### Preliminary Agenda

#### Day 1  
**Wed 14 Sept 2022**  
**4:30 pm - 6:00 pm**

<table>
<thead>
<tr>
<th>Time</th>
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<tr>
<td>4:30 pm</td>
<td>Welcome</td>
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| 4:45 pm| Opening Keynote ‘When Rare Disease Becomes a BIG Data Opportunity’  
  Moderator: Morrie Ruffin, AAIH  
  Panelists:  
  - Ryan Colburn - Pompe Patient and Innovator  
  - Stacie Calad-Thompson - BioSymetrics  
  - Robert Deans - Synthego  
  - Kazi Mustaquisan - Roche |
| 5:30 pm| Can Big Data Supercharge Research for 10K Rare Diseases?  
  - Kirk Lamoreaux, Cogenticity and Report Author  
  - Wendy Erler, Global Advocacy, Alexion Astra Zeneca Rare Disease, 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**

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<th>Time</th>
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<tr>
<td>7:00 am</td>
<td>Breakfast</td>
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<td>7:30 am</td>
<td>Welcome</td>
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| 7:35 - 8:00 am     | **RARE-X - Leveraging What’s ahead**  
  - Charlene Son Rigby, RARE-X CEO |
| 8:00 - 8:45 am     | **The Future is Now: Realizing the Possibilities of Patient Driven Research**  
  - Wendy Erler, Global Advocacy, Alexion, RARE-X Board Member  
  - Betsy Bogard, FOP Advocate, Ensoma, RARE-X Board Member  
  - Julia Vitarelo, Batten’s Advocate |
| 8:45 - 9:30 am     | **N-of-1 Therapies - The Promise and Opportunity**  
  Moderator: Walt Kowtoniuk, RARE-X Board Chair, Third Rock Ventures  
  Panelists:  
  - Luke Rosen - Founder KIF1a.org  
  - Tim Yu - Boston Children’s, N=1  
  - Sarah Glass - nLorem  
  - Chris Hart - 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, Ring14 USA
- Bina Shah, Project 8p
- Scott Demarest, Colorado Children’s

10:45 - 11:30 am  **Built to Accelerate - disease characterization, progression models and research roadmapping**
- Karmen Trzupek, RARE-X
- Rachel Groth, BridgeBio
- Wendy Chung, Columbia & Simons Searchlight

11:30 - 12:45 pm  **Innovation Roundtables [Two - 30 min sessions ]**

**Practical Insights**
Table: Community Engagement/Enrollment Strategies, Tom Hume, RARE-X
Table: Effective Researcher Collaborations - Scott Demarest, Colorado Children’s
Table: Pre-Competitive Consortia Models to accelerate research and Discovery, Karmen Trzupek, RARE-X
Table: Disease Concept Model: STXBP1 Case Study - Charlene Son Rigby, RARE-X
Table: The Criticality of Developing a Data Governance Strategy for the Long Game - Vanessa Vogel Farley, RARE-X

**Mentoring**
Table: Research Roadmapping: Rachel Groth, BridgeBio
Table: Attracting Researchers - what you should be prepared with: Julia Vitarello, Stop Batten
Table: The Importance of NH Data to support Cell & Gene Therapy - Wendy Chung, Columbia
Table: How to partner with industry in your NH Registry - Betsy Bogard
Table: Entrepreneur Bootcamp - Getting Involved: Yael Weis, Mahzi
**Cell & Gene Therapy**
Table: Partnering With Columbus Childrens, Gene Therapies
Laura Hameed, Executive Director
Table: Research Opportunities, California Institute of Regenerative Medicine, TBD
Table: ASO Nof1 Therapy Development - Partnering with nLorem, Sarah Glass Ph.D., Chief Operating Officer
Table: Accelerating Nof1 Research and Therapy Development - Tim Yu, Boston Children's
Table: Translating Molecular and Genetic Understanding into Oligonucleotide-Based Medicines (OBMs) - Chris Hart, Creyon Bio

**Research & Drug Repurposing**
Table: AI Driving Early Discovery - Glenn Morrison, Recursion
Table: Drug Repurposing and Partnering with Industry - Bruce Bloom, Heax
Table: Re-imagining and Scaling Drug Development for rare disease - Onno Faber, Rarebase
Table: AI for Drug Discovery - Accelerating Knowledge in Rare Disease - Lurong Pan, AlInnocence
Table: Engineered Cell Libraries for Fast Target Discovery - Robert Deans, Synthego

**Collaborations**
Table: Opportunities for Community Data Analysis - Maria Picone, TREND
Table: Effective EHR Collection to Advance Natural History Data, Nasha Fitter, Ciitizen/Invitae
Table: Neurodevelopmental Data Collaborations, What can we Learn - Jennifer Tjernagel, Simon’s Searchlight
Table: New Funding Models for Rare Disease Research - Alok Tayi, Vibebio

12:45 It’s a Wrap + Boxed Lunch & Networking