RARE DISEASE SCIENTIFIC WORKSHOP

COVID-19 Mitigation Strategies in Rare Disease Therapy Development & Delivery: Exchanging Best Practices – Moving to Permanent Solutions

Virtual Event · December 15, 2020

Registration Link: https://rarediseases.swoogo.com/scientificworkshop (password: SW2020)

Event Platform Link: https://eventmobi.com/rdsw/

10:00-11:15AM

When the World Tilted: Reflecting on the Forced Evolution of COVID-19 and Applying Innovations to a New Normal

Moderator: Emil Kakkis, MD, PhD – President & CEO, Ultragenyx & Founder & Member of the Board of Directors, EveryLife Foundation for Rare Diseases

Jenn McNary – Rare Mom, Patient Advocate, Co-Founder & Manager, One Rare & J. McNary Consulting

Kristie E.N. Clarke, MD, MSCR, FAAP, CDR – US Public Health Service, Team Lead, Disproportionately Affected Populations Team & Medical Epidemiologist Centers for Disease Control and Prevention

Janet Maynard, MD, MHS – Director, Office of Orphan Drug Products Development, US Food and Drug Administration

Christopher P. Austin, MD – Director, National Center for Advancing Translational Sciences, National Institutes of Health

Jean Moody-Williams, RN, MPP – Deputy Director, Center for Clinical Standards and Quality, Centers for Medicare and Medicaid Services

Stu Peltz, PhD – Chief Executive Officer, PTC Biotherapeutics

11:15-11:30AM

BREAK

11:30-12:45PM

CASE STUDY BREAKOUT SESSIONS – PLEASE CHOOSE ONE OF THE FOLLOWING:

'Size Matters' - Collecting Outcome Measures During the Pandemic Through Innovative Technology in Clinical Trials Moderator: Anne Pariser, MD – Director Office of Rare Disease Research, NCATS	**Clinical Trials 'On the Move': Deploying Innovations in Home Health & Mobile Services** Moderator: Isabelle Lousada – Founder & CEO, Amyloidosis Research Consortium	**Learning from the Trenches: Assessing Patient Experiences & Preferences During COVID-19** Moderator: Annie Kennedy – Chief of Policy & Advocacy, EveryLife Foundation for Rare Diseases
Cure VCP Disease Inc. & Nationwide Children's Hospital: *Using tele-research to kick-off functional assessment testing* Speakers: Sujata Patel, RPh & Lindsay Alfano, PT, DPT, PCS BridgeBio, Mass General, NYU Langone & UKE Hamburg: *Natural history conduct during COVID-19* Speaker: Rafael Escandon, DrPH, PhD, MPH Casimir: *Remote monitoring of disease progression in Duchenne Muscular Dystrophy* Speaker: Mindy Leffler Myasthenia Gravis Rare Disease Network (MGNET) & George Washington University: *Adapting disease specific outcome measures pilot trial for telehealth in Myasthenia Gravis (ADAPTteleMG)* Speaker: Amanda Guidon, MD	Homology Medicines, Premiere Research & Illingworth Research Group: *Implementing home health services in the first-ever PKU gene therapy clinical trial during the pandemic* Speaker: Joseph Donahue AGTC: *Coronavirus and clinical trials: AGTC provides support for the Rare Inherited Retinal Disease trial participants amid the pandemic* Speaker: Jill Dolgin, PharmD Roche/Genentech: *Ensuring treatment continuity for SMA patients in Risdiplam clinical trials during COVID-19 Pandemic* Speakers: Coralie Nguyen, PhD & Sibylle Fuhrer, PharmD Applied Therapeutics: *ACTION-Galactosemia clinical study – staying in action during COVID-19* Speaker: Shoshana Shendelman, PhD	The EveryLife Foundation for Rare Diseases: *Results from the US rare disease COVID-19 vaccine survey* Speaker: Jamie Sullivan, MPH X4 Pharma & Savvy Coop: *X4 asked patients* Speakers: Michele Rhee, MBA, MPH & Jen Horonjeff Parent Project Muscular Dystrophy: *An assessment of the Duchenne community's experience of the COVID-19 pandemic: Impact on care, clinical trials and access to approved therapies* Speaker: Ryan Fischer

12:45-1:15PM	LUNCH BREAK		
1:15-2:30PM	**CASE STUDY BREAKOUT SESSIONS – PLEASE CHOOSE ONE OF THE FOLLOWING:**		
	How Rare Disease Leaders Have Kept Life-Saving Research Momentum Strong During the Pandemic Moderator: Kristin Van Goor, PhD – Senior Director, Regulatory Policy & Intelligence, Vertex	**'In the Eye of the Storm': An Inside Look into State Health Laboratories & Newborn Screening Programs** Moderator: Amy Gaviglio, MS, CGC – Public Health Genetics Consultant, APHL & Expecting Health	**Rare Disease & COVID Urgency Aren't Mutually Exclusive: Ensuring Continuity of Care** Moderator: Sarah-Lloyd Stevenson – Director, Faegre Drinker
	BIO & Vertex: *An overview of industry experience from the BIO decentralized trial task force* Speaker: Kristin Van Goor, PhD Sanofi Genzyme: *A wholistic, patient centric & COVID-19 resilient approach to a rare disease clinical trial* Speaker: Kimberly Hawkins, MPH Sarepta: *Model-informed clinical trial mitigation for Duchenne Muscular Dystrophy* Speakers: Erica Koenig, PhD & Lilly East, PhD	APHL and Expecting Health: *An overview of COVID-19's impact on state newborn screening programs* Speaker: Amy Gaviglio, MS, CGC Iowa Department of Public Health: *Maintaining newborn screening quality during the COVID-19 pandemic & a natural disaster* Speaker: Kimberly Noble Piper, RN, BS, CPH, CPHG New York State Newborn Screening Lab & Northwell Health: *Continuation of a pilot study and patient follow-up during a pandemic* Speakers: Norma Tavakoli, PhD & Dorota Gruber, MD Wisconsin Public Health Laboratory: *Maintaining newborn screening quality during the COVID-19 pandemic* Speaker: Mei Baker, MD	*Can you hear me yet? Changing the way we listen to patients in the era of telehealth* Speakers: Christine Von Raesfeld & Idean Pourshams, MD Undiagnosed Disease Network: *COVID-19's Impact on the diagnostic odyssey and the work of the UDN* Speakers: Vandana Shashi, MD, MBBS & Troy Evans *Coping with an SMA diagnosis in a pandemic – the ups and downs of virtual care & when it just wasn't enough* Speakers: Kathryn Alexander & Kathryn Swoboda, MD
2:30-2:45PM	BREAK		
2:45-3:30PM	**Concluding Session: Our Collective Responsibility to Act** This session will feature a recap of key takeaways from the Case Study Panel Sessions & a discussion about how we move forward together. Moderator: Annie Kennedy – Chief of Policy & Advocacy, EveryLife Foundation for Rare Diseases *Peter Stein, MD – Director, Office of New Drugs, US Food and Drug Administration* *Christopher P. Austin, MD – Director, National Center for Advancing Translational Sciences, National Institutes of Health*		

Schedule and Speakers are subject to change.