

DAY 1

Wednesday 14 Sept 2022

4:30 pm - 6:00 pm

4:30 pm

Welcome

4:45 - 5:30 pm

Opening Keynote: When Rare Disease Becomes a BIG Data Opportunity

Moderator: Morrie Ruffin, Managing Partner, Adjuvant Partners; Co-founder AAIH

Panelists:

- Ryan Colburn, Pompe Patient and Innovator
- Stacie Calad-Thompson, Ph.D., BioSymetrics
- Robert Deans, Ph.D., Synthego
- Kazi Mustaqhusain, Roche

5:30 - 6:00 pm

Can Big Data Supercharge Research for 10K Rare Diseases?

- Kirk Lamoreaux, Cogenticity
- Wendy Erler, Alexion AstraZeneca, RARE-X Board Member

6:00 pm

Evening Reception Co-Hosted by RARE-X and Global Genes

DAY 2

Thursday 15 Sept 2022

7:00 am - 1:00 pm

7:00 am

Breakfast

7:30 am

Welcome

7:35 - 8:00 am

RARE-X - Leveraging What's ahead

- Charlene Son Rigby, RARE-X

8:00 - 8:45 am

The Future is Now: Realizing the Possibilities of Patient Driven Research

- Wendy Erler, Alexion AstraZeneca, RARE-X Board Member
- Betsy Bogard, FOP Advocate, Ensoma, RARE-X Board Member
- Julia Vitarello, Mila's Miracle Foundation

8:45 - 9:30 am

N-of-1 Therapies - The Promise and Opportunity

Moderator: Walt Kowtoniuk, Ph.D., RARE-X Chair, Third Rock Ventures

Panelists:

- Luke Rosen, KIF1a.org
- Tim Yu, M.D., Ph.D., Boston Children's Hospital, N=1
- Sarah Glass, Ph.D., nLorem Foundation
- Christopher Hart, Ph.D., Creyon Bio

- 9:30 - 10:00 am **RARE-X Open Science Data Challenge - Neuro + Case Study: ALS Kaggle Challenge**
Presenter: Charlene Son Rigby, RARE-X
Discussion with: Indu Navar, EverythingALS
- 10:00 - 10:45 am **Case Study: Collaborative Research Network Commission for Complex Copy Number Variants**
Moderator: Vanessa Vogel-Farley, RARE-X
 - Yssa DeWoody, Ph.D., Ring14 USA
 - Bina Shah, Project 8p
 - Scott Demarest, M.D., Colorado Children’s Hospital
- 10:45 - 11:30 am **Built to Accelerate - disease characterization, progression models and research roadmapping**
 - Karmen Trzupek, RARE-X
 - Rachel Groth, Ph.D., BridgeBio
 - Wendy Chung, M.D., Ph.D., Columbia & Simons Searchlight
- 11:30 - 12:45 pm **Innovation Roundtables [Two - 30 min sessions]**
- Practical Insights**
- Table 1: Community Engagement/Enrollment Strategies
Tom Hume, RARE-X
- Table 2: The Challenges and Alternate Models to Engage with Researchers: Scott Demarest, M.D., Colorado Children’s Hospital
- Table 3: Pre-Competitive Consortia Models to Accelerate Research and Discovery - Karmen Trzupek, RARE-X
- Table 4: Disease Concept Model: STXBP1 Case Study
Charlene Son Rigby, RARE-X
- Table 5: The Criticality of Developing a Data Governance Strategy for the Long Game - Vanessa Vogel-Farley, RARE-X
- Table 6: Citizen Science - Who, What, When, Why and How
Luke Rosen, KIF1a.org
- Mentoring**
- Table 7: Research Roadmapping - Rachel Groth, Ph.D., BridgeBio
- Table 8: From Mila to Millions- Julia Vitarello, Mila’s Miracle Foundation
- Table 9: The Importance of NH Data to Support Cell & Gene Therapy-
Wendy Chung, M.D., Ph.D., Columbia & Simons Searchlight
- Table 10: How to Partner with Industry in your NH Registry
Betsy Bogard, FOP Advocate, Ensoma, RARE-X Board Member

Cell & Gene Therapy

Table 11: Partnering With Columbus Children's Gene Therapies
Laura Hameed, Columbus Children's Foundation

Table 12: Research & Development Opportunities and How to Get Funding: Lila Collins, Ph.D. & Uta Grieshammer, Ph.D., California Institute of Regenerative Medicine (CIRM)

Table 13: ASO Nof1 Therapy Development: Partnering with nLorem
Sarah Glass Ph.D., nLorem Foundation

Table 14: Accelerating Nof1 Research and Therapy Development
Tim Yu, M.D., Ph.D., Boston Children's Hospital

Table 15: Translating Molecular and Genetic Understanding into Oligonucleotide-Based Medicines (OBMs)
Christopher Hart, Ph.D., Creyon Bio

Research & Drug Repurposing

Table 16: AI Driving Early Discovery - Glenn Morrison, Recursion

Table 17: Drug Repurposing and Partnering with Industry
Bruce Bloom, Healx

Table 18: Re-imagining & Scaling Drug Development for Rare Disease
Onno Faber, Rarebase

Table 19: AI for Drug Discovery: Accelerating Knowledge in Rare Disease
Lurong Pan, Ph.D., Alnnocence

Table 20: Engineered Cell Libraries for Fast Target Discovery
Robert Deans, Ph.D., Synthego

Table 21: Onboarding for Effective Drug Discovery: Getting to Proof of Concept Faster for Rare Disease Therapeutics
Casey McPherson, To Cure A Rose

Collaborations

Table 22: Opportunities for Community Data Analysis
Maria Picone, TREND

Table 23: Effective EHR Collection to Advance Natural History Data
Nasha Fitter, Ciitizen/Invitae

Table 24: Neurodevelopmental Data Collaborations, What Can We Learn
Jennifer Tjernagel, Simon's Searchlight

Table 25: New Funding Models for Rare Disease Research
Alok Tayi, Ph.D., and Bobby Bouthillette, Vibebio

12:45

It's a Wrap + Boxed Lunch & Networking