Data DIY
GlobalGenes.org
Tuesday, February 4, 2020

#DataDIY | GlobalGenes.org
Welcome and Introduction
Workshop 4: Becoming a Data-Centric Community
8:45–9:00

Christian Rubio
VP, Strategic Advancement
Global Genes

#DataDIY | GlobalGenes.org
Data DIY Workshop #4

Becoming a Data-Centric Community

• The Power and Potential of Data-Driven Organizations
• Managing Collaborations
• What More Can You Do With Data?

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WORKBOOK #1: The Whys and Hows of Patient Data Collection

Download your copy at: globalgenes.org/data-diy/

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WORKBOOK #2:
Data Trusts, Governance
and Collection Platforms

GOOD GOVERNANCE

OBJECTIVES
STAKEHOLDERS
TEAM
ELSI
SUSTAINABILITY

PLANNING

Download your copy at: globalgenes.org/data-diy/

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WORKBOOK #3: Collaborative Research Networks

Download your copy at: globalgenes.org/data-diy/

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**Patient-led Activities**
- Establish and fill registries
- Fund longitudinal natural history studies to find:
  - Full disease presentation
  - Biomarkers
  - Clinically meaningful endpoints
- Create tissue banks
- Fund genetic diagnosis and disease target research
- Assure intellectual property control and open access of tissue & models to researchers
- Promote disease awareness and genetic testing
- Investigate drug repurposing

**Knowledge Integration**

**Sponsor-led Activities**
- Define disease target for discovery effort
- Define therapeutic rationale (target interaction and drug / protein / gene technology) for discovery effort
- If drug, determine chemical library for screening
- Define and establish screening cascade
- Define cell & animal model outcomes for hit validation
- Propose potentially treatment-sensitive biomarkers and clinical endpoints based on therapeutic rationale

**Target Product Profile**
- Therapeutic technology
- Efficacy assessment targets
  - Biomarkers
  - Patient-relevant, validatable clinical endpoints
- Tolerability objective
  - Patient-acceptable levels of expected side-effects from this therapeutic approach
- Convenience objective
  - Minimum acceptable drug route and frequency
## AGENDA

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<td>Introduction to Workshop 4: Becoming a Data-Centric Community</td>
<td>Christian Rubio, VP, Strategic Advancement, Global Genes</td>
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<td>9:00–10:00</td>
<td>Refining Your Strategy: Building a Bridge to Treatment Options</td>
<td>David Spencer, PhD, Vice-Chairman of the Board, BioPontis Alliance for Rare Diseases</td>
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| 10:00–11:00 | Funding DIY: EB Research Partnership’s Multi-Stakeholder Platform Powered by Venture Philanthropy | Michael Hund, MBA, Chief Executive Officer, EB Research Partnership  
                      Ryan Jancaitis, Global Head, Product Management, Amazon Web Services |
| 11:00     | BREAK                                                                 |                                                                           |
| 11:15–12:15 | Leveraging the Power of Partnership: Managing Multi-Stakeholder Collaborations | Kristin Schneeman, Director, Programs at FasterCures                       |
| 12:15–1:15 | LUNCH                                                                 |                                                                           |
| 1:15–1:30 | Welcome Back                                                            | Christian Rubio, VP, Strategic Advancement, Global Genes                  |
| 1:30–2:30 | Creating a Global Network                                               | Lara Pullen, PhD, Co-founder, Chion Foundation  
                                      Maria Picone, Co-Founder/CEO at TREND Community  |
                                      Bernhard Suter, M.D., Asst. Professor, PEDs & Neurology, Baylor College of Medicine &  
                                      Texas Children's Hospital  
                                      Daniel G. Glaze, M.D., FAASM, Professor, PEDs & Neurology, Baylor College of Medicine |
| 3:30     | BREAK                                                                 |                                                                           |
| 3:45–4:45 | Expanding Your Engagement to Underserved Populations                   | Liz Horn, LHC Biosolutions  
                                      Linda Wade, President & CEO, Sickle Cell Assoc, of Texas, Marc Thomas Foundation  
                                      Jordan Shumway, Social Worker, Sickle Cell Assoc. of Texas, Marc Thomas Foundation  
                                      Khrystal K. Davis, JD, Founder, Texas Rare Alliance  
                                      Robyn Correll Carlyle, MPH, CHWI, Dia de la Mujer Latina, Inc. (DML) |
| 4:45–5:00 | Wrap Up, Final Questions, Next Steps, and Round Table                 | Christian Rubio, VP, Strategic Advancement, Global Genes                  |
Global Genes Question Wall

To participate, please visit https://datadiy4.cnf.io/ with your browser

Note: Responses and submissions are anonymous
Polling Question
Refining Your Strategy: Building a Bridge to Treatment Options
9:00–10:00

David Spencer, PhD
Vice-Chairman of the Board,
BioPontis Alliance for Rare Diseases
The drug will 'fix' the issue by inhibiting a protein's activity (e.g., small molecule), replacing the missing protein (e.g., enzyme replacement therapy for enzyme deficiency disorders), etc.

PK/PD - Pharmacokinetics and pharmacodynamics properties include potency, toxicity, etc.

Questions: datadiy4.cnf.io/
Disease Therapy Development Is Making The Leap From Academia To Industry

Barriers of entry

Academic researchers finding disease mechanisms

Reality: “publish or perish”, lifeblood = grant funds

Pharmaceutical industry developing & manufacturing drugs

Reality: “return on investment”, lifeblood = financial sustainability

Basic research -> Discovery/Preclinical -> Clinic to Market
Few Grants Exist For Drug Discovery And It Is Hard To Publish On Basics Of Drug Development

Academic researchers finding disease mechanisms

**Reality:** “publish or perish”, *lifeblood = grant funds*

Pharmaceutical industry developing & manufacturing drugs

**Reality:** “return on investment”, *lifeblood = financial sustainability*

Barriers of entry

- Lead discovery
- Preclinical development

Grants are far more available for advancements in basic research than the “applied research” to find and optimize potential cures

Basic research → Discovery/Preclinical → Clinic to Market
Especially in Rare Disease, Industry Needs Projects That Are More Advanced Than Academia Can Produce

Academic researchers finding disease mechanisms

**Reality:** “publish or perish”, lifeblood = grant funds

**Pharmaceutical industry** developing & manufacturing drugs

**Reality:** “return on investment”, lifeblood = financial sustainability

**Barriers of entry**

- Lead discovery
- Preclinical development

Pharmaceutical companies need a reasonable shot at return on investment, so rare disease projects, targeting smaller patient groups, need to be sufficiently de-risked

- Strong proof of preclinical efficacy and safety is needed

Basic research ➔ Discovery/Preclinical ➔ Clinic to Market
Other Gaps Exacerbate The Problem

• Drug development know-how
  – Patient organizations, even if funding basic research hoping to find a therapy, most often have to **invent their own pathway** and bridge over the valley of death. Most lack expertise and resources to do so

• Integration of the patient perspective
  – In rare disease, patients and caregivers can know more **about their disease than their treaters**, yet industry tends to involve patient organizations only when they near clinical testing

Questions: datadiy4 cnf io/
Other Gaps Exacerbate The Problem

• Financing
  – Only **charitable giving** can feed the earliest stages of drug discovery; venture capital or pharmaceutical industry funding are only obtainable in later phases
    • “Venture philanthropy” may start to bridge that gap, but too little is available

• Intellectual property (IP) management
  – Key information/resources (e.g., natural history studies, tissue banks) must be available for all researchers but the crucial IP relating to potential therapies must be kept bundled in one place, easing out-license to a drug development partner
  – The pressure on academics to publish may lead to inability to patent key findings, lessening industry interest
Paths To New Therapies

Drug repurposing

- **Pro:** if an already approved drug can ameliorate the disease, that is the quickest path
- **Con:** frequently not possible and any successful drugs may provide partial symptomatic relief instead of “disease modifying” effects
- **Con:** sometimes not possible to protect IP, preventing investment
- Some companies exist that use advanced methodologies to help find potential repurposed drugs
  - **BioVista:** Vizit-Medical software allows rapid exploration of connections between genes, proteins, diseases, drugs based on PubMed
Paths To New Therapies

“De novo” therapy discovery

- Small molecule drugs
  - **Pro:** high-throughput screening, precise control of structure and purity, tunable structures, less expensive manufacturing
  - **Con:** toxicity can be hard to predict and it is not always possible to address a rare disease pathway with a small molecule

- Biologics (large proteins made in cell culture)
  - **Pro:** can consist of or be modelled on normal proteins, more predictable toxicities, can interact with nearly all biological targets
  - **Con:** expensive to make, assuring purity can be difficult, usually i.v., can cause antigenicity

- Antisense oligonucleotides (AONs – small bits of synthetic RNA/DNA)
  - **Pro:** cheaper to manufacture, allow silencing of mutant protein production or “skipping” of pathogenic protein portions
  - **Con:** less useful for widely distributed tissues and for diseases in which the needed protein is deleted

- Gene therapy: may provide long-term cure but problems can include antigenicity of delivery virus, cancer risk, transfection of enough cells to restore function and high expense
Bridging The Gap

Patient organizations can provide an advanced foundation for drug discovery and development by partnering with academic scientists and clinicians

**Awareness**
- Use scientific literature, social media and TV to spread awareness and support fundraising efforts

**Diagnostics**
- Encourage development of specific diagnosis and “when to test” guidelines

**Registries**
- Find as many patients as possible

**Natural History Studies**
- Start natural history studies to describe disease variation/progress and potential clinical endpoints

**Tissue Banks**
- Start tissue banks open to all investigators to foster patient-relevant research

**Biomarkers**
- Develop biomarkers that can be used in clinical trials to correlate with disease status (to indicate progression or improvement)
Steps That Patient Organizations Can Take

• Attract people* with graduate training in the life sciences to work with you
• Work with them to find published experts in basic and clinical research on your disease area
• Find champions among the research/clinical scientists that will work closely with you and actively collaborate with other experts
• Use raised funds to support research that can lead to therapeutics, keeping control of the results
  – One starting place is the BioPontis Translational Research Readiness Tool
• Find scientists* with pharmaceutical drug development experience to help formulate therapeutic discovery strategies
• Make sure to use legal agreements that:
  – Reward all players in case of success
  – Grant access to crucial materials that support research (e.g., patient tissues, data on potential biomarkers)
  – Protect critical IP (therapeutic mechanisms, hits, potential lead compounds) so that successful discovery programs can be licensed in one bundle to potential sponsors

* Can be volunteers working under CDA
BioPontis Alliance For Rare Diseases: Mission And Goal

We are a nonprofit Foundation started by pharmaceutical industry professionals, academics, patient advocates and lawyers in order to identify promising science and work to transform it into potential therapies for rare neurological diseases

• Our goal is to validate promising research and complete preclinical development, applying philanthropic funds, to initiate the pathway to a final product that can be marketed by a development partner.

• Our method is to work collaboratively with key scientists, contract research organizations (CROs) and patient organizations to integrate the patient viewpoint as early as possible and leverage best practices in the pharmaceutical industry for drug discovery and preclinical development

Questions: datadiy4.cnf.io/
BioPontis History And Accomplishments

• Founded in 2015, we initiated operations to find promising basic research projects in neurological rare disease

• In 2016:
  – Charitable start-up funds were raised from the Baillet-Latour fund, pharmaceutical companies (Shire, GSK) and private individuals
  – A set of criteria for candidate projects was developed
  – A first project was initiated with Dr. Albena Jordanova at University of Antwerp and VIB: HINT1 activators for Charcot-Marie-Tooth (CMT) disease, a form of inherited peripheral neuropathy
  – Meetings were held with 17 neurological disease patient organization representatives to determine how best to integrate the patient perspective into the earliest stages of rare disease drug discovery and preclinical development ¹

• In 2017, on the basis of their request, BioPontis developed the Translational Research Readiness Tool (TRRT) and piloted the use with two patient organizations and their scientific advisors

¹ Summarized in white paper "Integrating Rare Disease Patients into Pre-Clinical Therapy Development; Finding our Way with Patient Input"
Integrating Rare Disease Patients into Pre-Clinical Therapy Development

Finding our Way with Patient Input

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The TRRT Probes Readiness In Several Areas

• Disease target, basic science of relation of target to disease, animal models, assays
• Therapeutic strategy – how to correct the defect
• Clinical knowledge base, including diagnosis, existence of registries, tissue banks, natural history studies, clinical endpoints and biomarkers (biochemical signs that may correlate with disease progression, symptoms or clinical interventions)
• Regulatory interactions
• Preparation and execution of clinical trials

“The exercise gave the FSH Society an opportunity to review the critical benchmarks that the field needs to meet in order to proceed with drug discovery and development, and to have several scientific experts weigh in on the current status of the field. Feedback from researchers indicated that the field would benefit from obtaining cells from a more genetically diverse population of patients, investing in research on differentiating iPSCs into cells with skeletal muscle morphology, and establishing sources that are more easily available to researchers. We’ll be able to continually update the RRT to measure our progress and efficacy of our funding.” – June Kinoshita, FSHD Society Executive Director
HINT1 is one of over 70 sub-types of CMT.

CMT is:

A disease of the peripheral nerves that control muscles (unlike the muscular dystrophies, which affect the muscles themselves).

Found in both genders and in all races and ethnic groups, it is the most commonly inherited peripheral neuropathy, affecting 2.6 million people worldwide.

CMT is slowly progressive, causing loss of normal function and/or sensation in the lower legs/feet and hands/arms. CMT is currently incurable, but not usually fatal, though it is severely disabling in a small proportion of cases.

https://www.cmtausa.org/understanding-cmt/what-is-cmt/
CMT Presentation And Origins

• Target organs
  – CMT can cause demyelination (CMT type 1), axonal death (CMT type 2) or both of peripheral motor and sensory nerves

• Symptoms
  – Progressive weakness and wasting of distal muscles, motor impairment, sensory loss and skeletal deformities

• Genetically heterogeneous
  – Mutations in more than 80 genes identified
  – Most are autosomal dominant but many are autosomal recessive
  – Affected systems include mRNA processing, mitochondrial maintenance, cytoskeleton organization, endosomal sorting, etc.

• Progressive, severely disabling, currently incurable
HINT1 Axonal Neuropathy

- HINT1 mutation is one of the top three most frequent genetic causes of autosomal recessive CMT and 80% of those showing the clinical hallmark of neuromyotonia – 79 cases identified to date

- A single point mutation, R37P, accounts for over 70% of HINT1-related axonal neuropathy in over 60 identified families

- The highest carrier prevalence ranges from western Europe through central Europe and the middle east, in some regions as high as 1 in 67-182 people

Questions: datadiy4.cnf.io/
Loss-of-function HINT1 Mutations

Loss of function due to:

- Instability of the protein (R37P, C84R)
- Loss of enzymatic activity (H112N)
- Nonsense-mediated decay
Summary: HINT1 Project Rationale

• HINT1 is histidine triad nucleotide binding protein 1, a highly conserved protein that hydrolyzes most compounds bound to AMP, binds histidine and regulates several transcriptional factors

• The R37P point mutation is heritable and is by far the most frequent cause of HINT1-related axonal neuropathy

• This form of axonal neuropathy is relatively severe and progressive, diagnosed in the first decade of life

• The goal of the drug discovery program is to find small molecules that can stabilize the protein (act as chaperones) to restore function and structure
  – Parents of HINT1 patients are unaffected, while they have only 50% HINT1 activity, indicating that even partial restoration of HINT1 activity might be sufficient to alleviate the disease
HINT1 Project Progress

• In 2016, when the project was started, there was good basic and clinical science demonstrating the tight connection between HINT1 mutation and the disease, but no real path to new therapeutics.

• Starting with the goal of finding protein stabilizers (primarily for R37P), the team, primarily consisting of investigators at the University of Antwerp and VIB, BioPontis personnel and support from AMRI, started development of assays to support characterization and high-throughput screening of compounds.

• Today, a number of screening hits have been found that may activate or stabilize HINT1.
  – Several confirmatory assays and cellular models have also been created to support evaluation of hits and lead compounds.

Questions: datadiy4.cnf.io/
Conclusions From The BioPontis Experience

• Getting drug development experts together with researchers and patient organizations in a non-profit backdrop can produce real results in starting the drug discovery process
  – One crucial element is establishing the right incentives to share and create IP and generate and atmosphere of mutual trust

• Non-profit fundraising to support a structured, repeatable process that can be used for many diseases is challenging
  – Emotional appeal that disease-specific organizations can create is hard to emulate

The gap can be crossed with the right tools, funding and people
Backups
Technology Partners: Incentivizing Technology Transfer With Academia

Shared economics and science

Economic & IP Sharing  Partnered Development

- Pro Rata share of all economic value of IP shared back to original IP source (university)
- Inventor is Development Team Participant
- Academic Partner/Inventor participates in new IP value created at BioPontis
- BioPontis' pro rata share of returns is re-invested into our mission

Questions: datadiy4.cnf.io/
BioPontis creates a multidisciplinary team for each Rare Disease Therapeutic Program. BioPontis Scientist leads each team and provides project management.
Alliances And Partnerships: We Start With Patients And Science

**Research Partners**
- Academia
- Patients’ Organizations
- Research Institutes

**Development to Proof-of-Concept**
- BioPontis licenses IP - manages IP and technical project development

- Early Discovery
- Lead Identification
- Lead Optimization
- Preclinical Development

**Patients and academia integrated into development team**

**Clinical Development and Commercialization**
- Biopharmaceutical company acquirer
- New Company - NEWCO
Board and Executive Management

**Scientists & Clinicians**

Chief Science Officer
**Gilbert Carnathan, PhD, RAC**
Drug discovery, regulatory and development
Prior: VP Cortech, Napro Biotherapeutics, Argos Therapeutics; President, Origin LLC

Board member
**Jean–Jacques Cassiman, MD**
Prof Em KU Leuven, human genetics
EPPOSI, ESHG (Sec Gen)
Academy Award Science Communication

Board member
**David Spencer, PhD**
Prior: CSO BioPontis Alliance, VP Bayer, COO/CSO Biolex, ProteoVec

Board member
**Warren Strittmatter, MD**
Prior: Chief Dept Neurology
Duke University

**Patients’ Organization**

Board member
**Susan Kahn**
Exec Director, Tay-Sachs and Allied Diseases Association

**International Law**

Board member
**Howard Liebman, Esq.**
Senior EU Partner, Jones Day,
President Amcham Belgium

**BioPharma Industry**

Board member
**Christian Policard, PhD**
Prior: SVP Sanofi France
EVP, Institute Pasteur

Executive Chairperson & co-founder
**Erik Tambuyzer, PhD**
Prior: Co-Founder Innogenetics,
SVP Genzyme, co-Founder EPPOSI,
former Chair EuropaBio

Questions: datadiy4.cnf.io/
Our Patient Integration Activities

You can download our research & papers:

• White paper: “Integrating Rare Disease Patients into Pre-Clinical Therapy Development”
  http://biopontisalliance.org/patient-input/

• Translational Research Readiness Tool: http://biopontisalliance.org/readiness-survey/
Polling Question
Questions?
Funding DIY: EB Research Partnership’s Multi-Stakeholder Platform Powered by Venture Philanthropy

10:00–11:00

Michael Hund, MBA,
Chief Executive Officer,
EB Research Partnership

Ryan Jancaitis
Global Head, Product Management,
Amazon Web Services
Funding DIY: EB Research Partnership’s Multi-Stakeholder Platform Powered by Venture Philanthropy

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Questions: datadiy4.cnf.io/
Rare Disease Innovation: Venture into Cures

Michael Hund, MBA
Chief Executive Officer
EB Research Partnership
@MichaelPHund
@EBresearch

www.ebresearch.org
OUR STORY

ONE MISSION: Accelerate treatments and cures for EB.
We have reached the threshold of a far more promising future for children born with EB. Leading researchers believe that both life-changing treatments and a cure are within reach. To realize our vision of a world without EB, we’re building a research model based on three core pillars.

**DATA PLATFORM**
The largest gathering of EB data imaginable, including clinical, genomic, and patient data underlying the disease.

**RESEARCH NETWORK**
An open and fluid collaboration consortium that brings together the often siloed academic, medical, and patient communities.

**IMPACT PORTFOLIO**
Direct investment in the most promising projects, including disease controlling, changing, and curing therapies.
IMPACT

$35MM
Raised since 2011

EB research projects funded

65

12x
The amount of clinical trials (2 - 24+)

4
Phase III clinical trials expected within a year

3
Companies formed

90%
Of revenue directed to research

www.ebresearch.org
2019 YEAR IN REVIEW

2019 YEAR IN REVIEW

$5.5 MILLION FOR RESEARCH FUNDED BY EBRP

21 PROJECTS APPROVED FOR ALL MAJOR EB SUBTYPES USING INNOVATIVE AND DIVERSE THERAPEUTIC TECHNIQUES

RESEARCH FUNDED IN 6 COUNTRIES

NEARLY 90% OF REVENUE DIRECTED TOWARDS RESEARCH

INDUCED PLURIPOTENT STEM (IPS) CELL TECHNOLOGY

PROTEIN EXPRESSION INDUCTION

BIOBANKING

GENE EDITING: CRISPR, BASE EDITING, VIRAL VECTORS

EXON SKIPPING

PREMATURE TERMINATION CODON (PTC) READTHROUGH

PROTEIN REPLACEMENT THERAPY

COMPUTATIONAL DRUG REPURPOSING

EBRP funds diverse and innovative research approaches to maximize the potential of discovering treatments and cures for every individual with EB, regardless of subtype.
COLLABORATION: iPS Cell Consortium

iPS-derived Skin Grafts for Epidermolysis Bullosa
iPS CELL CONSORTIUM

RESULTS:

A DECADE FASTER

IN THE CLINIC IN 5 YEARS INSTEAD OF 15
THE KEY: VENTURE PHILANTHROPY

Return on investment directed back into more research funding until a cure is found
EBRP’s Venture Philanthropy Definition:

In return for providing capital to de-risk early stage medical research, EBRP maintains the ability to share in the financial upside generated by research EBRP has funded.

• EBRP is agnostic regarding structure (equity, guaranteed ROI, warrants, etc.)

• EBRP seeks the ability to move IP forward with different party if commercial partner decides to no longer pursue

• EBRP is constantly weighing any conflicts of interest

• EBRP is as transparent to its community as possible
VENTURE PHILANTHROPY 101

1. Time is our greatest asset

2. The non-profit model is broken
   
   • Shift funding mindset from donation to investment
   • Non-profit is a tax status not a business model

3. No sector exists in which one group de-risks IP and passes its economics to the next party except non-profit
   
   • WHY?! Typical answer “just because”
   • Time to carve same financial pie differently than before

4. Venture philanthropy model benefits ALL parties involved because commercial partners use non-profit capital to de-risk opportunities
VENTURE PHILANTHROPY TOP 5

EBRP’s five absolute must-haves for venture philanthropy:

1. IP/Product that can be monetized & market opportunity for product (nice to have larger market possibility also)
2. Independent third-party validation of what you are funding
3. Non-profit commanding market presence
4. Investment/structuring expertise
5. Good legal advisors (pro bono even better)

Questions: https://datadiy4.cnf.io/
REASONS EB IS THE RIGHT RARE DISEASE FOR VENTURE PHILANTHROPY

1. Researchers have identified the genetic mutations

2. Methodologies on how to treat the condition have advanced to clinical trials

3. Easy to observe if a treatment is working

4. Protein deficiency at its core (long history of treating protein problems in medical research)

5. Large enough patient population for commercial solutions

6. Approaches to treating EB are likely applicable to >7000 other conditions and 350 million people

Questions: https://datadiy4.cnf.io/
VENTURE PHILANTHROPY LIFE CYCLE - LOOK FAMILIAR?

Questions: https://datadiy4.cnf.io/

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VENTURE PHILANTHROPY CASE STUDY 1

We fund a university to develop a treatment for severe EB that corrects gene mutations in skin cells under venture philanthropy agreement.

A public biotech company uses that intellectual property to make a treatment, we received stock in the company.

That stock grew to worth multiples beyond our original investment.

We sell our shares and direct that revenue right back into research projects.

3 YEARS

$500,000

6X Return on Investment

$3,000,000
VENTURE PHILANTHROPY CASE STUDY 2

EB Gene Editing & Larger Market Opportunity

- 10% of Americans = 31.9 Million People
- 7,000 Incurable Rare Diseases
- 5,400 Incurable Genetic Rare Diseases
  - EB = “Proof of Concept”
  - Gene Therapy

Works if We Control Intellectual Property
VENTURE PHILANTHROPY CASE STUDY 3

2018: EBRP awards $5 million to Netherlands-based public company ProQR for clinical development of QR-313.

Exon skipping: first-in-class RNA-based oligonucleotide designed to address the underlying cause of DEB due to mutations in exon 73 of the COL7A1 gene. Platform technology which can be used to address additional mutations.

2019: ProQR strategic spin out of their DEB activities into newly formed company, Wings Therapeutics, formed and financed by EBRP to take exon skipping forward. Continue to conduct clinical trials with QR-313 in exon 73 as well as progress other RNA molecules that are designed for other mutations that cause DEB.

www.ebresearch.org
VENTURE PHILANTHROPY KEY GOALS

1. Rapidly accelerate treatments and cures for EB
2. Create an innovative and sustainable funding model
3. Build a bridge between patients, research, and industry
4. Pioneer the model for all rare disease

Questions: https://datadiy4.cnf.io/
To maintain competitive advantage, digital businesses must innovate as rapidly as possible.
Creating a culture of Innovation

**Mechanisms:** Encoded behaviors that turn ideas into meaningful innovations – *Working Backwards methodology*, PR-FAQs, and detailed Narratives

**Generate Ideas:** We hire innovators, and we encourage new, disruptive ideas from across our organization

**Culture:** Hire builders & manage against our 14 Leadership Principles, Think Big & invest early – and accept failure as a critical part of the innovation process

**Architecture:** Empower innovators through self-service tools; utilize AWS cloud platform to **build quickly**, scale fast and minimize costs of failure

**Organization:** We **decentralize authority** to single-threaded owners leading small, empowered ‘two pizza’ teams that own what they create

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Where innovation begins

Start with the customer and work backwards
Applying Innovation at EB Research Partnership

Problem Statement
The current process to identify genomic data associated with a single or group of phenotypes, or vice versa, doesn’t exist as data is not regularly or easily shared across institutions.

Press Release
A user friendly means of sharing data and outcomes to increase the speed of access to genomics and phenotypic data to enable research breakthroughs in EB treatment and cures.

Targeted Personas
An Investigator has been given a project, searches for relevant data, and analyzes the result set. A Patient is an individual or relative of an individual that suffers from EB.

Success Criteria
A web portal that provides Investigators a means to quickly find and analyze relevant data, while providing self-reporting capabilities to patients and parents of patients to enable research.
EBRP DATA PLATFORM

- EB CCOD
- EHRs
- Bioinformatics

Natural History

Direct to Patient

- HIPAA, HITRUST, and/or GxP standards
- De-identified
- Security & Compliance
- Agile
- Scalable computation, storage, and analytics
- Simplified collaboration
- Ecosystem of partners and tools

EB Clinical Research Consortium

Industry
- Biotech/Pharma
- FDA
- Tech

Patients & Families

www.ebresearch.org
THANK YOU
Polling Question
Questions?
Break

Data DIY

We’ll be back @ 11:15am
Global Genes Question Wall

To participate, please visit https://datadiy4.cnf.io/ with your browser

Note: Responses and submissions are anonymous
Leveraging the Power of Partnership: Managing Multi-Stakeholder Collaborations
11:15–12:15

Kristin Schneeman
Director
FasterCures

Questions: datadiy4.cnf.io/
Collaborative Research and Development Is on the Rise
Framework Report

Landscape Analysis

Consortia-pedia Catalogue

COMMENTARY

COLLABORATIVE ENVIRONMENTS

Consortium Sandbox: Building and Sharing Resources

Mark D. Lim

Some common challenges of biomedical product translation—scientific, regulatory, adoption, and reimbursement—can best be addressed by the broad sharing of resources or tools. But, such aids remain underdeveloped because the undertaking requires expertise from multiple research sectors as well as validation across organizations. Biomedical resource development can benefit from directed consortia—a partnership framework that provides neutral and temporary collaborative environments for several, oftentimes competing, organizations and leverages the aggregated intellect and resources of stakehold- ers so as to create versatile solutions. By analyzing 359 biomedical research consortia, we tracked consortia growth around the world and gained insight into how this partnership model advances biomedical research. Our analysis suggest that research by consortia provides benefit to biomedical science, but the model needs further optimization before it can fully integrate into the biomedical research continuum.

Sci. Trans. Medicine, June 2014

Questions: datadiy4.cnf.io/

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Captured Key Features of Consortia

<table>
<thead>
<tr>
<th>Mission, Structure and Governance</th>
<th>Expectations</th>
<th>Decision-making authority</th>
<th>Culture of trust</th>
</tr>
</thead>
<tbody>
<tr>
<td>Financing</td>
<td>Memberships</td>
<td>In-kind contributions</td>
<td>Diversification</td>
</tr>
<tr>
<td>Human Capital</td>
<td>Leadership</td>
<td>Program management</td>
<td>Volunteer army</td>
</tr>
<tr>
<td>Intellectual Property</td>
<td>Defining “pre-competitive”</td>
<td>Master licensing agreement</td>
<td>Exclusivity</td>
</tr>
<tr>
<td>Data Sharing</td>
<td>Agreements</td>
<td>Standards</td>
<td>Public dissemination</td>
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<tr>
<td>Patient Participation</td>
<td>Patients as a stakeholder</td>
<td>Patient-led</td>
<td>Communication</td>
</tr>
<tr>
<td>Measuring Value and Impact</td>
<td>Impact &amp; mission statement</td>
<td>Assessment periods</td>
<td>Stakeholder directed</td>
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</tbody>
</table>

Questions: datadiy4.cnf.io/
Consortium Checklist

• Shared-scientific problem
• Concrete goal / broadly usable solution
• Who has the resources and expertise (don’t just go for those with $$)
• Individual champion who can rally the experts
• Formalized governance
• Project management - milestones/timeline, metrics
• Human capital
• Dissemination – Access, licensing, IP
What Do Consortia Produce?

- Tool Development: 144
- Biomarkers: 119
- Basic Research: 107
- Data Sharing: 81
- Product Development: 66

Questions: datadiy4.cnf.io/
Framework

Alignment of mission, expectations, and a clear understanding of the nature of the partnership are key to the vitality of any consortium. We present the following questions to better guide conversations and thinking around the structure of this unique model of partnership.

MISSION AND VISION
- Who are my partners? What incentives drive each of the organizations partaking in this consortium?
- Do we share an unmet need that can advance both a shared goal and our unique individual objectives?
- Can we coalesce around a shared vision for moving forward?
- What are the outputs and outcomes of this effort? Who are the beneficiaries? Is this consortium created to provide data, tools, and resources to benefit all partners and the broader research community?

TERMS OF ENGAGEMENT
- What assets and resources can each partner bring to the effort?
- What resources are needed to augment existing assets? How do you access those external resources?
- What policies and practices can each partner agree to, regarding:
  > Data-sharing
  > Intellectual property
  > Conflict of interest
  > Material-sharing
  > Confidentiality
  > Data Access

SUSTAINABILITY
- What accountability measures must be in place to track progress and impact?
  > Equitable and timely contributions of resources and effort from all participants
  > Scientific milestones on research projects
  > Strategic milestones on consortium progress toward mission
  > Other strategic measures and mission-driven considerations
  > Procedures to ensure return-on-investment to participants and sponsors
- How will metrics be used to provide real-time feedback, and how will these impact the trajectory of the consortium?
- Are there external factors that must be considered in the near- and long-term that could potentially shift the focus of the consortium or alter the nature of the partnership?
Patient Groups As Collaboration
Conveners

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Biomedical Research and Development

Through Multi-stakeholder Collaboration

When is collaboration the right solution?

How can foundations preserve their mission while accounting for their partners’ differing interests?

What are the main obstacles to creating collaboration?

What resources exist or are needed to address legal agreements, data sharing, and intellectual property?
Why Are Foundations Becoming Collaboration Conveners?

“Because we have no choice. None of us can do this alone.”

“Because we have the end in mind.”

Foundations can build a bridge between basic and applied science.

Foundations are driven by a sense of urgency to streamline processes, reduce redundancies, learn from failures, and enable communication.
Defining Collaboration

“If you’re doing it right, you’re not just a funder, you’re a partner.”

FasterCures defined consortia as initiatives characterized by:

- Integration of researchers in a non-competitive space
- Agreement on a mission that addresses a shared need
- A governance structure that values each stakeholder’s input
- An integrated research plan that utilizes each stakeholder’s resources/skills

Questions: datadiy4.cnf.io/
<table>
<thead>
<tr>
<th>FOUNDATION CONVENER</th>
<th>NAME OF COLLABORATION</th>
<th>PURPOSE</th>
<th>PARTNERS</th>
</tr>
</thead>
<tbody>
<tr>
<td>AMERICAN HEART ASSOCIATION (AHA)</td>
<td>AHA PRECISION MEDICINE PLATFORM</td>
<td>Allows researchers and clinicians to access and analyze vast and diverse data to facilitate collaboration and accelerate breakthroughs in prevention, treatment, and cures for heart disease and stroke.</td>
<td>Amazon Web Services, AstraZeneca, Cedars-Sinai Heart Institute, Dallas Heart Study, Duke Clinical Research Institute, Intermountain Medical Center Heart Institute, International Stroke Genetics Consortium, and Stanford Cardiovascular Institute</td>
</tr>
<tr>
<td>AMYLOIDOSIS FOUNDATION</td>
<td>AMYLOIDOSIS RESEARCH CONSORTIUM</td>
<td>Works to accelerate the development of advanced diagnostic tools and effective treatments for systemic amyloidosis.</td>
<td>25 academic research centers</td>
</tr>
<tr>
<td>CHILDREN’S TUMOR FOUNDATION</td>
<td>NF PRECLINICAL INITIATIVE</td>
<td>Works to accelerate proof of concept testing of potential effective repurposed drugs in neurofibromatosis-1 (NF)-relevant models, and to frontload the clinical pipeline with new drug candidates for NF1.</td>
<td>Four leading NF academic laboratories with plans in 2017 to expand to partners in the pharmaceutical industry</td>
</tr>
<tr>
<td>COPD (CHRONIC OBSTRUCTIVE PULMONARY DISEASE) FOUNDATION</td>
<td>COPD BIOMARKER QUALIFICATION CONSORTIUM</td>
<td>Pools existing data from clinical studies evaluating various biomarkers to provide sufficient information to qualify them so that the U.S. Food and Drug Administration (FDA) and the European Medicines Agency can use them to evaluate new treatments.</td>
<td>GlaxoSmithKline; Boehringer-Ingelheim; AstraZeneca; Pfizer; National Heart, Lung, and Blood Institute; and FDA</td>
</tr>
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<td>CURE DUCHENNE</td>
<td>COLLABORATIVE TRAJECTORY ANALYSIS PROJECT</td>
<td>Works to unleash the power of collaborative data science on clinical trial design, potentially helping the entire community to bring effective new therapies to patients more quickly.</td>
<td>Pfizer, BieMarin, Shire, Sarepta, PTC Therapeutics, Solid Biosciences, Catabasis Pharmaceuticals, Bristol-Myers Squibb, and Parent Project Muscular Dystrophy</td>
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<td>FOUNDATION FOR THE NIH</td>
<td>BIOMARKERS CONSORTIUM</td>
<td>Identifies, develops, and qualifies biomarkers to advance specific applications for diagnosing disease, predicting therapeutic response, and improving clinical practice using new and existing technologies.</td>
<td>National Institutes of Health (NIH), FDA, Centers for Medicare and Medicaid Services, more than 20 biopharma companies, and five nonprofits</td>
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<td>JDRF</td>
<td>ENCAPSULATION CONSORTIUM</td>
<td>Develops a product that will hide implanted beta cells from the immune system and allow people with type 1 diabetes to live life as if they don't have the disease.</td>
<td>More than 25 research institutions</td>
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<td>ONE MIND</td>
<td>TRACK-TBI</td>
<td>Coordinates a national collaboration among Level I Trauma Centers that will enroll 3,000 patients in the largest longitudinal study of TBI (traumatic brain injury) ever undertaken.</td>
<td>11 research universities</td>
</tr>
<tr>
<td>UNITIO, INC.</td>
<td>T1D EXCHANGE</td>
<td>Coordinates a network of clinical care and research centers, combined with a registry, biobank, and social network, offering researchers access to aggregated clinical, biological, patient-reported outcomes, and electronic health record data.</td>
<td>77 clinics, patients, physicians, researchers, and industry representatives</td>
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Many of the participants in the workshop have already convened and led multi-stakeholder, collaborative R&D initiatives with a wide range of objectives and partners. Here is a small sample of such initiatives.
Collaboration Life Cycle: Start-Up

Mission & Governance

• Alignment of goals and expectations

• A formal, transparent governance structure

• How much control a foundation should have in setting collaboration terms

Keeping all parties focused on the mission of the collaboration and accountable for their activities might necessitate a more active role for the foundation.
Collaboration Life Cycle: Start-Up

Human Capital

• Carefully consider how the initiative will be staffed
• Different partners bring different skill sets
Collaboration Life Cycle: Start-Up

Anticipating Future Needs

• Consideration given to the needs, or wants, down the road that can be anticipated or planned for up front

• Importance of bringing in industry, regulators, and even payers early on in planning
Collaboration Life Cycle: Building Relationships

• Building trust
  – Ensuring that all milestones in agreements are decided upon together
  – Understanding the incentives that drive each stakeholder in the collaboration
  – Instilling in the group a sense of purpose and pleasure via team-building

• Data sharing and reuse

• Protection and management of intellectual property

• Using the right tool for the ask

Questions: datadiy4.cnf.io/

Relevant Resources In The ‘Foundations As Collaboration Conveners’ Toolkit

• FNIH Biomarkers Consortium General Intellectual Property and Data Sharing Principles (NIH)
• Parkinson’s Progression Markers Initiatives Research Documents and SOPs (MJFF)
• CHDI Agreement Templates
• Sample Funder’s Addendum: Access to Research Tools (FasterCures)
• Confidential Disclosure Agreement for Research Consortium (MJFF)
• Key Research Agreement Terms and Definitions (FasterCures)
• Venture Philanthropy Legal Report: The Importance of an Interruption License (Schaner & Lubitz)
• 2014 Annual Summit White Paper (One Mind)

For more, go to train.fastercures.org/toolkits
Collaboration Life Cycle: Evaluation and Sustainability

• Defining success
  – It’s important to define success up front, in the service of transparency and accountability

• Maintaining focus
  – Lengthy and large-scale projects can create mission creep

• Surrogate markers of success
  – There have to be measures of successful collaboration other than objective achievement

• Sustainability
  – Redefining the value proposition to engage the biopharma industry’s interest/support

Questions: datadiy4.cnf.io/
Start-Up Challenges and Solutions

**CHALLENGES CITED BY WORKSHOP PARTICIPANTS**

What are the most significant challenges for partners internally and externally that need to be addressed?

- Engaging interest and buy-in from the right partners, including regulators
- Understanding and influencing stakeholder incentives
- Alignment of goals and expectations
- Need for different types of talent, management, and special expertise
- Governance structure
- Managing conflicts of interest for foundations, investigators, and companies
- Length of time to launch

**SOLUTIONS, TOOLS, AND RESOURCES CITED BY WORKSHOP PARTICIPANTS**

What tools and resources would be useful in streamlining the planning and execution of collaborative initiatives by foundations that do not currently exist?

- More thinking about role of foundation in management, governance of collaboration
- Sample organizational guiding principles
- Access to common agreements and common infrastructure (IRBs, trial networks) developed by NIH
- Templates for contracts, protocols, etc.
- Mentoring, peer-to-peer network for information and guidance
- Case examples and contacts of organizations that have done specific types of projects (e.g., trial networks, biomarker initiatives, etc.)
- Governance models, including information about international consortia
- Catalogue of resources that could benefit pre-competitive collaborations (e.g., preclinical animal study databases, etc.)

Questions: datadiy4.cnf.io/
CHALLENGES CITED BY WORKSHOP PARTICIPANTS

What are the most significant challenges for partners internally and externally that need to be addressed?

ESTABLISHING AND MAINTAINING
- Internal resources to manage the initiative
- Due diligence on choosing projects
- Data policies and practices, and allocating resources to support sharing
- IP policies—what is the foundation’s role, what solutions serve objectives
- Maintaining focus, guarding against mission creep
- Quality of science across all partners

SOLUTIONS, TOOLS, AND RESOURCES CITED BY WORKSHOP PARTICIPANTS

What tools and resources would be useful in streamlining the planning and execution of collaborative initiatives by foundations that do not currently exist?

ESTABLISHING AND MAINTAINING
- Good approaches to IP policies and management
- Training in how data sharing works
- Tools for system and stakeholder mapping
- Data and sample sharing policies and agreements
- Model consent language
- Examples of IRB efficiencies (e.g., reliance agreements)
- Antitrust policies for industry in collaborations
- Information about liability insurance for foundations engaged in health research activities
- Toolkit of resources other foundations have developed to address issues
- List of law firms, consultants, etc., who can provide support for tech transfer, statistical analysis, etc.
- Ideas for how to engage more data scientists
- Models for engaging with regulators, e.g., Research Roundtables, drug development tools meetings
**Evaluation and Sustainability Challenges and Solutions**

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**Evaluating and Sustaining**
- Tracking IP and commercialization of funded research
- Getting negative data shared publicly
- Measuring financial and non-financial ROI
- Metrics for organizational success, how you measure your contribution
- Educating and communicating with stakeholders, setting realistic expectations
- Process evaluation in addition to impact evaluation
- Mechanism to sunset large collaborations
- Measuring the impact of the scientific strategy versus the impact of the collaboration—do they need to be separate?
- Demonstrating the value to donors
- Sustainability model with industry

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**Evaluating and Sustaining**
- Examples of evaluation, success criteria
- Develop tools to more easily track IP generated from research funding
- Models for sustainability plans for registry operating costs
Key Takeaways

- Collaboration is necessary.
- Collaboration is not for the faint of heart.
- Foundations are uniquely positioned to be collaboration conveners.
- Investing in a strong framework during the start-up phase is worth it.
- Resources to streamline collaborations exist, but more are needed.

WORKSHOP WISDOM

“If you want to go fast, go alone; if you want to go far, go together.’ We’re asking the question, ‘Can we go quickly together?’”

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Explore TRAIN Toolkits

And Send Us Your Go-to Resources For Building Successful Partnerships

- **TOOLKIT**
  - Foundations as Collaboration Conveners

- **TOOLKIT**
  - Foundation-University Partnerships

- **TOOLKIT**
  - Foundation-Company Partnerships

Questions: datadiy4.cnf.io/
University-Foundation Relations

Bridges gaps in understanding between academic research institutions and patient foundations to streamline the technology transfer process

Tools stakeholders can use, including:

• Model provisions that address early-stage research, commercialization of inventions and royalty sharing

• Foundation-University Partnership toolkit

• Report highlighting key takeaways from TRAIN workshop on moving from transactional to transformative partnerships
Patient Organizations As Research and Data Partners

Questions: datadiy4.cnf.io/
Advancing Models of Patient Engagement

Patient Organizations as Research and Data Partners

• How can patient groups partner with large-scale shared data networks to conduct and support research of interest to patients?

• What data assets can patient organizations create that complement and add value to these networks?

• Partners:
  – People-Centered Research Foundation / PCORnet
  – National Evaluation System for health Technology (NEST) Coordinating Center
  – The Leona M. and Harry B. Helmsley Charitable Trust
  – Michael J. Fox Foundation for Parkinson’s Research
  – Sage Bionetworks (John Wilbanks)
  – > 400 patient organizations surveyed

Questions: datadiy4.cnf.io/
Advancing Models of Patient Engagement: Patient Organizations as Research and Data Partners

PART I: MAKING THE CASE

Advancing Models of Patient Engagement: Patient Organizations as Research and Data Partners

PART II: FOR PATIENT ORGANIZATIONS

Advancing Models of Patient Engagement: Patient Organizations as Research and Data Partners

PART III: FOR RESEARCHERS
Rationale for This Project

• Clinical innovation is changing to become more data- and patient-centric.
• Patient organizations are vital actors in the emerging patient-centered medical research and innovation system.
• Infrastructure for sharing, aggregating and analyzing data from a variety of sources is growing.
• Patient-generated health data represents the next target of opportunity for integration into real-world evidence.
• This type of data is generally not readily available in the environment of shared data networks – but ideally it should be.

What can be done to start to close this gap?

Questions: datadiy4.cnf.io/

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Data Types and Platforms Used by Patient Organizations

Source: Milken Institute
Patient Organizations Are Investing in Health Data

• 88% of survey respondents have supported creation or maintenance of data resources
  – Used primarily for discovery and observational research but also for preclinical and clinical research and post-market surveillance.
  – Include patient registries, online platforms or social networks for patients and caregivers, patient-reported outcomes, biorepositories, natural history, gene sequencing data, and mobile health data collection and/or studies.

• They are investing because:
  – They can aggregate data for a patient population across many institutions and derive unique insights,
  – They have a unique level of trust with their patient communities,
  – They are driven by the interest or request of their patient communities,
  – These data are not being collected and/or shared by providers or researchers, and
  – They need industry-standard information to de-risk investment in treatments for their diseases.
Patient Groups Are Bringing Their Data Assets to Partnerships

• > 75% survey respondents share de-identified patient data with partners, most with academic and industry researchers
• > 75% do not charge a usage fee
• > 50% require committee review of data requests and a data-use agreement
• Many dictate terms regarding ownership and control of the data and the return of results to the foundation and/or to patients.
• Results include publications, basic biological insight, research tools or infrastructure, clinical studies, and preclinical work
• 80% said their data have not been integrated with other sources for research.

“In late 2018, the Food and Drug Administration (FDA) approved an inhaled levodopa powder to treat “off” episodes in people with Parkinson’s disease (PD). The Michael J. Fox Foundation (MJFF) provided “de-risking” funding for early clinical trials of the therapy, the first to reach market approval. MJFF’s decision to fund this and other industry and academic projects aimed at alleviating “off time” was based on patient reports through a large-scale survey that such alleviation is a significant unmet need for their quality of life. MJFF also engages with industry and government partners in a rigorous study of patients’ benefit-risk preferences regarding devices used to treat PD, with the aim of including those preferences in clinical trial criteria.”
## Types and Examples of Data-Sharing Infrastructure

<table>
<thead>
<tr>
<th>Repository</th>
<th>Platform</th>
<th>Network</th>
</tr>
</thead>
<tbody>
<tr>
<td>Data sets are uploaded and made available to qualified researchers for download and secondary use</td>
<td>An environment that enables data sharing and access as well as aggregation and analysis</td>
<td>An infrastructure that links and provides access to data sets and research/analytical services across multiple independent institutions, without data residing in a central repository</td>
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</tbody>
</table>

- dbGaP
- GenBank
- Cancer Imaging Archive
- Yale Open Data Access Project
- Clinical Study Data Request

- Vivli
- Project Data Sphere
- ImmPort
- Synapse
- tranSMART

- PCORnet
- NEST
- Sentinel
- GAAIN
- MDEpiNet Coordinated Registry Networks
Recommendations

If we all agree that patient-generated health data has value and that patient organizations have value as research and data partners, how do we enable more and better collaboration among these stakeholders, particularly in the context of shared data networks?

• Improve the capacity of patient organizations and other stakeholders to partner
• Optimize the development and use of patient-generated health data
• Develop a framework for partnership in the context of shared data networks
Any Partnership Framework Must Include:

- Capacity-building
- Benefit to patients
- Compensation
- Reciprocity

Questions: datadiy4.cnf.io/
Example of a Maturity Model

Big Data Maturity Assessment Dimensions

**ORGANIZATION**
- Leadership
- Funding
- Strategy
- Culture
- Value

**INFRASTRUCTURE**
- Development
- Technologies
- Architecture
- Integration
- Scope

**DATA MANAGEMENT**
- Diversity, volume, speed
- Processing
- Storage
- Quality
- Access

**ANALYTICS**
- Skills
- Mindset
- Techniques
- Applications
- Delivery methods

**GOVERNANCE**
- Policies
- Structure
- Compliance
- Stewardship
- Security and privacy

Source: Transforming Data With Intelligence, tdwi.org
What Does It Mean to Be a “Research-Ready” Patient Organization?

A research-ready organization is one with the:

• Expertise

• Funding strategies

• Engagement with external constituencies, and

• Patient resources

Needed to add distinctive value to the R&D process in service of the needs of the patients they represent.

Questions: datadiy4.cnf.io/

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## Proposed Partnership Maturity Model

<table>
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<th>EXPERTISE</th>
<th>LEVEL I</th>
<th>LEVEL II</th>
<th>LEVEL III</th>
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</thead>
<tbody>
<tr>
<td><strong>Minimal professional staff</strong></td>
<td></td>
<td>Has engaged, non-conflicted SAB</td>
<td>May have business or management advisory board</td>
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<tr>
<td><strong>SAB primarily funded scientists</strong></td>
<td></td>
<td>Has created research roadmap for disease that drives strategies and keeps it up-to-date</td>
<td>May have hired an alliance development person on staff</td>
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<td>Has a Chief Scientific or Medical Officer</td>
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<td><strong>EXPERIENCE</strong></td>
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<tr>
<td><strong>FUNDING STRATEGIES</strong></td>
<td></td>
<td>Funds development of tools and resources</td>
<td>Funds or invests in private companies</td>
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<td></td>
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<td>Funds translational science</td>
<td>Engages in or convenes multi-stakeholder collaborative R&amp;D efforts</td>
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<td>Funds translational science</td>
<td>Willing to accept high risk</td>
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<td><strong>EXTERNAL ENGAGEMENT</strong></td>
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<td>Builds relationships with key stakeholders across ecosystem</td>
<td>Has IP policies for university &amp; company grants</td>
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<td>Has transparent conflict of interest policy for industry relationships</td>
<td>Convenes research roundtables to discuss challenges with key stakeholders</td>
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<td>Has provided input to FDA formally or informally</td>
<td>Has interacted with payers around value of and access to treatments</td>
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<td><strong>PATIENT RESOURCES</strong></td>
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<td>Collects robust natural history data in registry</td>
<td>Has multiple platforms/methods for collecting patient data</td>
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<td>Aids in recruiting patients for trials</td>
<td>Collects data utilizing common data models and standards</td>
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</table>
Your Turn

• What has your journey with collecting and sharing patient data been like?
• What might you have done differently?
• What do you think this journey might optimally look like?
• What inputs would help you improve?
Polling Question
Questions?
Lunch
Data DIY
We’ll be back @
1:15pm
Educational courses centered around rare disease.

RAREUNIVERSITY.COM
Global Genes Question Wall

To participate, please visit https://datadiy4.cnf.io/ with your browser

Note: Responses and submissions are anonymous
Welcome Back
1:15–1:30

Christian Rubio
VP, Strategic Advancement
Global Genes
## Setting The Stage

<table>
<thead>
<tr>
<th>Prader-Willi Syndrome</th>
<th>Cataplexy</th>
</tr>
</thead>
<tbody>
<tr>
<td>A complex genetic condition characterized by chronic hunger (hyperphagia) and obesity.</td>
<td>A sudden and transient episode of muscle weakness with full conscious awareness.</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Narcolepsy</th>
<th>Pitolisant</th>
</tr>
</thead>
<tbody>
<tr>
<td>A chronic sleep disorder characterized by overwhelming daytime drowsiness and sudden attacks of sleep.</td>
<td>The only FDA-approved treatment for people with narcolepsy that is not a controlled substance.</td>
</tr>
</tbody>
</table>
It Starts With The Kids!

“There’s no way I’m working this hard for science. I’m working this hard for kids. We’re doing all of this as volunteers instead of sleeping.”

— Lara Pullen, Ph.D. Chion Foundation
Look Beyond The Obvious

Always question assumptions. Consider repurposing.
Find The Right Team

• Science
• Research
• Technology
• Communication
• Networking
• Medicine
• Fundraising
• Regulatory
Use The Scientific Method

Observation
Social Listening

Hypothesis
Scientific Literature

Experiment
Access/Crowdsource

Conclusion
Publish

Questions: datadiy4.cnf.io/
Listen And Observe

Observation
Social Listening

Questions: datadiy4.cnf.io/

#DataDIY | GlobalGenes.org
REVIEW THE SCIENTIFIC LITERATURE

Hypothesis
Scientific Literature

Questions: datadiy4.cnf.io/
Get Access And Crowd Source Data

- FDA Personal Importation
- Off-label Prescription
- Expanded Access

Questions: datadiy4.cnf.io/
Publish, Publish, Publish

Conclusion

Publish

Questions: datadiy4.cnf.io/

#DataDIY | GlobalGenes.org
Bridge The Gap To FDA

“The FDA is committed to collaborating with patients, caregivers, and advocates, as well as incorporating the various perspectives from these groups into the FDA’s regulatory decision-making processes.”

— Nina L. Hunter, FDA Office of Medical Products and Tobacco
LET’S GO

• Form your team
• Listen to your community
• Think outside the box to form a hypothesis
• Investigate the best way to access drugs
• Crowdsource response to drugs
• Publish
• Take message to the FDA
Polling Question
What More Can You Do with Data?

Clinical Guidelines

2:30–3:30

Paige Nues
Director
Rettsyndrome.org

Dan Glaze, MD
Professor
Baylor College of Medicine/Texas Children’s Hospital

Bernhard Suter
MD, Assistant Professor
Baylor College of Medicine/Texas Children’s Hospital

Questions: datadiy4.cnf.io/

#DataDIY | GlobalGenes.org
What Is Rett Syndrome?

- Rett syndrome is a rare non-inherited genetic neurological disorder that affects ~1:10,000 females (and even more rarely in males)
- Begins to display itself in missed milestones or regression at 6-18 months
- Leads to severe impairments, affecting nearly every aspect of life: ability to speak, walk, eat, digest and breathe easily
- The hallmark of Rett syndrome is near constant repetitive hand movements while awake
- Cognitive assessment in children with Rett syndrome is complicated
- 1983: Becomes widely known as a distinct disorder, clinical diagnosis
- 1999: Causative gene discovered by mutations in X-linked MECP2, encoding methyl-CpG-binding protein 2
- 2020: >95% of classic RTT have MECP2 mutations
The Beginning of Diverse Data Sets: 1980’s

• U.S. Contact registry (International Rett Syndrome Association now Rettsyndrome.org)

• Five (5) Clinics in U.S.
  – Houston, TX (Dr. Daniel Glaze)
  – Birmingham, AL (Dr. Alan Percy)
  – Baltimore, MD (Dr. Sakkubai Naidu)
  – Portland, OR (Dr. Saronjini Budden)
  – San Diego, CA (Dr. Richard Haas)

• Seven (7) Clinics International and Parent Associations
  – Canada (Dr. Patrick MacLeod)
  – Sweden (Prof. Bengt Hagberg)
  – Germany (Prof. Folker Hanefeld)
  – UK (Dr. Alison Kerr and Angus Clarke)
  – Spain (Dr. Merce Pineda)
  – Italy (Dr. Michele Zapella)
  – Japan (Dr. Yoshiko Nomura and Dr. Masaya Segawa)
  – Australia (Dr. Helen Leonard and Dr. John Christodoulou)
Landscape Post-Gene Discovery

• Discovery of causative gene energizes the field
• Brings together Rett syndrome advocacy groups and researchers to
  – Find patients and test for genetic marker
  – Discover mechanism of the disease
  – Discover pathways to treatments and clinical trials
  – Assess disease state: genotype/phenotype
• Five (5) Clinics in U.S.

Questions: datadiy4.cnf.io/
Data: Building a foundation of fundamental knowledge of rare disorders

• 2003 the NIH Office of Rare Diseases Research (ORDR) created the Rare Diseases Clinical Research Network (RDCRN)

• Clinical research important in the value chain to identify cures and treatments for rare diseases

• Address common issues with rare disease research
Data: Building a foundation of fundamental knowledge of rare disorders

• Hurdles
  – Few patients, widely dispersed - challenge for recruitment
  – Few expert centers for diagnosis, management and research

• Solution
  – Clinical research consortia

• As of 2019, NIH awarded $31 million in grants; over 23 consortia representing 200 rare disorders, and $7m for data coordination

Questions: datadiy4.cnf.io/
Cost-Share Research Infrastructure

• Data Management Coordinating Center (DMCC) at the University of South Florida
• All RDCRN protocols are also listed on ClinicalTrials.gov
• Provide technology, tools and support of study disease and data analysis

Questions: datadiy4.cnf.io/
Cost-Share Research Infrastructure

• Provide online protocol management which includes: patient enrollment and randomization for clinical trials, data entry and collection using data procurement standards, adverse event reporting, and protocol training for research staff

• Oversees the RDCRN Contact Registry

Questions: datadiy4.cnf.io/
U54: Rett Syndrome Natural History Study Data Collection

• Build a foundation of fundamental knowledge
• Establish uniform protocols
• A sophisticated database
• Cost-share research infrastructure
• Enlist more centers where the patients are
U54: Rett Syndrome Natural History Study Data Collection

- A sophisticated database
- Cost-share research infrastructure
- Enlist more centers where the patients are
Natural History Study 1: 2003

- Cooperative agreement of 3 disorders: Rett syndrome (RTT), Angelman syndrome and Prader-Willi syndrome
- Focus: Longitudinal natural history
- Data sets unique to each disorder
Natural History Study 1: 2003

• Three sites investigating RTT: Baylor College of Medicine, University of Alabama Birmingham, Greenwood Genetic Center

• Goal: enroll 1,000 Rett syndrome patients
Natural History Study 1: 2006

- Purpose: expand phenotype-genotype studies and set stage for clinical trials
- Principal sites: Baylor, Greenwood Genetic Center, and UAB
Natural History Study 1: 2006

- **Hurdle**
  - Few patients, widely dispersed - challenge for recruitment
  - Few expert centers for diagnosis, management and research

- **Solution**
  - **Travel Clinics** funded by IRSA (now Rettsyndrome.org):
    Oakland CA, Chicago IL, New Brunswick NJ, Tampa/Miami FL

- DMCC: adds Contact Registry
Natural History Study 2: 2009

• Focus: Longitudinal natural history
• Goal: increase enrollment to 1350
• Principal sites: Children’s Hospital Boston, Baylor, Greenwood Genetic Center, and UAB
Natural History Study 2: 2009

- IRSF (now Rettsyndrome.org) continues support for Rett portion and travel sites CA, IL, NJ, FL
- Outcomes: Increase data analysis and publish on Rett syndrome and the emerging related disorders: CDKL5 Deficiency Disorder, FOXG1 syndrome, and MECP2 Duplication Disorder
Natural History Study 3: 2014

• New cooperative agreement for: Rett syndrome, MECP2 Duplication disorder and MECP2 positive, non-Rett individuals, and Related Disorders CDKL5 Deficiency disorder and FOXG1 syndrome

• Collaboration of investigators and patient advocacy groups

• Single comparative data set
Natural History Study 3: 2014

- Principal sites: expand to 11
- IRSF provides support for 3 more sites, and administers grant for entire consortium
- Mechanism: Rare Diseases Clinical Research Network (RDCRN), an initiative of the Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Sciences (NCATS)
  https://ncats.nih.gov/rdcrn
Natural History Studies: Research Goals

• Longitudinal natural history data on Rett syndrome, MECP2 Duplication disorder, CDKL5 Deficiency disorder, and FOXG1 syndrome
• Genotype/phenotype factors affect clinical course and clinical severity
• Event-related potentials (VER and ABR) on disorders
• Blood/saliva samples for DNA, RNA, and metabolomics on disorders
# Natural History Studies

<table>
<thead>
<tr>
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<tbody>
<tr>
<td>• Classic RTT: 955</td>
<td>• Classic RTT: 628</td>
</tr>
<tr>
<td>• Atypical RTT: 166</td>
<td>• Atypical RTT: 81</td>
</tr>
<tr>
<td>• Non RTT: 99 (includes 35 DUP)</td>
<td>• MECP2 DUP: 72</td>
</tr>
<tr>
<td></td>
<td>• MECP2 +,Non RTT: 55</td>
</tr>
<tr>
<td></td>
<td>• CDKL5: 66</td>
</tr>
<tr>
<td></td>
<td>• FOXG1: 58</td>
</tr>
<tr>
<td></td>
<td>• DNA, RNA, and Metabolomic samples: 638</td>
</tr>
<tr>
<td></td>
<td>• Event-related potentials: 152</td>
</tr>
</tbody>
</table>
Important NHS Findings

- Revised diagnostic criteria in collaboration with international experts
- Phenotype-genotype interactions
- Impaired growth and development
- Impaired GI function
- Scoliosis prevalence and surgical impact
- Sexual maturation
- Prolonged longevity
- Epilepsy prevalence and trends
- Awake breathing patterns and trends
NHS Findings (continued)

• Hand stereotypies
• Quality of Life Assessments
• Qualitative Rating Scales
• Behavioral profiles in RTT
• Mutations in males
• Other genes producing RTT features
• Gallbladder Dysfunction in RTT
• Vitamin D Deficiency in RTT
• Caregiver burden for RTT
• Metabolomic Profiles in RTT
NHS Findings (continued)

- Phenotypic Effect of Xq28 Duplication
- Severity Assessment in CDKL5 Deficiency
- Scoliosis: Comorbidities and Predictors
- Changing face of survival in RTT
- Age of diagnosis in RTT
- Cardiac arrhythmias in RTT
- Severity Assessment in CDKL5 Deficiency
- Scoliosis: Comorbidities and Predictors

TWENTY-EIGHT outcomes increase Rett syndrome insights
NHS Findings (under review)

- Motor Behavioral Assessment factor analysis
- Comparison of epilepsy among all four disorders
NHS Findings (in preparation)

- Anxiety in RTT
- Primary Care Guidelines in RTT
- Event related potentials: all four disorders
- Anthropometrics in RTT
- Hand function in RTT
- Sleep disorders in RTT, Prader-Willi, and Angelman syndrome
- Dystonia with aging in RTT
- Ambulation in RTT
- Psychopharmacologic usage in all four disorders
- Parental age risk for MECP2 mutation
- AED usage in RTT
Outcome: Resource-rich, research-ready

• Industry takes notice

• Stage set for clinical trials
  – Build a foundation of fundamental knowledge
  – Establish uniform protocols for clinical data
  – A sophisticated database
  – Cost-share research infrastructure
  – Enlist more centers where the patients are
  – Formalize network of clinical providers
Outcome: Clinical Guidelines and the P3 Learning Collaborative

- **P3 - Project, Process, Publish**
- Develop evidence-based guidelines and quality indicators
- Four focus areas 2019-2020:
  - *Communication, Epilepsy/Rett spells, GI, Prolonged QT (cardiac)*
- Led by Key Thought Leaders
- Progress shared with overall Learning Collaborative (monthly)
- Progress reported to North American Clinic Discussion Group (quarterly)
Natural History Data and the Evolution of Rett Related Disorders

Bernhard Suter, MD
Assistant Professor
Baylor College of Medicine / Texas Children’s Hospital
NIH Natural History Study

• Longitudinal natural history data on Rett syndrome
• Later also on Rett related disorders: MECP2 Duplication Disorder, CDKL5 Disorder, and FOXG1 Disorder
"The clinical Pattern of the Rett Syndrome"

Pasky Hanefeld, MD

A case of infantile spasms who later developed many characteristics of Rett syndrome

Seizures at the age of one year were her first clinical symptom

Progressive encephalopathy, stereotyped movements, ataxia and microcephaly

At 2 years hand stereotypies (wringing-washing) appeared

The Clinical Pattern of the Rett Syndrome

Pasky Hanefeld, MD

A case of infantile spasms who later developed many characteristics of Rett syndrome is reported.

Seizures at the age of one year were her first clinical symptom. Progressive encephalopathy, stereotyped movements, ataxia and microcephaly appeared at 2 years. Hand stereotypies (wringing-washing) appeared.

[Hanefeld 1985]
CDKL5 as Rett variant

CDKL5/STK9 is mutated in Rett syndrome variant with infantile spasms:

• In 1985, Hanefeld described a variant with early appearance of seizures.
  – Normal perinatal period, followed by emergence of seizures (infantile spasms).

• Two patients with signs of Rett syndrome: acquired microcephaly and stereotypic midline hand movements.

• Generalized convulsions and myoclonic seizures at 1.5 months in the first patient and spasms at 10 days in the other

• Suggesting a diagnosis of the Hanefeld (early seizure) variant.

• Found frameshift deletions in the CDKL5 gene in both patients
The CDKL5 disorder is an independent clinical entity associated with early-onset encephalopathy

• Most individuals with CDKL5 mutations have severe developmental delay from birth*, seizure onset before the age of 3 months

• Less than one-quarter (of n=86) met criteria for early-onset seizure variant RTT.

• Seizures and sleep disturbances are more common

• Features of regression and spinal curvature are less common

[Fehr, 2013]
Implications

• **Clinical guidelines** distinct for each disorder:
  – e.g. current Rett guidelines have sections on scoliosis management, that likely would not be such a prominent issue for CDKL5

• **Clinical severity scales**
  – e.g.: “Severity Assessment in CDKL5 Deficiency Disorder” [Demarest, 2019]
  – Severity assessments are instructive for drug studies
Many “Rett related” genes

- Patients who were diagnosed with Rett syndrome or Rett-like syndrome because of their phenotype
- Negative for mutations in the MECP2, CDKL5 or FoxG1 genes
- Whole exome sequencing revealed 69 novel causal genes
- Much work to be done, to accurately distinguish between them, establish clinical baselines, develop clinical guidelines, etc.
“Current clinical evidence does not support a link between TBL1XR1 and Rett syndrome.”

We reviewed the clinical presentation of TBL1XR1 prompted by one Rett-like patient, and NO other patient did fulfill Rett criteria

- Based on the revised clinical criteria our patient was diagnosed with typical RTT
- Review of published cases > none met criteria for the diagnosis of either typical or atypical Rett syndrome.
- None of the cases in the DECIPHER Database fit Rett
- In fact, different mutations in TBL1XR1 appeared to result in differing phenotypes, rather than a single clearly defined clinical entity
Conclusion

• Commit to data and data harmonization
• Adapt as funding needs change
• Seek common ground with related disorders and leverage
• Listen to research data to inform and improve patient care through published guidelines
Polling Question
Questions?
Break
Data DIY
We’ll be back @
3:45am
Global Genes Question Wall

To participate, please visit https://datadiy4.cnf.io/ with your browser

Note: Responses and submissions are anonymous
Expanding Your Engagement to Underserved Populations

Panel Discussion
3:45–4:45

Liz Horn, PhD, MBI
Principal
LHC Biosolutions

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Our Panelists

Linda Wade
President & CEO
Sickle Cell Association of Texas, Marc Thomas Foundation

Jordan Shumway
Social Worker
Sickle Cell Association of Texas, Marc Thomas Foundation

Khrystal K. Davis, JD
Founder
Texas Rare Alliance

Robyn Correll Carlyle, MPH, CHWI
Dia de la Mujer Latina, Inc. (DML)
Linda Wade
President & CEO
Sickle Cell Association of Texas, Marc Thomas Foundation

Jordan Shumway
Social Worker
Sickle Cell Association of Texas, Marc Thomas Foundation
HISTORY OF THE ORGANIZATION

• The Sickle Cell Association of Texas Marc Thomas Foundation started in 1997 by a husband and wife who wanted to advocate for families suffering from sickle cell disease

• Pastor Marc Thomas suffered from sickle cell disease, pulmonary hypertension, congestive heart failure and wore oxygen 24 hours a day

• He passed away from sickle cell disease at the age of 46 but his wife and daughter continue his legacy and mission
What is Sickle Cell Disease?

• Sickle Cell Disease is a genetic disorder that affects red blood cells.

• The CDC reports that Sickle Cell Disease occurs in approximately 1 of every 365 African-American births and 1 in every 16,300 Hispanic-American births. Approximately 100,000 Americans are affected.
Accomplishments

• Successfully assisted and lobbied for legislation in Texas to begin testing all newborns for Sickle Cell Trait regardless of race.

• Two Board Members served on the Texas Sickle Cell Advisory Committee and two are on the Sickle Cell Task Force

• Met with Former President Bush to discuss Sickle Cell issues. Appointed under the President Bush Administration as a United States Health and Human Services Advisor on Blood Safety and Availability.

• Received an exclusive White House Oval Office invitation to meet with President Obama to discuss Sickle Cell Disease issues in his first 100 days in office.
Client Demographics

Understanding our Sickle Cell Community in Texas: Region

Understanding our Sickle Cell Community in Texas: Age Groups
Office Locations and Staff

- Licensed Clinical Social Worker/Certified Case Manager, Licensed Social Workers, Community Health Workers
- All staff are Certified Hemoglobinopathy educators and are HIPAA certified
- Bilingual Services (Spanish)
- Office Locations in Austin, Houston, San Antonio, and Dallas
- Serve clients in 52 Texas counties, 17 of which are rural
Services and Programming

- We serve more than 750 hundred clients with:
  - Certified Case Management, Medical Case Management, Referrals, SSI/SSDI Assistance, Housing, Transportation assistance and more
  - Counseling, Transition Services, Care Coordination, Medical Home Placement, Scholarship Program, Support & Limited Financial Assistance
  - Camp Cell-A-Bration, Camp Next Level, and ESCAPE Retreat
  - SOS Support Group Meetings and free Sickle Cell Testing
  - Sickle Cell Empowerment Conferences, Walks, and Events
• Patient surveys to assess symptoms, severity, and ongoing needs
• Demographic data on clients served to assess needs and future planning
• Pre- and post-surveys at Camps to assess the educational impact
Contact Information

info@sicklecelltx.org
1-844-994-HOPE
Austin: (512) 458-9767
Houston: (713) 534-1712
San Antonio: (210) 447-7553

www.sicklecelltx.org
Khrystal K. Davis, JD
Founder
Texas Rare Alliance
Texas Rare Alliance Mission

Working to improve access and health outcomes for Texas rare disease patients through education and advocacy
Texas Rare Alliance Meetups

Meetups for stakeholders in the Texas rare disease community

- Dallas-Fort Worth
- Austin (Webcast)
- San Antonio
- Houston
  - English
  - Spanish (Webcast)
Underserved Populations in Texas

• 38% Latino
• 8% cannot speak English well or at all
• 12% African American
• 18% have no health insurance
• 13% live below the poverty line
• 30% of counties have no hospitals
• 13% of counties have no doctors
Newborn Screening Advocacy in Texas

2019 Texas Legislative Session – Texas Rare Alliance Advocacy

• Newborn Screening Deficit Funding
• Newborn Screening Preservation Account
Newborn Screening in TX

• NBS is the best opportunity to combat socioeconomic disparities and democratize the diagnosis
• Texas NBS program screens 400,000 babies a year twice for 54 genetic conditions
• NBS in Texas is an overwhelming success for newborns with screened conditions, but there are more than 7,000 rare diseases
• We are not even close to screening for 1% of the rare diseases through newborn screening
Rare Disease Diagnosis in Children

• It takes 5-7 years to obtain an accurate rare disease diagnosis
• Many children with a rare disease will not survive to their 5th birthday
• We cannot continue to allow babies and children to die without obtaining an accurate rare disease diagnosis and treatment whenever possible
• More than 300 additional conditions would qualify for inclusion on the RUSP, but lack an approved genetic test
Project Baby Dillo

• Follow the success of Project Baby Bear in CA
• Pilot study provided rWGS of NICU & PICU Medi-Cal patients
  – Proven health outcomes
  – Proven economic benefits

“rWGS is faster, better and cheaper than traditional treatments. It has led to vastly improved outcomes for the babies themselves and it saved taxpayer dollars.” – Village News, CA Assembly Member Marie Waldron
Project Baby Dillo

- Project Baby Dillo would provide WGS to NICU and PICU patients insured by Texas Medicaid with unknown etiologies
- Ability to diagnose and treat many more of the 7,000 rare diseases
- Texas Rare Alliance will provide caregiver experience and preference data on access to the diagnosis
- Data from Project Baby Bear, Project Baby Dillo and others will provide data to support the implementation of newborn WGS
Project Baby Dillo Next Steps

• Recruit advocacy partners to join the Project Baby Dillo Task Force
• Recruit Hospitals to participate in Project Baby Dillo
• Identify a partner to perform WGS
• Conduct caregiver surveys to obtain PED and PPD on access to the diagnosis
• Hold advocacy events to educate members of the rare disease community, HCPs, Representatives, Senators, and their health policy staff.
• Secure sponsors for Bills in the Texas House & Senate to provide funding for Project Baby Bear
Project Baby Dillo

- Hunter was admitted to the NICU at birth with respiratory failure
- 11-day NICU stay
- Discharged without a diagnosis
- Diagnosed with Spinal Muscular Atrophy Type 1 - the leading genetic cause of death in infants.
- SMA now has approved treatments
Robyn Correll Carlyle, MPH, CHWI
Dia de la Mujer Latina, Inc. (DML)
Día de la Mujer Latina

A grassroots, community-based organization dedicated to:

• Culturally and linguistically proficient education
• Facilitating early detection screening
• Culturally preventative care interventions
• Promoting wellness with resource information
• Promotores/Community Health Workers (P/CHW) training
• Patient Navigation for follow-up services
• Training CHWs to be clinical trial navigators for under-represented populations
Community Health Workers

Impact

From Seattle, Washington to Miami, Florida, DML has left its health imprint at more than 39 cities across the nation.
### Promotor (a) /CHW Curriculum

#### Teaching Skills
- Plan + Effect Presentation
- Teach on “How to talk” to your doctor
- Teaching skills for behavior change

#### Communication Skills
- Understanding Health Literacy
- The art of observing, listening, communicating effectively

#### Service Coordination Skills
- Medicaid/CHIP/CHIP Perinatal
- Understanding the role of community health centers
- Affordable Care Act
- Patient Navigation

#### Advocacy Skills
- Understanding HIPAA
- How to advocate about health issues for Latinos or other minority groups
- Learn about the Legislative process

#### Interpersonal Skills
- Cultural Competence
- How to motivate your patient & their family about follow up
- Informal counseling

#### Capacity-Building Skills
- Build community resiliency by promoting prevention.
- Disaster Preparedness-
- Community Emergency Response Team (CERT)

#### Organizational Skills
- How to prepare for a Health Fiesta or cultural event
- How to prepare and analyze Pre and Post Surveys with SWOT

#### Knowledge Base on Specific Health Issues
- Breast Cancer & Survivorship
- Cervical Cancer - Human Papilloma Virus (HPV)
- Heart and Stroke
- Diet & Nutrition
- Autoimmune Diseases

#### Knowledge Base - Specific Health

- HIV/STD
- Diabetes
- Mental Health First Aid
- Behavioral Health
- Viruses & Vaccines

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DML’s Community Health Worker/Promotores Outreach & Training

- Health Fiestas
- Patient Navigator
- Telehealth Navigators
- Clinical Trial Navigator
- Medical Providers
- Personal-Care Navigator
- School-based Navigator
- Disaster Recovery

Triage at Community Health Centers

Health Fiestas

Patient Navigator

Telehealth Navigators

Clinical Trial Navigator

Disaster Recovery

Medical Providers

Personal-Care Navigator

School-based Navigator
Promotores/CHWs as Clinical Trial Navigators

Program Goal

• Train Promotores/CHWs to work with researchers to recruit and retain populations currently under-represented in clinical trials.

• Recruit those who complete the certified training as Clinical Trial Navigators to participate in a variety of research opportunities.
For Further Questions or Information:

Venus Ginés, MA, CHWI
281-489-1111
www.diadelamujerlatina.org
president@diadelamujerlatina.org

Robyn Correll Carlyle, MPH, CHWI
robyn@adjuvantmedia.com
Polling Question
Questions?
What's Next?

4:45–5:00

Thanks and Goodbye to Our Online Guests
Wrap Up
Educational courses centered around rare disease.

RAREUNIVERSITY.COM
NEXT:

IMAGINING THE FUTURE OF RARE DISEASE

Download a copy at GlobalGenes.org/Next-Report
STAY CURRENT WITH RARE DISEASE

GLOBAL GENES™
RARECast™
with Daniel Levine

RAREDaily
by Global Genes

globalgenes.org/rare-daily
RARE PATIENT ADVOCACY SUMMIT

SAVE THE DATE!

September 21-23, 2020
Sheraton San Diego
Marina Hotel

GLOBALGENES.ORG/PASUMMIT