexible trials. New designs that incorporate phase 0/1 trials into the spectrum of phase 2 can be accepted, thus skipping the need for separate clinical trials for each phase. Also, separate clinical trials for biosimilars need to be formulated to achieve effective approval for a new trial.

It was suggested that a few of the regulations need to be considered to conduct clinical trials in India which include proof of efficacy similarity against the reference biological product, identification of risks associated with differences in biosimilars and the reference products. Furthermore, an insightful novel idea was put forward by the panel which suggested that instead of looking for the similarities in science, switch to the differences that can lead to resourceful identification of variations in assessing the genotoxicity and immunogenicity related to the drugs. Introducing drug repurposing through proper academic research and proof of concept can be aspired for efficient orphan drug development research.

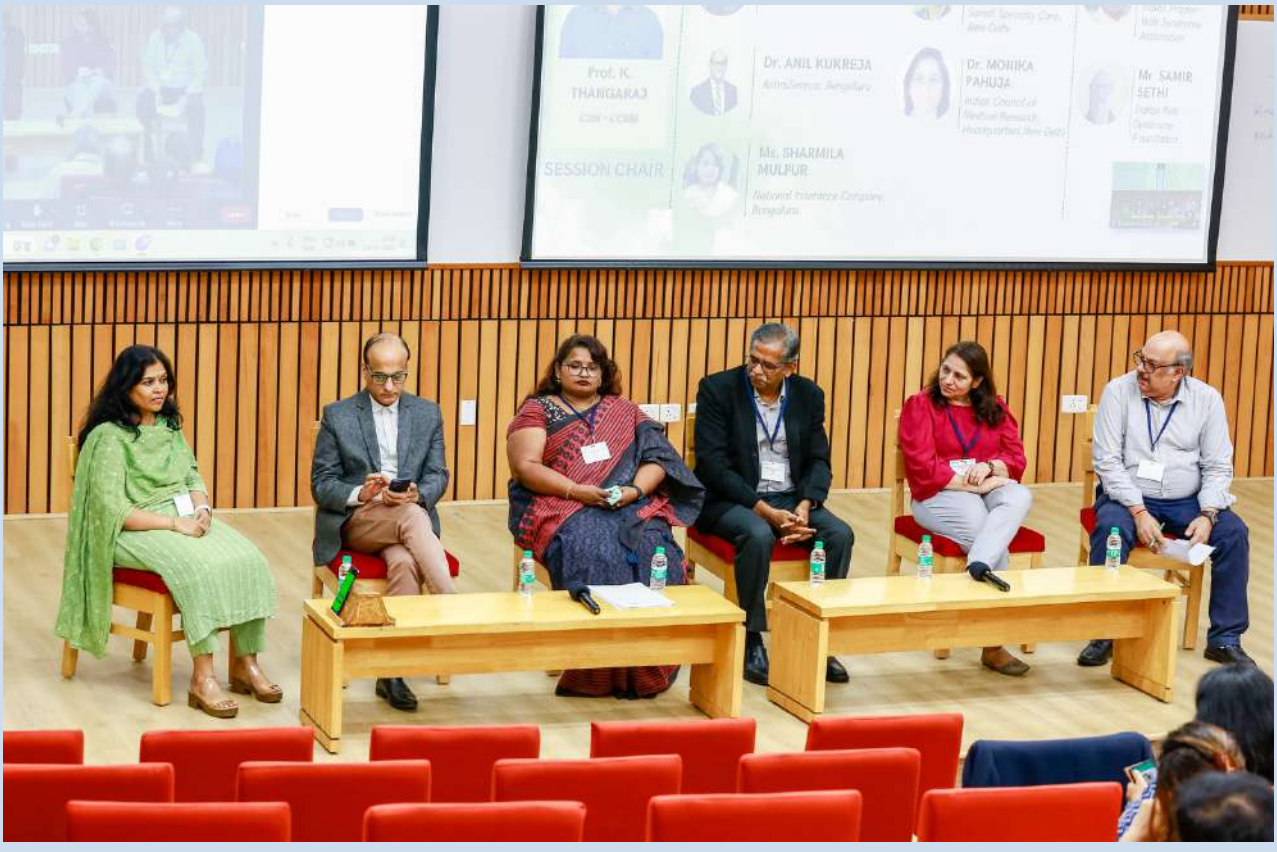
**"Comprehensive suggestions include covering lifelong care needs, such as respiratory care for SMA patients and expenses for blood transfusions in Gaucher's disease, under the insurance umbrella for rare genetic diseases."**

Because of the low number of affected populations with RGDs, they have been recognised in the Orphan Drug Act which is formulated for drugs with a limited market. With the narrow market, drug manufacturers need some incentives to invest in this cause. Therefore, government of India need to formulate certain laws to boost the production of these drugs indigenously.

Another point of discussion was the possible challenges that can come across in developing therapies in the form of market challenges or small population sizes for RGDs. Even after developing therapies, the challenge of the market comes with high costs and with the idea of developing drugs for a small population, the industry interference is limited. This can be tackled by introducing smart acceleratory pathways from government funding and certain waivers for clinical trials. There is a need to look through a global perspective, and the stringent rules that hinder pharmaceutical companies from looking into RGD need to be amended.




*From left: Prof. K Thangaraj, Mr. Samir Sethi, Dr. Charu Gautam, Dr. Anil Kukeraja, Ms. Sharmila Mulpur, Mr. Parmeshwaran Sitaram, Dr. Monika Pahuja, Mr. Sri Harsha*




*From left: Dr Charu Gautam, Dr. Anil Kukreja, Ms Sharmila Mulpur, Mr. Parmeshwaran Sitaram, Dr. Monika Pahuja, Mr. Samir Sethi*

# *SESSION 5*
# *PROGRESS AND WAY FORWARD IN RGD FIELD*

**SESSION CHAIR**

**Prof. VIJAY CHANDRU**

*IISc*

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**Prof. DEEPA BHAT**

*JSS Medical college, Mysuru*

**Dr. RADHA RAMA DEVI**

*Rainbow Children's Hospital, Hyderabad*

**Prof. B K THELMA**

*University of Delhi*

**Mr. SRI HARSHA**

*Indian Prader-Willi Syndrome Association*

**Prof. K THANGARAJ**

*CSIR- Centre for Cellular and Molecular Biology, Hyderabad*

# Rare Genetic Diseases Research Summit Calls for Collaboration to Conquer Challenges

The Rare Genetic Diseases Research Summit (REDRESS) 2023 concluded with a resounding call to action, urging researchers, clinicians, pharmaceutical companies, and patient advocacy groups to join forces and tackle the challenges of rare genetic diseases (RGDs).

The final session, aptly titled "Progress and Way Forward in RGD Field," saw Prof. Thelma, a scientist from the University of Delhi, deliver a powerful message: "Give to the society what we have in hand." Her words resonated with the audience, highlighting the collective responsibility to improve diagnostics, therapeutics, and overall care for RGD patients in India.

The session delved into various aspects of the RGD landscape, with Dr. Deepa Bhat, a genetic counsellor, emphasizing the need for practical solutions like expanded government schemes and public-private partnerships. She stressed the importance of incorporating diverse expertise, including psychologists, public health experts, and clinicians, into teams addressing RGD issues.

Sri Harsha, a parent of a child with RGD, shared his personal experience, highlighting the critical role of government intervention and access to specialized healthcare facilities in managing his child's condition. Dr. K. Thangaraj, a geneticist, brought a unique perspective, explaining how his research on population evolution in India revealed the existence of distinct sets of RGDs in each community due to endogamy practices. He emphasized the crucial role of population genetics research in raising awareness and preventing future occurrences of these diseases.

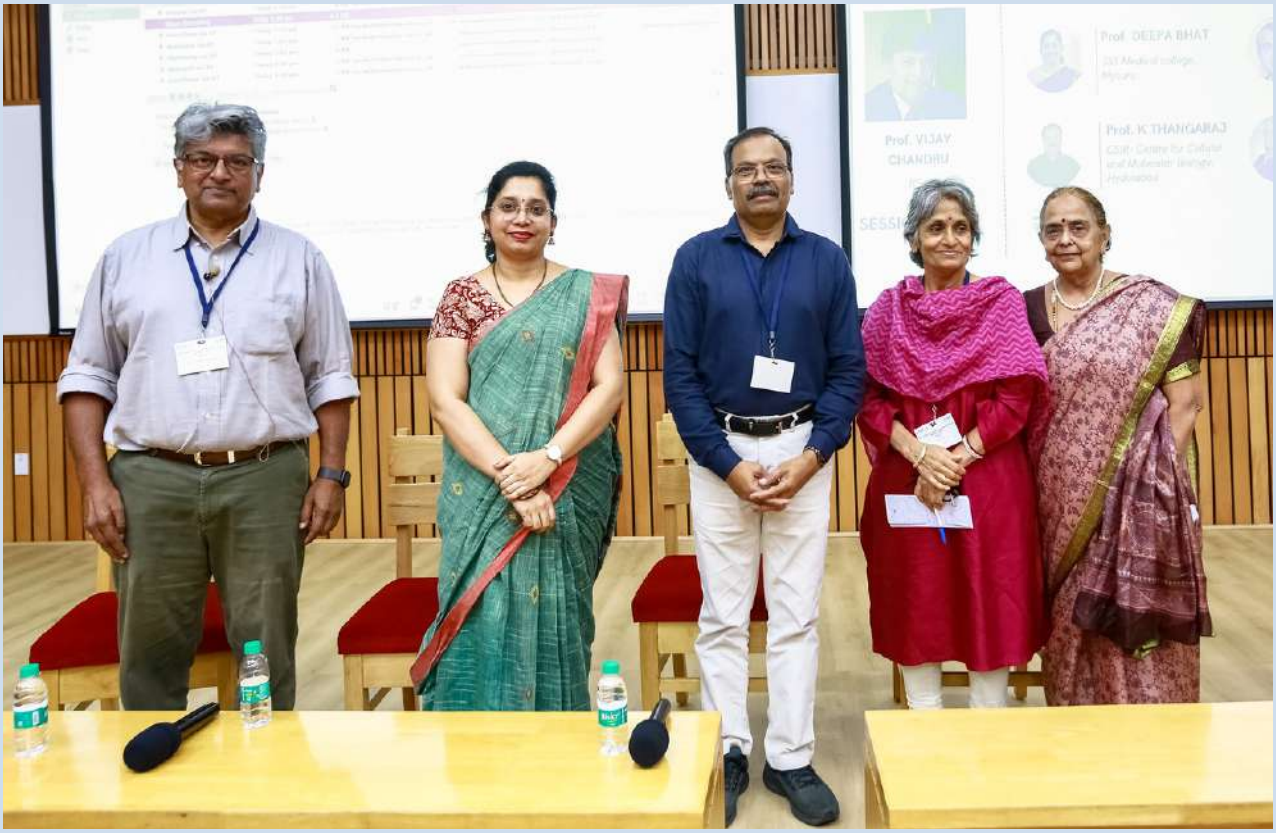
The session chair, Dr. Vijay Chandru, proposed leveraging AI to bridge the gap between community workers and tertiary healthcare providers. He also emphasized the need for cost-effective solutions and innovative technologies, like gene therapy, to ensure their accessibility to patients. Prof. Thelma echoed this sentiment, advocating for mandatory Newborn Screening for 50-60 treatable disorders to facilitate early diagnosis.

The discussion revealed a shared understanding of the challenges and opportunities in the RGD field. Dr. Radha Rama Devi and Dr. Bhat highlighted the stigma surrounding RGDs, adding an additional layer of suffering for patients.

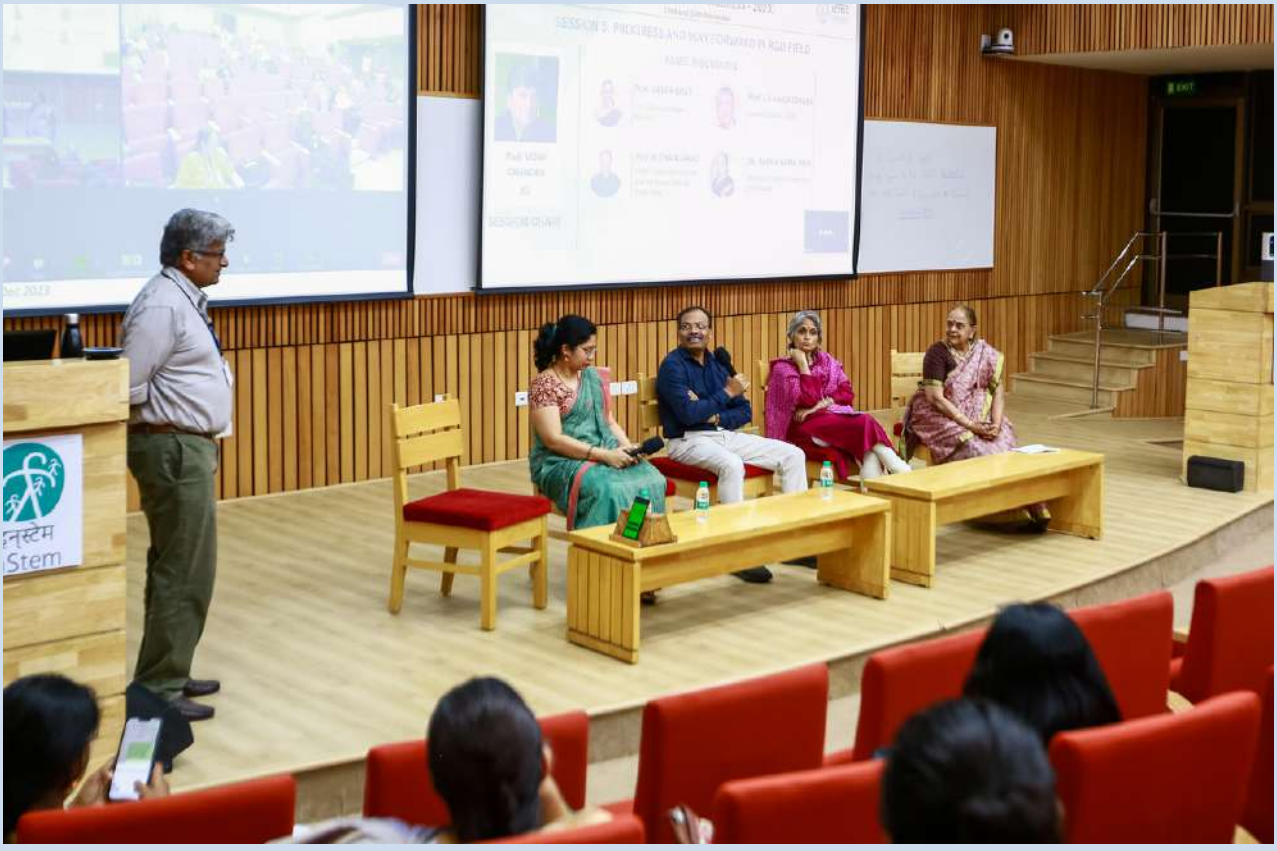
Closing the summit, Dr. Rakesh Mishra, Director of TIGS, thanked the diverse group of participants and emphasized the ongoing commitment to finding solutions for RGDs. He outlined key takeaways, including:

- Early diagnosis: Making prenatal and newborn screening mandatory for treatable disorders.
- Treatment: Exploring repurposed and low-cost drugs, establishing infrastructure for drug testing, and fostering collaboration partnerships between government, administrative and private stakeholders - both in R&D and policy
- Teamwork and collaboration: Building strong partnerships between researchers, clinicians, and industry stakeholders.
- Science and R&D: Utilizing cutting-edge technologies such as CRISPR and mRNA technologies to bring down costs and ensure quality manufacturing.
- Funding: Exploring philanthropic funding, CSR initiatives, and the National Research Foundation.
- Information and databases: Creating a federation of databases and leveraging digital tools for virtual consultations.

Dr. Mishra concluded with a message of hope, stating that "a few success stories are all that are needed for this model to go forward." With the enthusiasm and expertise displayed at REDRESS 2023, the future of RGD research in India appears bright, offering a glimmer of hope for countless individuals and families affected by these debilitating conditions.




*From left: Prof Vijay Chandru, Prof Deepa Bhat, Prof K Thangraj, Prof B K Thelma, Dr Radha Rama Devi*



*From left: Prof Vijay Chandru, Prof Deepa Bhat, Prof K Thangraj, Prof B K Thelma, Dr Radha Rama Devi*

# ANNEXURES

The REDRESS 2023 Poster presentation received over 80 abstracts from 53 institutes across India. The abstracts were invited under 8 categories of Basic Research, MedTech/DigiHealth, Clinical Research Regulatory/IP, Translational Research, StartUp/Industry, Diagnostics and Policy Research. From the shortlisted abstracts, 43 authors successfully presented their work at REDRESS 2023 summit.

| Name (Last name, First name) | Title | Affiliation/Institute name |
|---|---|---|
| Karthik C | Lentiviral Vectors in Gene Therapy of β-Globinopathies; A Pursuit for a Strong Backbone | CMC-Centre for Stem Cell Research |
| Shakhapur, Nidhi | Artificial intelligence to diagnose rare genetic disorders - Early Diagnosis of Mitochondrial Genetic Disorders Using Deep Learning Approach | Vellore Institute of Technology |
| Srivastava, Esha | A study to assess the quality of life of beta thalassemia major patients in India | Tata Institute for Genetics and Society |
| Singh, Manoj | Better Policy Making and Implementation for Rare Diseases | Power In Me Foundation |
| Anjum, Uzma | Shifting Knowledge Landscapes in Rare Diseases Policy Formation: A Study of Advocacy, Definitions, and Collaboration in India | Indian Institute of Technology, Bombay |
| Dr Challa, Viveka Santhosh Reddy | Clinically and Laboratory Profile and Outcome of Allen Herndon Dudley Syndrome- AHDS/ MCT8 Deficiency: Four Genetically Confirmed Cases from a Single Tertiary Care Center. | Indira Gandhi Institute of Child Health (IGICH), Bengaluru |
| Wagle, Aseem | Determination of Tyrosinemia metabolic biomarkers in human plasma by GCMS | Shimadzu Analytical (India) Pvt. Ltd. |

| Name (Last name, First name) | Title | Affiliation/Institute name |
|---|---|---|
| Kodandapani, Sreelakshmi | Clinical utility of fetal autopsy and genetic testing in fetal structural anomalies: An experience from malnad region: Karnataka | Malnad hitech diagnostic centre |
| Babu, Sharath | Childhood Onset Spinocerebellar Ataxia (SCA) Type 7- Two cases, Masquerading as Mitochondrial Disorders from southern India. | Indira Gandhi Institute of Child Health |
| Babu, Sharath | Pompe Disease Masquerading as GM1 Gangliosidosis: Coarse Facies with Extensive Mongoloid Spots and Pyramidal Signs | Indira Gandhi Institute of Child Health |
| M Govind | Modelling vascular impairments associated with ACDC disease using NT5E gene-edited human aortic endothelial cells. | PhD Scholar, Manipal Institute of Regenrative Medicine (MIRM) |
| Lakshminarayana, Shilpa | A novel homozygous mutation in aspartoacylase gene causing Canavan disease | Bangalore Medical college and research Institute |
| Kumar, Amit | Topoisomerase 1 is critical for the development and function of thymus | Jawaharlal Nehru Centre For Advanced Scientific Research |
| Madhavan, Niharika | Improving Creutzfeldt-Jakob disease (CJD) Diagnosis and Understanding the Microbiota-Gut-Brain Connection: A Multidisciplinary Approach in India. | SIMATS School of Engineering |
| Vishwakarma Aman | Meta-Predictor: A machine learning tool to predict the phenotype from a given genotype of mutation in MPS-IVA | Institute of Bioinformatics and Applied Biotechnology, Bengaluru, India |

| Name (Last name, First name) | Title | Affiliation/Institute name |
|---|---|---|
| Markose, Annsmol | The Spectrum of Succinic Semialdehyde Dehydrogenase Deficiency (SSADH)- Largest Case Series from India | Indira Gandhi Institute of Chid Health |
| Digumarthi V S Sudhakar | Genetic dissection of antenatally detected fetal abnormalities: uncovering promising candidate genes | ICMR- National Institute for Research in Reproductive and Child Health |
| Kumar, Jeevan | Clinical and molecular insights into Indian families with Epidermolysis Bullosa | Department of Medical Genetics, Kasturba Medical College, Manipal, Manipal Academy of Higher Education, Manipal, India |
| Singh, Nivedita | In silico modeling and binding site analysis of Cerebroside Sulfotransferase (CST) – an important therapeutic target for developing substrate reduction therapy of Metachromatic Leukodystrophy | Banaras Hindu University |
| Santhoshkumar, Rashmi | A rare case report of autosomal recessive Myotilinopathy in an Indian patient | Senior Scientific Assistant, Department of Neuropathology, NIMHANS, Bangalore |
| Rafi, Shabnam | Multigenerational Inheritance of Swyer Syndrome: Exploring Genetic Patterns in a Familial Context | Indian Institute of Science |
| Kansagara, Gaurav | The matricellular protein mindin regulates dermal fibroblast plasticity and functional heterogeneity in the autoimmune disease scleroderma | Institute for Stem Cell Science and Regenerative Medicine (inStem) |

| Name (Last name, First name) | Title | Affiliation/Institute name |
|---|---|---|
| S Divya | Unravelling the complexity of anaemia and insights into molecular biomarkers and targeted therapies | Amrita Vishwa Vidyapeetham, Chennai |
| Amrissh | Spectrum of sickle cell anemia and thalassemia in a teaching institute of South India | Centre for stem cell research vellore CSCR |
| Prasad G, Pannaga | GAA REPEAT EXPANSION IN THE FIRST INTRON OF FGF14 CAUSING SPINOCERBELLAR ATAXIA-27B IN THE INDIAN POPULATION | National Intitute of Mental Health and Neuro Sciences |
| Rama Lakshmi Bv | Genetic Investigation & screening in the Remote areas of South India | BharathMD Foundaton |
| Panigrahi, Lokesh | Therapeutic rescue of neutrophil maturation arrest by base editing of ELANE in severe congenital neutropenia | Centre for Stem Cell Research (CSCR), A Unit of inStem, Bengaluru |
| Kanimozhi, Elangovan | Pilot Study on Cytogenetics of Acute Lymphoblast(ALL) – Experience from a single Laboratory | Center For Medical Genetics |
| Mohana Sundaram, Sivaraj | Thyroid hormone transporter MCT8 deficiency in Indians: A rare condition and future prospects | Indian Institute of Technology, Indore from 16th November |
| Rajan, Anagha | Comprehensive analysis of a rare type of breast cancer | Research Scholar-Amrita Vishwa Vidyapeetham Chennai |

| Name (Last name, First name) | Title | Affiliation/Institute name |
|---|---|---|
| Sreelekshmi R S | Leveraging digital polymerase chain reaction for the diagnosis of Spinal Muscular Atrophy: Identification of causative mutations in SMN1 gene and assessment of copy number of SMN2 gene | Tata Institute for Genetics and Society (TIGS) |
| Purushottam, Meera | Direct-to-Consumer Genetic Testing in India - a SWOT Analysis | Molecular Genetics Laboratory |
| Dr Vikas K | Clinical and Laboratory Profile of Sixty-eight (68) children with defects in mitochondrial complexes from a single tertiary care centre in Southern India. | Indira Gandhi Institute of Child Health |
| Kataria, Sunny | A novel mouse model for human systemic sclerosis | Institute for stem cell science and regenerative medicine (inStem) |
| Tripathi, Ranu | Warburg Micro Syndrome type 1: A Neuro-Ophthalmologic and Endocrine syndrome | All India Institute of Medical Sciences, Bhopal |
| Thullimelli, Beulah | Leveraging Electronic Health Records for Rare Genetic Diseases' Diagnosis and Management, Using Genetico App | Board of Genetic Counseling India (BGCI) |
| Sen, Ritoprova | Elucidating the mechanism of mitophagic alteration in mitochondrial diseases | Integrated Ph.D in Biological Sciences/ JNCASR |
| Dubey, Rahul | Role of genomic non-coding trinucleotide repeat expansion in neurological disorders | JNCASR |

| Name (Last name, First name) | Title | Affiliation/Institute name |
|---|---|---|
| Sardar, Rahila | Decoding the genetic Insights of Neonatal-Onset of Primary Congenital Glaucoma through whole exome sequencing. | Vgenomics India Private Limited |
| Dr. Atreyee Ghosh | Digital Twin: Advancing Early Diagnosis and Management of Rare Diseases through Digital Innovation | IIT Indore |
| Sacikala | A rare mosaic case of deletion Xq in a female child suspected of Hemophilia | Center for Medical Genetics |
| Kumar, Sujith | Clinical and Genetic Correlates of Orofaciodigital Syndrome VI - A Case Study | MS Ramaiah Medical College |

# SNAPSHOTS: POSTER PRESENTATIONS