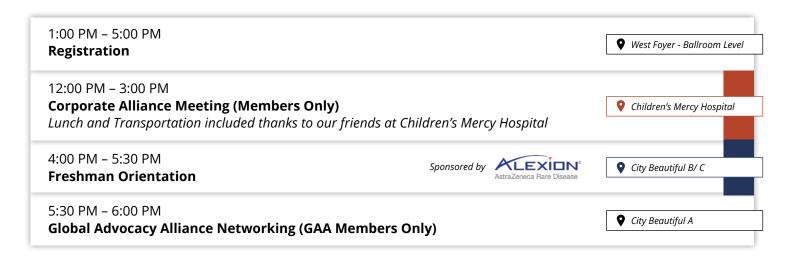
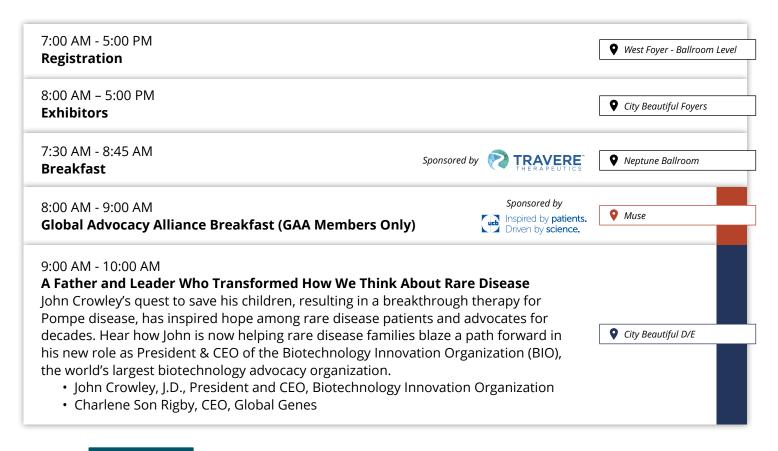
# **AGENDA**

# Membership Meetings - Wednesday, September 25, 2024



# RARE Advocacy Summit - Thursday, September 26, 2024



10:00 AM

# RAREly Told Stories film screening: "Rarely Have You Seen a Fox Like This" by Claudia Parker, Author, FOXP1 Parent and Advocate

City Beautiful D/E

10:15 AM - 11:15 AM

**Track Sessions and Expert Office Hours** 

#### 10:15 AM – EMPOWERING THE RARE INDIVIDUAL

#### Sponsored by

**YLEXION** 

#### **Living with Visible and Invisible Disabilities**

Explore how to take back your power with tips for allies, what to do when people miss the mark, and understanding "disabilities as dynamic".

- Moderator: Mackenzie Abramson, Senior Manager, Research Program Communications, Global Genes
- Kendra Gottsleben, Marketing Communication Specialist, Center for Disabilities, Sanford School of Medicine, University of South Dakota
- Richie Kahn, Clinical Researcher, Co-founder and COO, Canary Advisors
- Soniya Vadher-Patel, Social Media Influencer, Entrepreneur and Mental Health Advocate

# 10:15 AM - COMMUNITY AND CAPACITY BUILDING

#### Sponsored by

global rare diseases 🧗

# Think Like a Start-Up: Grow your Patient Advocacy Group

Discover principles of fast organization building: how to create scalable and sustainable models, collect metrics, learn, adapt and adopt legal structures to protect your IP.

- Moderator: Charlene Son Rigby, CEO, Global Genes
- Michael Hund, CEO, EB Research Partners
- Sunitha Malepati, Founder, Buffalo Initiative and Vice President, CACNA1A Foundation
- Mark Wilhelm, Partner, Troutman Pepper

#### Sponsored by

# Know (and Build) Your Strength as a Research Partner

10:15 AM – GROWING YOUR ORGANIZATION'S RESEARCH READINESS

Join a hands-on workshop featuring the FasterCures Research Partnership Maturity Model. Learn about the four domains of research capacity, and identify next steps to increase your organization's partnership value.

• Facilitator: Raymond Puerini, Associate Director, FasterCures

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#### - 11

10:15 AM - EXPERT OFFICE HOURS

# Expert Office Hours - By Appointment Only

Attendees can meet one-on-one with leaders, industry members, researchers, and healthcare professionals to gain insights on topics like building capacity, advancing research, supporting communities, and avoiding costly detours. These 15-minute sessions are reserved in advance through the Attendee Hub event app.



11:15 AM - 11:30 AM

**Session Transition / Break** 

City Beautiful Foyers

City Beautiful B/C

City Beautiful D/E

11:30 AM - 12:30 PM

# **Track Sessions and Expert Office Hours**

#### 11:30 AM – EMPOWERING THE RARE INDIVIDUAL

Sponsored by

**KLEXION** 

# Help for the Rare Caregiver

Helping the helper is essential. We'll look at supports that provide the most benefit as well as research that shows what works, and provide actionable advice to ease caregiver burdens.

- Moderator: Megan Nolan, Founder, Rare Parenting
- Daniel DeFabio, Director, Community Engagement, Global Genes
- Bridgette Kelleher, Ph.D., Associate Professor, Psychological Services, Purdue University
- Bret Koncak, Rare Dad, Co-founder, mejo

# 11:30 AM - COMMUNITY AND CAPACITY BUILDING

Sponsored by

## **Activate Your Community**

Get expert tips on building connections, attracting and managing volunteers, creating lieutenants and outreach through community and corporate partnerships.

- Moderator: Effie Parks, Rare Mom; Podcast Host, Once Upon a Gene
- Lara Bloom, President and CEO, Ehlers Danlos Society
- Samantha Charleston, Director, Programs and Community Engagement, Orphan Disease Center, University of Pennsylvania
- · Rob Long, Executive Director, Uplifting Athletes
- · Luke Rosen, Co-Founder, KIF1A.org

# →Chiesi global rare diseases 🧖

# 11:30 AM - GROWING YOUR ORGANIZATION'S RESEARCH READINESS

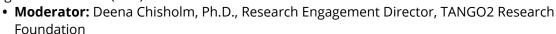
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#### **Unlocking the Future: The Role of Natural History Studies**

Hear about best practice and case studies for improving patient outcomes, uncovering insights on disease and treatment, and identifying what is acceptable to the U.S. Food and Drug Administration (FDA).



- · Lea Ann Browning-McNee, Director of Communication and Engagement, Reagan-Udall Foundation for the U.S. FDA
- Elizabeth Cassidy, Research Project Manager, Tuberous Sclerosis Alliance
- Kasha Morris, Co-Founder, TANGO2 Research Foundation

# 11:30 AM - EXPERT OFFICE HOURS

# **Expert Office Hours - By Appointment Only**

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12:30 PM - 1:50 PM

**Lunch and Poster Presentations** 





2:00 PM - 3:00 PM

**Track Sessions and Expert Office Hours** 

#### 2:00 PM - EMPOWERING THE RARE INDIVIDUAL

Sponsored by

**ALEXION** 

#### Medical Gaslighting and Post Traumatic Stress Disorder (PTSD)

Medical outliers are sometimes doubted or dismissed. Learn what gaslighting is, what to do about it, and the impact of PTSD.

- **Moderator:** Mackenzie Abramson, Senior Manager, Research Program Communications, Global Genes
- · Amanda Griffith-Atkins, Marriage and Family Therapist, Founder, Amanda Atkins Counseling Group
- Carla D'Imperio, Family Support Specialist, Phelan-McDermid Syndrome Foundation
- Suzanne Edison, Mental Health Coordinator, CureJM

# 2:00 PM - COMMUNITY AND CAPACITY BUILDING

Sponsored by

#### **Understand Newborn Sequencing and Screening (NBS)**

NBS can be a path to both diagnosis and new members. Learn why genetic diagnosis is key, how to create infrastructure to support diagnosis, and how to insure access to services and equity.



- **Moderator:** Annie Kennedy, Chief of Policy, Advocacy and Patient Engagement, EveryLife Foundation for Rare Diseases
- Wendy Chung, M.D., Ph.D., Chief of Pediatrics, Boston Children's Hospital
- Stacey Seeloff, Director, Strategic Alliances, Rady Children's Institute for Genomic Medicine
- Brooke Thomas, Chief Programs Officer, Alabama Rare

#### 2:00 PM - GROWING YOUR ORGANIZATION'S RESEARCH READINESS

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#### A Guide to Biomarkers and Outcome Measures

How are biomarkers and outcome measures different? How can biomarkers be used as surrogate endpoints? And what is the role of Patient Advocacy Groups in identifying them?



- Pat Furlong, Founding President and CEO, Parent Project Muscular Dystrophy
- Celia Goeldner, Clinical Expert Team Leader, Clinical Scientist, Roche

# 2:00 PM - EXPERT OFFICE HOURS

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## **Expert Office Hours - By Appointment Only**

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3:00 PM - 3:15 PM

**Session Transition / Break** 

City Beautiful Foyers

3:15 PM - 4:15 PM

#### **Track Sessions and Expert Office Hours**

#### 3:15 PM - EMPOWERING THE RARE INDIVIDUAL

Sponsored by

### My Genetic Test Says VUS: What's Next?

What does "variants of unknown significance (VUS)" mean? When and how do you pursue reanalysis? Are these variants different for marginalized populations? Find out.

- Moderator: Abigail Sveden, Genetic Counselor, Boston Children's Hospital
- Dustin Baldridge, M.D., Ph.D., Assistant Professor, Washington University in St. Louis
- Christina Gurnett, M.D., Ph.D., Director, Division of Pediatric and Developmental Neurology, Washington University in St. Louis
- Michele Herndon, Program Director, Undiagnosed Diseases Network Foundation

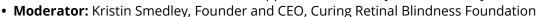


#### 3:15 PM - COMMUNITY AND CAPACITY BUILDING

Sponsored by

#### **Disability and Access**

This panel will focus on best practices for ADA website compliance, accessibility for research and clinical trials, and how best to support the needs of your community.



- · Rachelle Cook, Rare Disease Advocate
- Kendra Gottsleben, Marketing Communication Specialist, Center for Disabilities, Sanford School of Medicine, University of South Dakota
- · Lori Renna Linton, Rare Disease Advocate and Filmmaker



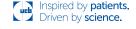
#### 3:15 PM - GROWING YOUR ORGANIZATION'S RESEARCH READINESS

Sponsored by

#### **Quick Wins & Long-Term Investments in Data**

Use the power of data to make a difference for patients and improve life now for your community members. Maximize users for broadest impact, move the needle with inhome collection and use data to create and maintain clinical guidelines.

- **Moderator:** Zohreh Talebizadeh, Ph.D., Senior Director, RARE-X Research Program, Global Genes
- Wendy Chung, M.D., Ph.D., Chief of Pediatrics, Boston Children's Hospital
- · Marianne Clancy, Executive Director, Cure HHT
- Jennifer Foss-Feig, Ph.D., Principal Investigator, Foss-Feig Lab, Icahn School of Medicine, Mount Sinai



#### 3:15 PM – EXPERT OFFICE HOURS

Sponsored by

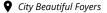
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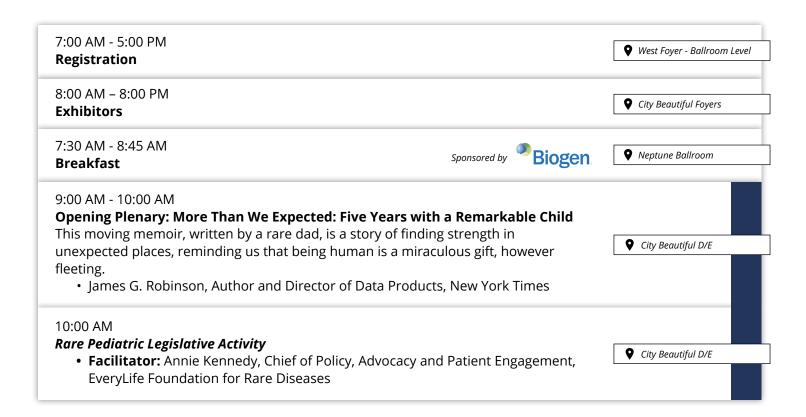
4:15 PM - 4:30 PM

**Session Transition / Break** 





# RARE Advocacy Summit - Friday, September 27, 2024



City Beautiful D/E

10:15 AM - 11:15 AM

## **Track Sessions and Expert Office Hours**

### 10:15 AM – EMPOWERING THE RARE INDIVIDUAL

Sponsored by

#### **Rare Siblings**

There are unique concerns around siblings to someone who has a rare disease. Parents often give them less attention or focus. The siblings feel they must do more than their typical peers due to the medical care needs in their home. We'll hear from rare siblings on how to manage the unique role.

- Moderator: Noah Seidman, Rare Sibling
- Matt Flesch, Vice-President, Communications & Patient Advocacy, Amgen
- · Felix Townsin, Rare Sibling



# 10:15 AM - COMMUNITY AND CAPACITY BUILDING

Sponsored by

### **Empowering Advocacy: Federal, State and Patient Insights**

Join the EveryLife Foundation for Rare Diseases for an update on advocacy opportunities at the state and federal levels. Hear directly from advocate panelists for tips and tools to take home.



- · James Griffin, Advocate
- · Paloma Juarez, Advocate
- Stephanie Riordan, Director of Patient Programs, EveryLife Foundation for Rare Diseases
- Shannon von Felden, Senior Director of Advocacy, EveryLife Foundation for Rare Diseases
- · Anneliese Williams, Advocate

#### 10:15 AM - GROWING YOUR ORGANIZATION'S RESEARCH READINESS

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### Overcoming the "Valley of Death" in Research

Navigating from bench to bedside is one of the greatest challenges rare disease leaders face. What do PAGs need to do to de-risk drug development for partners? What internal and external expertise can you tap? Is venture philanthropy an option?

- Moderator: Danny Levine, Life Sciences Writer, Global Genes
- Sharie Haugabook, Ph.D., Senior Project Manager and Drug Development Lead,
   Therapeutics for Rare and Neglected Disorders (TRND), National Institutes of Health (NIH)
- · Christopher Hopkins, Ph.D., CEO, Devine Bio
- · Michael Hund, CEO, EB Research Partnership
- · Yael Weiss, Ph.D., CEO, Mahzi Therapeutics

#### 10:15 AM - EXPERT OFFICE HOURS

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# **Expert Office Hours - By Appointment Only**

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11:15 AM - 11:30 AM

**Session Transition / Break** 

City Beautiful Foyers

11:30 AM - 12:30 PM

### **Track Sessions and Expert Office Hours**

### 11:30 AM - EMPOWERING THE RARE INDIVIDUAL

#### Sponsored by

ALEXION<sup>®</sup>

#### Your Child's Rights: IEPs and 504s

The battle to get the best Individualized Education Program agreed to and adhered to is a top complaint for rare parents. Our panel will help you understand your child's rights and make sure your child isn't offered "less than."

- Moderator: Shannon Bennett, Patient Navigator, Global Genes
- Elle Cole, Advocate, Sickle Cell Disease
- · Piper Paul, Attorney, Piper Paul Law

### 11:30 AM - COMMUNITY AND CAPACITY BUILDING

#### Sponsored by

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#### **Master Rare Disease Conference Planning**

Our panel of experts will help you develop impactful family conferences and scientific meetings, including in-person and virtual events. Find out how to identify key funding sources, create inclusive and accessible events, and maximize outreach with effective marketing. Based on a new toolkit developed by Simons Searchlight and Global Genes.

- Moderator: Erica Jones, Outreach Manager, Simons Searchlight
- Jessica Fein, MitoAction Advocate and Author, Breath Taking: A Memoir of Family, Dreams and Broken Genes
- Ashley Point, President, Koolen-De Vries Syndrome Foundation
- · Michele Whiteside, Director, Programs and Events, Global Genes

#### Sponsored by

# **Looking for the Common in Rare**

Hear case studies from industry and advocacy. Our panelists will be discussing how each collaboration evolved, what the outcomes were, and factors that led to success.

- Moderator: Danny Levine, Life Sciences Writer, Global Genes
- Simon Frost, CEO and Co-founder, Tiber Capital Group

11:30 AM - GROWING YOUR ORGANIZATION'S RESEARCH READINESS

- Karmen Trzupek, Senior Director, Scientific Programs, Global Genes
- Joni Rutter, Ph.D., Director, National Center for Advancing Translational Sciences (NCATS), National Institutes of Health (NIH)



#### 11:30 AM - EXPERT OFFICE HOURS

#### **Expert Office Hours - By Appointment Only**

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12:30 PM - 1:50 PM

**Lunch and Poster Presentations** 

Sponsored by Pharming

Neptune Ballroom

2:00 PM - 3:00 PM

#### **Track Sessions and Expert Office Hours**

# 2:00 PM - EMPOWERING THE RARE INDIVIDUAL

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**KLEXION** 

#### **Career Considerations for Rare Adults**

Overcome common challenges faced by rare adults in the workforce such as dealing with prejudice, asking for medical accommodations, and when/how to disclose a health

- Moderator: Mackenzie Abramson, MPH, Senior Manager, Research Programs Communications, Global Genes
- Dakota Fisher Vance, Global Patient Advocacy Associate Director, BioCryst Pharmaceuticals
- Kendra Gottsleben, Marketing Communication Specialist, Center for Disabilities, Sanford School of Medicine, University of South Dakota
- · Lori Renna Linton, Rare Disease Advocate and Filmmaker

# 2:00 PM - COMMUNITY AND CAPACITY BUILDING

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#### Working Together for Advocacy, Research and Therapies

Hear about a collaboration between GeneDx, Akouos and Hands & Voices that is breaking down silos for patients and families.



- Moderator: Gay Grossman, Patient Advocacy and Engagement Lead, GeneDx
- Alan Gilstrap, Senior Director, Advocacy and Engagement, Akouos
- Amanda Singleton, Clinical Lead, Biopharma Partnerships, GeneDx
- Janet DesGeorges, Executive Director, Hands & Voices

# 2:00 PM - GROWING YOUR ORGANIZATION'S RESEARCH READINESS

Sponsored by

#### **Using AI to Improve Diagnosis**

Learn how researchers are using AI and real world data to detect rare diseases earlier, clarify diagnoses and find solutions for the undiagnosed.

- Moderator: Danny Levine, Life Sciences Writer, Global Genes
- Erik A. Feingold, CEO and Co-founder, FDNA
- Melissa Haendel, Ph.D., Director of Precision Health and Translational Informatics, University of North Carolina, Chapel Hill
- · Kat Schmolly, M.D., Founder, zebraMD

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# 2:00 PM - EXPERT OFFICE HOURS

# **Expert Office Hours - By Appointment Only**

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3:15 PM - 3:30 PM

**Session Transition / Break** 



• City Beautiful Foyers

3:15 PM - 4:15 PM

# **Track Sessions and Expert Office Hours**

#### 3:15 PM – EMPOWERING THE RARE INDIVIDUAL

Sponsored by

# Astra Zenaca Bare Disease

#### **RAREly Told Stories Workshop**

This hands-on workshop presented by Global Genes and The Disorder Channel covers the basics of filmmaking, editing, principles of storytelling, and what's unique to rare disease films.

- Daniel DeFabio, Director, Community Engagement, Global Genes
- Claudia Parker, Rare Mom, Entrepreneur, Filmmaker

# 3:15 PM - COMMUNITY AND CAPACITY BUILDING

Sponsored by



# **Innovative Funding Strategies**

We've gathered a stand-out panel of leaders to share their novel models of funding – and brainstorm revolutionary new approaches.

- Moderator: Megan Golden, Co-Founder and CEO, Mission: Cure
- Sunitha Malepati, Vice President, CACNA1A Foundation and Founder, Buffalo Initiative
- Bina Maniar Shah, Founder, CEO and President of the Board of Directors, Project 8p Foundation

#### 3:15 PM - GROWING YOUR ORGANIZATION'S RESEARCH READINESS

Sponsored by

#### Is Our Community Ready for Gene Therapy?

Find out how advocacy groups, researchers and industry use the GENE TARGET framework to determine readiness for gene therapy, identify gaps in the translational research pipeline and prioritize funding and research efforts.

- Moderator: Shruti Mitkus, Ph.D., Director, Patient Services, Global Genes
- Raymond Belanger-Deloge, Translational Genomic Medicine Specialist, Rosamund Stone Zander Translational Neuroscience Center, Boston Children's Hospital
- Matt Fuller, Ph.D., Executive Director, Gene Therapy Research, Ultragenyx
- · Ashley Point, President, Koolen De-Vries Syndrome Foundation

# Inspired by patients. Driven by science.

### 3:15 PM - EXPERT OFFICE HOURS

ponsored by

# **Expert Office Hours - By Appointment Only**

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4:15 PM - 4:30 PM

**Session Transition / Break** 

• City Beautiful Foyers

4:30 PM - 5:30 PM

#### Creating a Global Ecosystem for Rare Disease Knowledge

How are advancements in data science making this a hopeful time for the rare disease community? And how can we bring together the macro and micro to accomplish even more?

City Beautiful D/E

- Moderator: Charlene Son Rigby, CEO, Global Genes
- John Wilbanks, Senior Fellow, Datasphere Initiative
- Peter Goodhand, CEO, Global Alliance for Genomics and Health

5:30 PM

#### RAREly Told Stories film screening: "Pass Me the Potatoes, Please"

by Lori Renna Linton, IB English teacher, Patient Advocate for HSP

City Beautiful D/E



5:30 PM - 6:30 PM

# RARE Champions of Hope Awards Ceremony

Open to all attendees

Sponsored by AMGEN 1

City Beautiful D/E

The RARE Champions of Hope Awards celebrate, honor and recognize those in the rare disease community who are raising the bar through their innovative approaches to research, programming and advocacy to create meaningful impact in the rare disease space. Awardees are beacons of hope in the rare disease community who lean in, take risks and lead collaborative efforts to positively impact the lives of rare disease patients, caregivers and families across the globe.

#### Awards will be presented in the following categories:

- RARE Champion in Advocacy (Organization)
- RARE Champion in Advocacy (Individual)
- RARE Champion in Industry
- RARE Champion in Research
- RARE Champion in Health Equity

6:30 PM - 8:30 PM

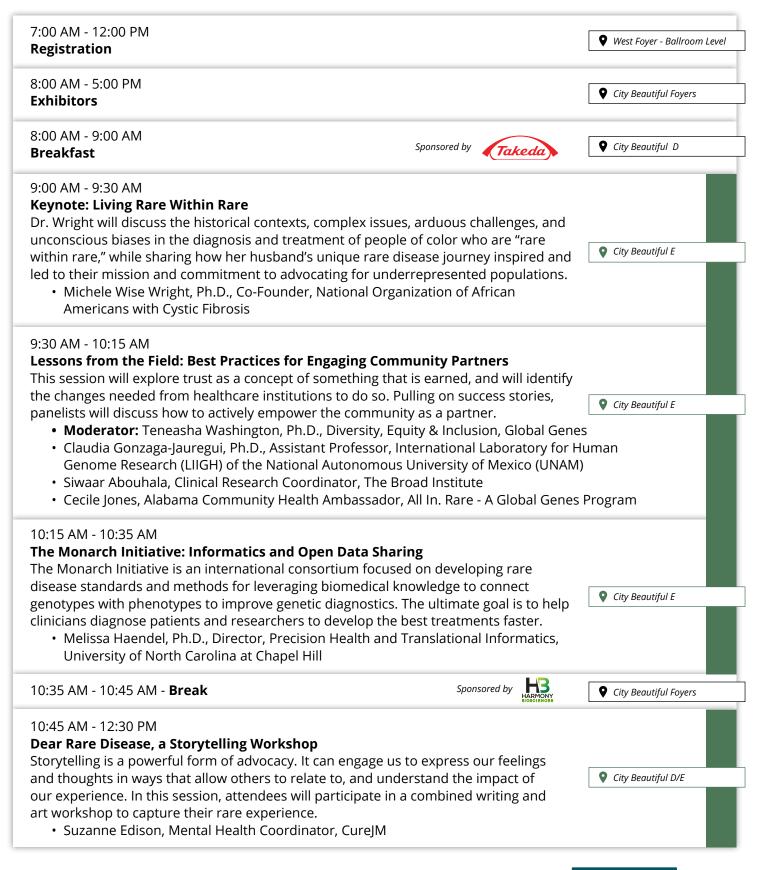
#### **RARE Champions of Hope Reception**

Open to all attendees

Sponsored by AMGE



# RARE Health Equity Forum - Saturday, September 28, 2024



# 12:30 PM - 2:30 PM Sponsored by General Sponsored by Sanofi • City Beautiful D **Lunch & Table Talks** 2:30 PM - 2:50 PM Data Driven Music: An Exploration of Rare Disease Harmony Professor Taylor will discuss music driven advocacy through demonstration of his music composition based on a catalog of 230 rare diseases that was curated by City Beautiful E Dr. Aditi Kantipuly. Twelve categories of disease correspond to the 12 Indian chakras, which provide a background harmony for each category. • Stephen Taylor, Professor of Music, University of Illinois at Urbana-Champaign 2:50 PM - 3:25 PM Film Screening: "Holding Moses" Holding Moses chronicles the heartfelt story of a queer mother and Broadway performer who learns how to parent her profoundly disabled son. This documentary folds together several areas in the DEI space including assumptions about caregivers, City Beautiful E shame narratives around disability, and LGBTQ parents. • Moderator: Daniel DeFabio, Director, Community Engagement, Global Genes Randi Rader, Featured Subject, Frankly Speaking Films Rivkah Beth Medow, Director, Frankly Speaking Films 3:25 PM - 4:10 PM Diversity in Rare Clinical Trials: What's Working and What Isn't Historically marginalized racial groups, women and other minoritized communities are notably underrepresented in clinical trials, despite efforts and investment by advocates and health equity leaders. This panel will discuss effective strategies to City Beautiful E address the lack of diversity in clinical trials. Moderator: Kim Moran, Ph.D, Senior Vice President and Head, U.S. Rare Diseases, UCB • R'Kes Starling, Founder, Reveles Clinical Research • Richie Kahn, Clinical Researcher, Co-founder and COO, Canary Advisors Tanvee Varma, M.D., Resident Physician, Brigham and Women's Hospital 4:10 PM - 5:00 PM **Rural Rare Healthcare** Rural residents often encounter barriers to healthcare including workforce shortages, health insurance status, transportation issues, health literacy, and stigma in rural communities. These disparities are even greater among historically marginalized communities and rare disease patients. Panelists will discuss the work they are doing City Beautiful E to combat these challenges. • Moderator: Shobana Kubendran, Genetic Counselor, KU School of Medicine-Wichita Ana Cohen, Ph.D., Assistant Director, Molecular Genetics Laboratory, Children's Mercy • Nadia Falah, M.D., Clinical Geneticist, Johns Hopkins All Children's Hospital • Abdallah F. Elias, M.D., Chief Medical Officer, Shodair Children's Hospital 5:00 PM - 5:15 PM City Beautiful E **Closing Remarks** 5:30 PM - 7:30 PM Genentech Sponsored by City Beautiful Foyers **Closing Reception**