



RARE PATIENT ADVOCACY SUMMIT
PRESENTED BY GLOBAL GENES
SEPTEMBER 24-25, 2015

SUMMIT OVERVIEW

A rare diagnosis changes everything. It crashes plans and dreams, knocks you off your feet, and requires a continual investment of time and money as you try to determine what should be your next step. The purpose of the RARE Patient Advocacy Summit is to help patient ADVOCATES become successful ACTIVISTS and provide the discussion, insights, and tools to move down this advocacy path, equipped and prepared.

The Summit will offer practical advice, case studies, and networking opportunities as we learn from one another. The goal is to have patient advocates walk away with a better understanding of the challenges they will face and ways they can be the most effective in advocating for their disease/disorder.

You will:

- Hear directly from experts
- Better understand how to overcome the challenges facing caregivers
- Learn about new advances in science
- Learn ways to become an unstoppable advocate and an effective organization
- Become equipped and educated on successful lobbying
- Focus on drug development and how patients are playing a critical role
- Network, Network. Network!
- Have fun connecting with other advocates who share your passion for advocacy

By attending this two-day event, you will meet new friends and colleagues, gain a new perspective on the complexities and questions that need to be considered in order to become effective advocates for rare disease, and be prepared to help make advances in the rare disease space.

SUMMIT DAY 1:

7:00 BREAKFAST BUFFET

Sponsored by Genzyme

8:00 WELCOME & MEETING KICK OFF

Nicole Boice, Introduces Summit Advisory Board

8:15 OPENING KEYNOTE

Bonner Paddock – Ironman Competitor, Marathoner, Extreme Climber, Author, and Patient Advocate, Cerebral Palsy

9:00 PRESENTATION - BUILDING A GAME PLAN

Building a Game Plan – Beginning with the end in mind, otherwise the journey looks impossible

Presenter: Pat Furlong, Founding President and CEO, Parent Project Muscular Dystrophy

9:45 BREAK

Sponsored by Aegerion Pharmaceutical

10:00 FIRESIDE CHAT

THE 10 THINGS YOU NEED TO DO ONCE YOU HAVE A DIAGNOSIS

Presented by Caregiver Action Network and Partners on the Path

What are my next steps?

Discussion with Matt Wilsey, Parent Advocate, NGLY1 and President of Grace Wilsey Foundation interviewed by Daniel Levine, Global Genes' RARECast, Principal, Levine Media Group

11:00 INTERACTIVE SESSION

THE 10 THINGS YOU NEED TO PURSUE RESEARCH FOR YOUR DISEASE?

What are the options for research and why is it so hard?

Presenter: Brad Margus, Parent Advocate, Founder and Chairman, A-T Children's Project, Founder and CEO, Exigence Neurosciences

12:00 LUNCH OUTSIDE COURTYARD

Sponsored by Alexion Pharmaceuticals

1:15 PANEL PRESENTATION

SEQUENCING, DATA SHARING & CONSENT:

Understanding your genetic information, owning your data and sharing with others to drive research and drug development

Moderator: Peter Goodhand, Executive Director, The Global Alliance for Genomics and Health

Panelists: Denise Perry, Senior Genetic Counselor, Illumina Clinical Services Laboratory

(Sequencing 101), Megan Doerr, MS, LGC, Principal Scientist, Governance Sage Bionetworks (Consent),

David Glazer, Director of Engineering, Google Genomic

2:15 TRANSITION TO TRACKS





	TRACK 1 DEEP DIVE TRACK – SEQUENCING	TRACK 2 DEEP DIVE TRACK – DATA AND CONSENT	TRACK 3 DEEP DIVE TRACK – RARE DISEASE HOT TOPICS	TRACK 4 DEEP DIVE TRACK – SCIENCE BRIEFS
2:30	<p>SEQUENCING DEEP DIVE 1: FROM DATA TO MEANING</p> <p>Why your medical information/genetic data is critical to helping find treatments. What can researchers do with more information?</p> <p>Hosts: David Glazer, Director of Engineering, Google Genomics; Anthony Philippakis, Cardiologist, Brigham and Women’s Hospital Research Scientist, Broad Institute of MIT and Harvard, Venture Partner, Google Ventures</p>	<p>DEEP DIVE 1: INDIVIDUALS – WHERE DO I PUT MY DATA AND WHAT DOES IT MEAN?</p> <p>The benefits of owning medical data – The Do’s, the Don’ts and other considerations.</p> <p>Host: Liz Horn, PhD, Co-Principal Investigator, Phelan-McDermid Syndrome Data Network</p>	<p>RARE DISEASE HOT TOPICS 2: CLINICAL TRIAL TRAINING – PRE-DRUG DEVELOPMENT</p> <p>When and how patients should become engaged with industry. Tools and strategies to take back to your community.</p> <p>Host: Bray Patrick-Lake, BS, MFS, Director, Stakeholder Engagement, Clinical Trials Transformation Initiative, Duke University</p>	<p>SCIENCE DEEP DIVE 1: WHAT IS EVOLUTIONARY PHARMACOLOGY AND HOW IS IT SPURRING RESEARCH?</p> <p>Host: Ethan Perlstein, PhD, Host, Founder and CEO, Perlstein Labs</p>
3:20	<p>BREAK OR GRAB A SNACK – GO TO A SHORT LEARNING SESSION</p>			
	<p>GRAB & GO SESSION: INNOVATIONS IN CARRIER SCREENING</p> <p>Host: Shivani Nazareth, Genetic Counselor and Director of Women’s Health, Counsyl</p>	<p>GRAB & GO SESSION: RARE DISEASE RESEARCH MICROGRANTS</p> <p>Host: David Cox, Executive Director, Rare Disease Foundation</p>	<p>GRAB & GO SESSION: SBIR GRANTS AND INTERESTING FUNDING PARTNERSHIPS</p> <p>Host: Timothy McDaniel, PhD, Senior Vice President of Emerging Opportunities, Translational Genomics Research Institute</p>	<p>GRAB & GO SESSION: CROWDMED – PATIENT RELIEF FUND</p> <p>Host: Jared Heyman, Founder and CEO, CrowdMed</p>
3:40	<p>SEQUENCING DEEP DIVE 2: THE IMPORTANCE OF SHARING GENETIC DATA WITH RESEARCHERS</p> <p>Host: Dr. Matt Huentelman, Associate Professor in the Neurogenomics Division, Translational Genomics Research Institute</p>	<p>DEEP DIVE 2: HOW DO YOU SHARE YOUR DATA FOR RESEARCH AND WHO OWNS WHAT?</p> <p>Host: James O’Leary, Chief Innovation Officer, Genetic Alliance</p>	<p>RARE DISEASE HOT TOPICS 2: PCORI</p> <p>Host: Suzanne Schrandt, Deputy Director Patient Engagement, Patient-Powered Research Networks</p>	<p>SCIENCE DEEP DIVE 2: NEW MODELS FOR DRUG DISCOVERY</p> <p>Host: Chris Gibson, Co-Founder and CEO, Recursion Pharmaceuticals</p>



-
- 4:40** **SEQUENCING DEEP DIVE 3: INCIDENTAL FINDINGS - THE ETHICAL DEBATE**
Host: Stephanie Gandomi, Genetic Counselor & Medical Affairs, Invitae
- DEEP DIVE 3: CREATING YOUR CONSENT TO SHARE YOUR DATA**
Navigating the different options for sharing data.
Host: Megan Doerr, Principal Scientist of Governance Team, Sage Bionetworks
- RARE DISEASE HOT TOPICS 3: ACCESS TO TESTING: Insurance, Billing, Reimbursement**
Host: Shannon Stasi, Licensed Genetic Counselor, Department of Laboratories and Pediatric Laboratory Utilization Guidance Services, Seattle Children's Hospital
- SCIENCE DEEP DIVE 3: PRECISION GENOME - GUIDED MEDICINE**
Host: Reid Robison, CEO and Co-Founder, Tute Genomics
-

5:30 **END OF DAY ONE**

- 5:45** **SPECIAL SHORT FILM SCREENING: THE LION'S MOUTH OPENS**
Post Screening Discussion with Marianna Palka, Writer, Producer, and Patient Advocate, Huntington's Disease and Adriana Venegas, Patient Advocate, Huntington's Disease
-

FRIDAY, SEPTEMBER 25, 2015

SUMMIT DAY 2:

- 7:30** **BREAKFAST BUFFET**
Sponsored by Walgreens
-
- 8:30** **WELCOME AND DAY 2 KICKOFF**
(10 Minute Ask and Community Call): 21st Century Cures – Lisa Schill, Parent Advocate and Vice President, RASopathies Network USA and Max Bronstein, Senior Director of Public & Government Affairs, EveryLife Foundation
-
- 8:45** **PRESENTATION: MAPPING DRUG DEVELOPMENT: THE ROLE PATIENTS PLAY**
Presented by Gayatri Rao, Director, Office of Orphan Products Development, FDA
-
- 9:45** **BREAK AND TRANSITION TO TRACKS**
Sponsored by Retrophin
-





	TRACK 1 DEVELOPING THERAPIES, WHERE PATIENTS PLAY	TRACK 2 THE RARE JOURNEY - DIAGNOSIS TO TREATMENT. STORYTELLING TRACK <i>Sponsored by: The Patient Experience Project</i>	TRACK 3 RARE DISEASE HOT TOPICS: WHAT CAN YOU DO!	TRACK 4 SUPPORT & ORGANIZATIONAL GROWTH
10:00	ACADEMIC RESEARCHERS - UNDERSTANDING THEIR MOTIVATION: Where do patients fit? Hosts: Christian Rubio, Senior Director Digital Marketing, Platform Q Health; Christina Waters, Chief Executive Officer and Founder, RARE Science	THE JOURNEY TO DIAGNOSIS: Host: Caroline Loewy, Founder and Treasurer, KCNCQ2 CURE Alliance; Parent Advocate; Board Member, Global Genes; Board Member, TGEN Center for Rare Childhood Disorders	CAREGIVING IN THE US: Landmark Study Results & introduction to Rare Disease Care-giving Study Host: Grace Whiting, Director of Strategic Partnerships, National Alliance for Caregiving	EARLY STAGE NPO PLANNING: Where to start/restart - begin with the end in mind. Host: Mary Cobb, Principal, Patient Vue
11:00	INDUSTRY - UNDERSTANDING THEIR MOTIVATION: Where do patients fit? Who do patients talk to? Host: Susan Stein, CEO, Connexion Healthcare	THE JOURNEY TO DEVELOP A TREATMENT: Hosts: Barbara Wuebbels, Vice President of Patient Advocacy, Audentes; Erin Ward, Co-Founder and President, MTM-CNM Family Connection, Inc.; Holger Fischer, Founder, ZNM-Zusammen Stark!; Alison Rockett Frase, Founder and Vice President, Joshua Frase Foundation; Anne Lennox, Trustee, Myotubular Trust	CLINICAL CARE - PATIENTS DESERVE THE BEST CARE: Building Comprehensive Care Centers Hosts: Kelly Ranallo, Founder and President, Turner Syndrome Alliance; Dina Matos, Executive Director, CARES Foundation; Stephanie Okey, Former Head of North America and SVP, Rare Diseases business and General Manager US, Genzyme	ENSURING ORGANIZATIONAL GROWTH: Creating Organizational Longevity Host: Jean Campbell, Principal, JFC Consultants
12:00	LUNCH OUTSIDE COURTYARD Sponsored by Shire			
1:10	GOVERNMENT, NIH - WHAT ARE THEIR MOTIVATIONS: How to engage with the NIH? Navigating the Institutes. Host: PJ Brooks, Division of Clinical Innovation and Office of Rare Diseases Research at National Center for Advancing Translational Sciences (NCATS), NIH	DRUG APPROVALS, THE RARE JOURNEY TO APPROVAL: How can you bring your voice to the FDA approval process to effectively communicate with Advisory Panels and FDA decision makers? Hear stories from patients and advisors who have navigated the final step to drug approval, participated in town hall ▶	SHOW ME THE MONEY! Where to go and when to ask for it when looking to fund research! Hosts: Deb Geraghty, VP Commercial Strategy, Dimension Tx; Alison Silva, Executive Vice President, Chief Operating Officer and Co-Founder, The Orphan Group	POWERING UP YOUR CARE TEAM: Building & sharing your knowledge base with your healthcare team, communities. Hosts: Todd Kozikowski, CEO, Yabidu; Lisa Schill, Parent Advocate and Vice President, RASopathies Network USA



1:10	GOVERNMENT, NIH - WHAT ARE THEIR MOTIVATIONS:	DRUG APPROVALS, THE RARE JOURNEY TO APPROVAL:	SHOW ME THE MONEY	POWERING UP YOUR CARE TEAM:
		meetings, letter writing campaigns. Learn best practices and build confidence in your storytelling skills!		
		Hosts: Joff Masukawa, President, Diligentia LLC Eileen Crowley, Communications & Advocacy Specialist, 3D Communications Tracy VanHoutan, Co-founder, Noah's Hope Melissa Hogan, Founder and President, President Saving Case & Friends, Inc.		
2:10	GOVERNMENT, FDA - WHAT ARE THEIR MOTIVATIONS:	MANAGING DATA AT THE ORGANIZATIONAL LEVEL		LEGISLATIVE UPDATE: 21ST CENTURY CURES
	How to engage with FDA & tools for approval (Breakthrough designation, accelerated approval)	Navigating the different options for sharing data.		How public policy and legislation impact the patient's ability to access treatments and the availability of treatments.
	Host: Carrie Jones, Health Communications Strategist, JPA Health Communications	Host: Liz Horn, PhD, Co-Principal Investigator, Phelan-McDermid Syndrome Data Network		Hosts: Max Bronstein, Senior Director of Public & Government Affairs, EveryLife Foundation; Jen Bernstein, Vice President, Horizon Government Affairs; Lisa Schill, Parent Advocate and Vice President, RASopathies Network USA
3:15	RARE TRIBUTE TO CHAMPIONS OF HOPE - FRIDAY AWARDS			
5:30	SUNSET COCKTAIL PARTY- OUTSIDE COURTYARD			
	<i>In partnership with Mauli Ola Foundation Sponsored by Takeda</i>			
