February 22, 2022

RARE DISEASE DAY
at Nationwide Children's

Program

4 pm — Pre-Program Breakout Sessions

1. Paint Party I – Acrylic
2. Paint Party II – Watercolor
3. Superhero Stories & Songs with Magical Adventures
4. Moderated Chat: Patients - Teens & Young Adults
5. Moderated Chat: Parents, Grandparents & Caregivers
6. Moderated Chat: Providers and Research Professionals

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5 pm — Welcome and Opening Remarks

Keynote Address: Linking Patients, Families & Physicians: The DADA2 Experience
Chip Chambers, MD
Assistant Clinical Professor of Surgery
Vanderbilt University
Founder, DADA2 Foundation

Research: You Will Be Found: The Search for Answers in Rare Disease
Peter White, PhD
Senior Director, Institute for Genomic Medicine
Nationwide Children's Hospital

Advocacy: Rare Diseases Care in Public Health
Murugu Manickam, MD, MPH, FACMG
Section Chief, Genetic and Genomic Medicine
Nationwide Children's Hospital

Patient Support: Supporting Your Child's Coping with a Rare Disease
Alana Goldstein-Leever, PsyD
Pediatric Psychologist
Nationwide Children's Hospital

6:20 pm — Break

6:30 pm — Patient and Family Discussion Panel
Joselin Linder – Moderator
Chris Camboni
Alex Demedeiros
Madison Evans
Eszter Hars, PhD
Demi Montgomery
Emily Neu

7:25 pm — Closing Remarks

Chip Chambers, MD

Dr. Chip Chambers is the former Chief of Endocrine Surgery at Vanderbilt University Medical Center in Nashville, Tennessee USA, where he continues to hold his clinical faculty appointment. Dr. Chambers' two children were diagnosed with the rare disease Deficiency of Adenosine Deaminase 2 or DADA2 in March 2014, just one month after the first articles describing the disease were published in the New England Journal of Medicine. As a physician and educator with years of experience in the academic, clinical, and business side of healthcare, Dr. Chambers is unusually well prepared to lead the DADA2 Foundation that forges productive and innovative partnerships between clinicians, researchers, biotech, pharma and most importantly patients. He is a compelling advocate for rare disease patients, a tireless networker, and the co-author of multiple scientific papers on DADA2. Dr. Chambers also worked for 12 and a half years at HealthSpring, one of the nation's exclusively Medicare Advantage plans, which was acquired by Cigna in 2012 and went on to be named Cigna Medicare.

Alana Goldstein-Leever, PsyD

Dr. Alana Goldstein-Leever is a pediatric psychologist at Nationwide Children's Hospital and a clinical assistant professor at the Ohio State University. Dr. Goldstein-Leever is the team psychologist for the Department of Rheumatology and the Neuroimmunology Clinic within the Department of Neurology. She provides psychological services to children, adolescents, and young adults with rheumatic diseases, as well as autoimmune and neuro-inflammatory disorders. Dr. Goldstein-Leever is engaged in clinical research focused on pediatric pain management and serves as a clinical supervisor across the internship and fellowship programs within the Department of Psychology.

Joselin Linder

Joselin Linder is a writer and author. Her work has appeared in The New York Times and The New York Post. Her memoir, The Family Gene, published by HarperCollins explores the history of medical genetics and the unnamed genetic disease only found in fourteen members of her family. The New Yorker called the book, "Surprisingly Buoyant," a phrase she hopes to have stitched on a t-shirt. You can usually find her geeking out about science and/or her three dogs: Dee Dee, Orson, and Orson's tail.

Patient and Family Panelists

Chris Camboni

Alex Demedeiros

Madison Evans

Eszter Hars, PhD

Demi Montgomery

Emily Neu

Murugu Manickam, MD, MPH, FACMG

Murugu Manickam, MD, MPH, FACMG, is a clinical geneticist/genomicist at Nationwide Children's Hospital as an associate professor of Clinical Pediatrics, with a joint appointment at the Wexner Medical Center at The Ohio State University Medical Center. Dr. Manickam received his medical degree from the Royal College of Surgeons in Ireland, completed a residency in internal medicine-pediatrics at Case Western/MetroHealth Medical Center in Cleveland and completed a fellowship in clinical genetics at the University of North Carolina, Chapel Hill. He is board certified in internal medicine and clinical genetics and received an MPH from UNC-Chapel Hill.

His specialty clinical interests are Down Syndrome and Neurofibromatosis but sees many rare clinical disorders. Additionally, he is a national expert in secondary findings from clinical testing and preventative/anticipatory care for rare disorders. Other areas of interest are the societal and ethical considerations of genetic testing and health disparities with genetics. He has been an invited lecturer and moderator at national and international meetings on the genetics and genomics.

Peter White, PhD

Peter White, PhD, is a genomics scientist and innovator, driven to create a future where no child with genetic disease goes undiagnosed and every child with cancer receives optimal treatment based upon their personal genome sequence. He currently has the position of Senior Director in The Institute for Genomic Medicine at Nationwide Children's Hospital, one of America's largest not-for-profit freestanding pediatric health care systems. He also has the appointment of tenured Associate Professor of Pediatrics in the School of Medicine at The Ohio State University.

He directs the Computational Genomics Group, leading a team of 30 high performing bioinformatics scientists, data scientists, software engineers and developers. He has substantial expertise in cloud computing and its applications in a combined healthcare and research setting. Leveraging the flexibility of the cloud, his group develops highly optimized solutions to address the substantial processing, networking and big data challenges arising from genomic science.

Peter received his PhD in Molecular Biology from the University of Cambridge, England and completed his postdoctoral training in the Department of Genetics at The University of Pennsylvania, Philadelphia. He has over 20 years of experience in the field of genomics and computational biology, is the recipient of multiple awards from the National Institutes of Health and has authored over 80 peer reviewed publications with over 14,000 citations.

Thank you!

Rare Disease Day at Nationwide Children's is made possible through generous support from:

The Jeffrey Modell Foundation for Primary Immunodeficiencies Diagnostic and Research Center at Nationwide Children's Hospital

Division of Allergy and Immunology, Nationwide Children's Hospital

Division of Genetics and Genomic Medicine, Nationwide Children's Hospital

Sanofi Genzyme - MPS I Registry

Presenters

Chip Chambers, MD
Alana Goldstein-Leever, PsyD
Murugu Manickam, MD, MPH, FACMG
Peter White, PhD

Patient and Family Discussion Panelists

Joselin Linder - Moderator
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Eszter Hars, PhD
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Emily Neu

Activity Breakout Group Hosts

Tami Burgett, RN
Alessandra Derick
Mark Drew, PhD
Shani Grove
Jackie Maher, RN
Katy Oberle
Diana Sumano Vicente
Paint the Town
Magical Adventures

Thank you for permission to use the following videos

Christina and Charlotte Loccke – The Land of Rare Diseases!
Flying Horse Farms – 500 miles
CARRA – Research is Hope

Planning Committee Leadership

Vidya Sivaraman, MD	Co-Chair	Rheumatology
Mari Mori, MD	Co-Chair	Genetics and Genomic Medicine
Joanne Drew, MFA	Co-Chair	Rheumatology
Roshini Abraham, PhD		Diagnostic Immunology Laboratory
Stacy Ardoin, MD MS		Rheumatology
Mitchell Grayson, MD		Allergy and Immunology
Dan Koboldt, MS		Institute for Genomic Medicine
Kim McBride, MD		Genetics and Genomic Medicine
Elizabeth Varga, MS, LGC		Institute for Genomic Medicine

Planning Committee

Dawn Allain, MS, LGC	OSU Genetic Counselling Graduate Program
Kevin Flanigan, MD	Center for Gene Therapy
Jesse Hunter, PhD	Institute for Genomic Medicine
Mahmoud Kallash, MD	Nephrology
Katelin Krivchenia, MD	Pulmonology
Peter Mustillo, MD	Allergy and Immunology
Rachel Notestine	Institute for Genomic Medicine
Hiren Patel, MD	Nephrology
Benjamin Prince, MD	Allergy and Immunology
Melissa Rose, DO	Hematology and Oncology
John Spencer, MD	Nephrology
Rachel Williamson	Institute for Genomic Medicine

Technical and A/V Support

Angel Ramona Hatfield	Hematology and Oncology
Mike Hettinger	RISI

Marketing

Catherine Roebuck	Marketing

Community Education

Marcie Rehmar	Community Education

This event would not have been possible without the teamwork and support from the Division of Rheumatology.

#RAREDISEASEDAY

RAREDISEASEDAY.ORG

EQUITY
FOR PEOPLE LIVING WITH A RARE DISEASE

WHAT DOES EQUITY MEAN FOR PEOPLE LIVING WITH A RARE DISEASE?

Equity in practice means meeting people's specific needs and **eliminating barriers preventing their full participation in society.** For people living with a rare disease equity means social opportunity, non-discrimination in education and work, and equitable access to health, social care, diagnosis and treatment.

On Rare Disease Day we call for action for people living with a rare disease to have equal opportunities to realise their **full participation in family, work and social life.**

The long-term goal of Rare Disease Day over the next decade is increased equity for people living with a rare disease and their families.

THE CHALLENGE

The **300 million people living with a rare disease around the world** and their families face common challenges in their daily lives. As a vulnerable and neglected population they are disproportionally affected by stigma, discrimination and social marginalization, within their social environment as well as society at large.

There are **over 6000 rare diseases** that are chronic, progressive, degenerative, disabling and frequently life threatening. Due to the rarity of each individual disease and scattered populations, expertise and information is scarce. In health and support systems designed for common diseases people living with a rare disease face inequities in accessing diagnosis, care and treatments.

People living with a rare disease also **face discrimination at work, school and leisure.** For instance in a EURORDIS Rare Barometer survey on 'Juggling care and daily life: The balancing act of the rare disease community' 94% of the respondents who reported difficulties in accessing higher education declared that the disease has limited their professional choices.

KEY STATISTICS OF RARE DISEASES

#SOCIALOPPORTUNITY

HOW CAN WE ACHIEVE EQUITY FOR PEOPLE LIVING WITH A RARE DISEASE?

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1 ADVOCATING FOR *SOCIAL INCLUSION* FOR PEOPLE LIVING WITH A RARE DISEASE

In order to achieve equitable social inclusion for people living with a rare disease, they must first have access to holistic care covering the 360° spectrum of health, social and everyday needs as argued in a EURORDIS position paper on Achieving Holistic Person-Centred Care to Leave No One Behind.

The UN 2030 Agenda and its Sustainable Development Goals (SDGs) provide an important framework for addressing the full spectrum of needs of people living with a rare disease. The Goals target important issues including education, gender, work and inequality. **Addressing the needs of people living with a rare disease is central to achieving the UN 2030 Agenda,** the SDGs and its pledge to leave no one behind. Global institutions protecting human rights seek to address the health care challenges of people living with a rare disease. The Human Rights Council Resolution on access to medicines and vaccines recognises the importance of development, access and affordability of treatments for rare diseases.

The following legislation promotes social inclusion for people with a disability, including those whose disability is a result of living with a rare disease: The **UN Convention on the Rights of People with Disabilities (CRPD)** encourages governments to develop and implement policies and practices targeting the most marginalized groups of persons with disabilities such as those living with a rare disease.

2 INCLUDING RARE DISEASES IN *UNIVERSAL HEALTH COVERAGE* TO LEAVE NO ONE BEHIND

Universal Health Coverage (UHC) is a safeguard of equity and people living with a rare disease need to be included in national strategies for UHC and essential health service packages. **UHC ensures all people, everywhere, can access the quality essential health services they need without being exposed to financial hardship.** UHC includes three dimensions of coverage: in terms of population, services and proportion of costs covered.

UHC is firmly rooted in the right to health and requires strong political leadership. In September 2019 all 193 UN Member States adopted the Political Declaration on UHC, which includes the need to strengthen efforts to address rare diseases. While countries around the world are at different stages in the development of policies in support of the rare disease community, **any country can today take decisive action to support a greater integration of rare diseases in its UHC model** or approach. It is possible to make great progress at a rapid pace even if starting from very little.

Governments can focus on 1) extending coverage to people living with a rare diseases by promoting visibility, codification and diagnosis; 2) Including other services and adapting existing ones to the needs of the rare disease population; and 3) Protecting the rare disease population from further financial hardship.

RARE IS *MANY* RARE IS *STRONG* RARE IS *PROUD*

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A UNITED NATIONS GENERAL ASSEMBLY RESOLUTION ON PERSONS LIVING WITH A RARE DISEASE

To ensure equity for people living with a rare disease, it is necessary for countries to collectively **promote measures that are multidisciplinary, holistic and person-centred**, and that ensure non-discrimination and opportunities to contribute to society.

To ensure these whole-of-society measures, the rare disease community, represented by the NGO Committee for Rare Diseases, Rare Diseases International and EURORDIS, are calling on UN Member States to **adopt a United Nations General Assembly Resolution on persons living with a rare disease and their families.**

A UNGA Resolution would promote global collaboration, national strategies and policies, as well as the inclusion of people living with a rare disease in the work of the United Nations and its agencies and bodies.

LEAVE NO ONE BEHIND!

MILESTONES IN ACHIEVING EQUITY

1983 – Us Orphan Drug Act

1999 – EU orphan drug regulation

2008 – First Rare Disease Day

2016 – UN High Level meeting, Inauguration of the NGO Committee for rare diseases

2017 – European Referece Networks launched!

2019 – Rare Disease Day policy event at the UN

Rare diseases mentioned by United Nations Office of the High Commissioner for Human Rights (OHCHR) and in the political declaration on universal health coverage.

2020 – Rare diseases are recognised as a vulnerable population within the first report of the 'State of commitment of UHC'

2021 – The Executive Board of the WHO agrees to provide guidance on expanding access to effective treatments rare and orphan diseases.

HOW CAN I *ADVOCATE* FOR EQUITY?

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1. Write a *LETTER TO A KEY POLICY-MAKER* in your community to highlight the importance of addressing the needs of people with a rare disease and share this fact sheet with them.

2. Hold a *RARE DISEASE DAY ONLINE EVENT* that highlights a key local or national policy issue for the rare disease community and invite a decision-maker to attend.

3. Use *MEDIA ATTENTION* on Rare Disease Day to launch a new initiative for the rare disease community.

KEY *RESOURCES* AND INFORMATION

ABOUT RARE DISEASES

- Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database, European Journal of Human Genetics, by EURORDIS-Rare Diseases Europe, Orphanet & Orphanet Ireland.
- EURORDIS press release: New scientific paper confirms 300 million people living with a rare disease worldwide
- What is a rare disease webpage (on rarediseaseday.org)
- Rare Disease Day 2020 Infographics
- Dr Tedros tweet 28.02.19
- Opening Remarks by H.E. Mr. Sven Jürgenson, Permanent Representative of Estonia at the Rare Disease Day Policy Event, New York, 21 February 2019

THE IMPACT OF RARE DISEASES ON DAILY LIFE

- EURORDIS position paper: Achieving Holistic Person-Centred Care to Leave No One Behind
- Rare Barometer survey report on 'Juggling care and daily life: The balancing act of the rare disease community'
- Rare Barometer survey infographic on 'Juggling care and daily life: The balancing act of the rare disease community'
- Disability and rare disease: towards person-centred care for Australians with rare diseases, The McKell Institute
- Rare Barometer survey on rare disease patients' experience of COVID-19

HUMAN RIGHTS OF PEOPLE LIVING WITH A RARE DISEASE

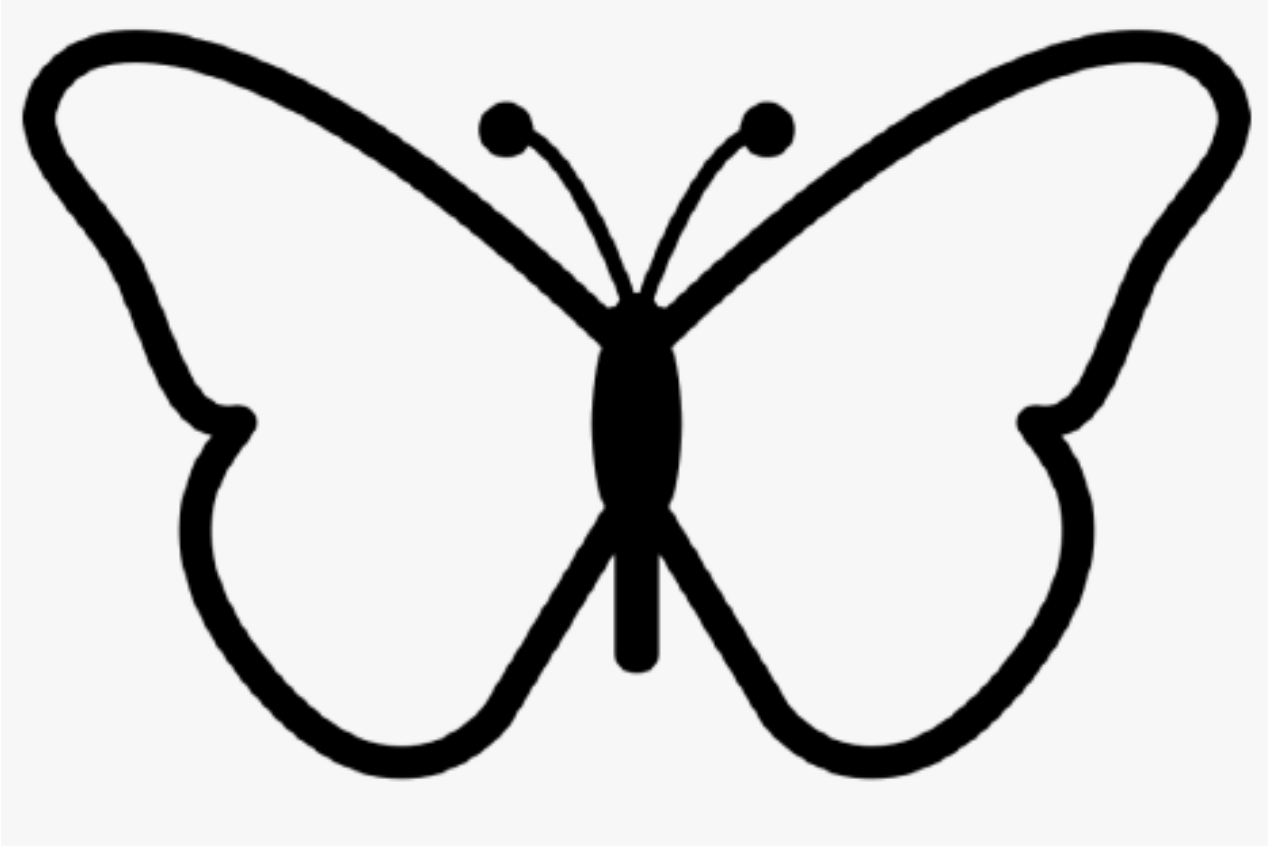
- Human Rights Council Resolution on access to medicines and vaccines
- UN Convention on the Rights of People with Disabilities
- Report of the Special Rapporteur on the rights of persons with disabilities to 73rd Session of the UNGA.

RARE DISEASES IN THE SUSTAINABLE DEVELOPMENT GOALS AND UNIVERSAL HEALTH COVERAGE

- Rare Diseases International position paper: Universal health coverage
- Universal Health Coverage campaign toolkit
- State of UHC implementation report
- UHC for Rare Diseases Campaign
- Development of the roadmap on access to medicines and vaccines 2019-2023
- News article, RDI: United Nations human rights body stresses the need to address rare diseases within Universal Health Coverage
- EURORDIS press release: UN Member States include rare diseases in political declaration on universal health coverage
- NGO Committee for rare diseases press release: Rare Disease Day 2019 Policy Event at the United Nations
- NGO Committee for rare diseases event report: Rare Disease Day 2019 Policy Event at the United Nations
- NGO Committee for rare diseases event report: The Right to Health: The Rare Disease Perspective, Rare Diseases International Policy Event 2017
- NGO Committee for rare diseases event report: Global Gathering for Rare Diseases: Inauguration of the NGO Committee for Rare Diseases 2016

Alone We Are Rare.

Together We Are Strong.



Share with us using the hashtag **#RareDiseaseDayNCH** on social media!

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Benefiting NATIONWIDE CHILDREN'S

When your child needs a hospital, everything matters.

Image from ultracoloringpages.com

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Show Your Rare

Email questions and comments to RareDiseaseDay@NationwideChildrens.org

Tell us the story of you at Flutter.NationwideChildrens.org

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