



## Exchange Innovator Roundtables Agenda & Topic Descriptions

### Practical Insights

#### **Table 1: Community Engagement and Enrollment Strategies**

*Tom Hume, Marketing & Communications Lead, RARE-X*

So you generated excitement, launched your data collection program months ago, and now your enrollment has stalled. What can you do? Join this roundtable for a lively discussion of promotional best practices and re-engage tips to jump-start your community enrollment again.

#### **Table 2: The Challenges and Alternate Models to Engage with Researchers**

*Scott Demarest, MD, Ph.D., Colorado Children's*

With the traditional model of engaging researchers becoming more outdated, hear about the realities and challenges of working with Academic Researchers, how to best approach new models, and if and when they would be appropriate.

#### **Table 3: Pre-Competitive Consortia Models to Accelerate Research and Discovery**

*Karmen Trzupsek, Sr. Director Scientific Programs, RARE-X*

RARE-X is actively working with multiple rare disease groups to develop collaborative consortia, where advocacy groups representing different but symptomatically related rare disorders come together to develop shared strategies for research and investment.

#### **Table 4: Disease Concept Model: STXBP1 Case Study**

*Charlene Son Rigby, CEO, RARE-X*

Disease concept models document the symptoms and impacts important to a specific patient population, and provide a summary of the lived experience of individuals and their families. These models are developed via concept elicitation interviews with caregivers and clinical experts. With the growing importance of patient input into therapy development, disease concept models can be used to guide selection of clinical endpoints and clinical outcome assessments, as well as in discussions with multiple stakeholders including biopharma and regulatory agencies. The STXBP1 Foundation funded development of the first disease concept model for STXBP1 disorder through a novel approach: working with an academic center and genetic counseling program, and



## **Exchange Innovator Roundtables Agenda & Topic Descriptions**

applying AI methods to analyze interview content. The study identified STXBP1 symptoms of concern that had previously not been documented in the literature including gastrointestinal and respiratory issues. During this Innovator Roundtable, we will review this case study and discuss the value of disease concept models in enabling successful therapy development.

### **Table 5: The Criticality of Developing a Data Governance Strategy for the Long Game**

***Vanessa Vogel-Farley, Research & Data Governance Lead, RARE-X***

Keeping data secure, private, accurate, available and usable requires a robust Data Governance plan. Careful development, implementation and maintenance of a Data Governance Strategy enables data utility for its original intended purpose as well as yet to be identified utilizations in a manner that aligns with participant consent and choice. This roundtable will include an overview of the components of an effective Governance program including, the. Informed Consent process and documentation, how data governance and technology need to work together, data sharing, as well as international considerations.

### **Table 6: Citizen Science - Who, What, When, Why and How**

***Luke Rosen, [KIF1a.org](http://KIF1a.org)***

Getting Rid of the Box: unconventional, collaborative approaches that build and galvanize nano rare communities. This discussion might focus on how to work with the scientific, medical, biotech, and patient communities to accelerate discovery and development of treatments. But it might focus on any shot on goal to help families living with rare and often degenerative conditions.



## **Exchange Innovator Roundtables Agenda & Topic Descriptions**

### **Mentoring**

#### **Table 7: Research Roadmapping**

***Rachel Groth, Ph.D., VP, Head of Neuroscience Research, BridgeBio***

The decision by a pharma company to launch a drug discovery effort for a particular rare disease is informed by a multitude of factors, including an understanding of the underlying disease biology, the clinical manifestations that impact quality of life for patients and caregivers, and the feasibility of conducting a clinical trial. Discussions in this session will be focused on key areas that patient organizations can contribute to become “research ready” to accelerate drug development.

#### **Table 8: How Do We Get from Mila to Millions?**

***Julia Vitarello, Founder & CEO, Mila’s Miracle Foundation***

What does the future look like where individualized medicines are an integral part of how we treat disease, and what are the obstacles to getting there? This brings up a number of topics including data - ie. the incredible importance of families standing up to academics and industry to ensure their childrens’ data is transparently shared, ie. N1C/RareX database. I could also touch on other topics like the new risk-benefit analysis for 1/very few, and the controversial but hot topic for families of when “N-of-1” makes sense (ie many more families pushing for entering with one child/one good case right in front of us with a drug tested on that patient’s cells knowing that there are more who could eventually benefit.)

#### **Table 9: The Importance of NH Data to support Cell & Gene Therapy**

***Wendy Chung, M.D., Ph.D., Columbia & Simon’s Searchlight***

Natural history data for rare disease are critical to understand the disease manifestations, how they change over the life course, and what aspects are most important to patients and families. For some rare diseases there is significant variation across individuals, in part related to access to care and genetic variation. Natural history data are critical to plan clinical trials, assess feasibility and utility of outcome measures, and provide run-in data for participants to decrease the reliance on placebo control trial design.



## **Exchange Innovator Roundtables Agenda & Topic Descriptions**

### **Table 10: How to Partner with Industry to Build Real-World Datasets**

***Betsy Bogard, FOP Advocate, Ensoma, RARE-X Board Member***

Building collaborations with industry can be mutually beneficial for both the patient advocacy community and biopharma. Real-world data present a particularly rich opportunity for collaboration. Designing and generating natural history and other real-world datasets benefits from the skills and resources of both parties. Attendees participating in this session will hear from an expert who has worn both hats. You will have the opportunity to better understand industry's needs and uses for real-world evidence and the critical role a patient community can play in building it.

### **Cell & Gene Therapy:**

#### **Table 11: Partnering With Columbus Childrens, Gene Therapies**

***Laura Hameed, Executive Director, Columbus Children's Foundation***

Entrepreneur or Philanthropy: Creative models to break down barriers to gene therapy advancement for ultra-rare diseases.

#### **Table 12: Research & Development Opportunities and How to Get Funding**

***Lila Collins Ph.D. & Uta Grieshammer Ph.D., California Institute of Regenerative Medicine (CIRM)***

CIRM will provide an overview of our revamped funding programs under our 2022-2027 Strategic Goals – Consisting in: Discovery, Translational, Clinical, Education and Infrastructure Research Programs. CIRM presenters will engage into answering any questions from attendees.

#### **Table 13: ASO Nof1 Therapy Development - Partnering with nLorem**

***Sarah Glass Ph.D., Chief Operating Officer, nLorem Foundation***

The n-Lorem Foundation is dedicated to the principle that the individual is the indivisible unit of human value. To be optimally fair to the many, we must be fair to the individual.



## **Exchange Innovator Roundtables Agenda & Topic Descriptions**

To be generous to the many, we must be generous to the individual. For the human collective to achieve its maximum potential, the individual must have the opportunity to create maximum value. n-Lorem is the only non-profit using a mutation-driven approach to provide personalized experimental ASO medicines for nano-rare patients, for free, for life.

### **Table 14: Translating Molecular and Genetic Understanding into Oligonucleotide-Based Medicines (OBMs)**

*Christopher Hart Ph.D., CEO, Creyon Bio*

Creyon Bio is creating unprecedented efficiency, accelerating timelines, and ultimately lowering costs for drug development and changing how precision medicines are created for patients whether treating disease impacting one patient or millions. Traditional trial-and-error approaches to screening gene-based medicines cannot scale up to meet the increasingly rapid pace of genomic discoveries. Creyon has built a process to create the right dataset and to leverage best-in-class AI and machine learning that together extract out the design rules that then allow for engineering knowably safe and effective Oligonucleotide-Based Medicines (OBMs). Creyon's vision is to create a platform that can quickly advance novel OBMs into the clinic, ultimately avoiding months or years of lab studies that precede most clinical trials, making optimal OBMs rapidly available for patients who need them.

## **Research & Drug Repurposing**

### **Table 15: AI Driving Early Discovery**

*Glenn Morrison, Recursion*

Recursion is the clinical-stage biotechnology company industrializing drug discovery by decoding biology. Enabling its mission is the Recursion OS, a platform built across diverse technologies that continuously expands one of the world's largest proprietary biological and chemical datasets. Recursion leverages sophisticated machine-learning algorithms to distill from its dataset a collection of trillions of searchable relationships across biology and chemistry unconstrained by human bias. By commanding massive experimental scale – up to millions of wet lab experiments weekly – and massive



## **Exchange Innovator Roundtables Agenda & Topic Descriptions**

computational scale – owning and operating one of the most powerful supercomputers in the world, Recursion is uniting technology, biology and chemistry to advance the future of medicine.

### **Table 16: Drug Repurposing and Partnering with Industry**

***Bruce Bloom, Healx***

Healx is a mission-driven technology company, pioneering the next wave of drug discovery by re-engineering the entire process from the ground up. At the core of our approach is Healnet, the next generation AI platform we built to attack the drug discovery problem in a massively parallel, hypothesis-free way. We deploy Healnet across three core areas: drug redevelopment, combination therapy development, and molecular enhancement. In each of these areas, Healnet uses cutting-edge AI to rapidly discover novel disease biology and modes of action - without being limited to a single target - and identify non-obvious connections between that biology and existing compounds. Our team then applies their deep drug discovery and development expertise to accelerate these novel treatment opportunities towards the clinic. Furthermore, our portfolio management and modeling tool guides our disease selection and drug matching workflows to ensure we continually optimize the commercial and societal return of our programmes.

### **Table 17: Re-imagining and Scaling Drug Development for Rare Disease**

***Onno Faber, CEO, Rarebase***

Rarebase is a public benefit precision medicine company that leverages cutting-edge technology and biology to discover and develop treatments for the millions of people worldwide living with a rare disease.

### **Table 18: AI for Drug Discovery - Accelerating Knowledge in Rare Disease**

***Lurong Pan, Ph.D., Alnnocence***

Rare disease patients face insurmountable challenges, including misdiagnosis and a lack of disease-modifying therapies. The Rare AI Institute's precision medicine platform leverages multi-omics, structure biology and clinical data to better characterize the patient's disease and identify existing drugs which could be repurposed to treat them.



## **Exchange Innovator Roundtables Agenda & Topic Descriptions**

The institute is building the infrastructure needed to make the platform available to patients and their physicians worldwide.

### **Table 19: Engineered Cell Libraries for Fast Target Discovery**

***Robert Deans, Ph.D., Chief Scientific Officer Synthego***

Synthego is a dynamic next gen organization that is accelerating discovery by; Rapidly aggregating genetic disease variants into a common test bed (iPSC) for associated function, using in silico structure predictions to model disease, incorporating disease relevant diversity for drug screening, incorporating bioinformatics to identify disease modifying genes secondary to the driver allele. Learn what this means for your disease and your research driving efforts.

### **Table 20: Onboarding for Effective Drug Discovery**

***Casey McPherson, Everlum Bio, To Cure A Rose***

Getting to proof of concept in rare disease therapeutic development is critical, and there are opportunities for patient advocacy entrepreneurs to drive this. The goal is to do so faster and cheaper. What does this approach look like, how is Everlum building a process to support this accelerated work, and where does new science like the development of cell lines to aso's play a roll in the timeline and cost.

## **Collaborations**

### **Table 21: Opportunities for Community Data Analysis**

***Maria Picone, Co-Founder & CEO, TREND***

TREND Community's mission is to improve the quality of life for all of us living with rare and chronic disease. Our technology leverages social media to better understand disease burden. Our community-driven approach is accelerating the discovery and delivery of new treatments.



## **Exchange Innovator Roundtables Agenda & Topic Descriptions**

### **Table 22: Effective EHR Collection to Advance Natural History Data, *Nasha Fitter, Ciitizen/Invitae***

The most important thing a rare disease community can do is create a natural history study, documenting the symptoms patients face. There are many ways to do this wrong. Come learn from rare mom Nasha how the Ciitizen platform offers a digital platform for rare groups to build these studies, and how companies have used the data successfully in IND filings.

### **Table 23: Neurodevelopmental Data Collaborations, What Can We Learn *Jennifer Tjernagel, Senior Project Manager, Simon's Foundation***

Simon's Searchlight is an international research program that is creating an ever-growing database and resource network. We currently study over 150 genes that cause rare neurodevelopmental disorders, and our list is always expanding. You or your family member must have a genetic diagnosis of one of these conditions in order to join. The study is international, and families can participate in several languages. You and families like yours share valuable information and experiences that leading geneticists and scientists around the world can use to improve the lives of people living with rare genetic neurodevelopmental disorders. These researchers can also invite families to participate in their research studies in the future. People with genetic diagnoses, their families, and scientists play equal parts in this journey. Your unique experience could hold the clues that scientists need to find answers for you and others with rare genetic disorders. Learn more about Simon's Searchlight and how to register by visiting our website: [SimonsSearchlight.org](http://SimonsSearchlight.org)

### **Table 24: New Funding Models for Rare Disease Research**

*Alok Tayi, Ph.D. & Bobby Bouthillette, Vibebio*

The biggest obstacle to treating patients with overlooked diseases isn't finding potential treatments – it's funding them. Vibe Bio is creating a different, mission-driven approach to developing medicines, one that taps new sources of capital and empowers and unites the patients who would benefit from new therapies.