



A New Era of Rare Disease Diagnostics and Therapeutics

Wednesday, February 24, 2021
Day 1

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| 10:00 am - 10:30 am EST
(3:00 pm - 3:30 pm GMT) | Opening Remarks and Introductions
James M. Wilson, MD, PhD
Rose H. Weiss Professor and Director, Orphan Disease Center,
Perelman School of Medicine, University of Pennsylvania |
| 10:30 am - 10:40 am EST
(3:30 pm - 3:40 pm GMT) | George Weiss
Founding Donor, Orphan Disease Center |
| 10:40 am - 10:45 am EST
(3:40 pm - 3:45 pm GMT) | Keynote Speaker Introduction
Jonathan A. Epstein, MD
Executive Vice Dean and Chief Scientific Officer,
Perelman School of Medicine, University of Pennsylvania |
| 10:45 am - 11:30 am EST
(3:45 pm - 4:30 pm GMT) | Keynote Speaker Rare Disease Research: New Methods,
New Partnerships, New Hope
Francis S. Collins, MD, PhD
Director, National Institutes of Health |
| 11:30 am - 12:30 pm EST
(4:30 pm - 5:30 pm GMT) | Panel Discussion Ethics |
| Panel Chair | Holly Fernandez Lynch, JD, MBE
Assistant Professor of Medical Ethics and Health Policy,
Perelman School of Medicine, University of Pennsylvania |
| Panelists | Alison Bateman-House, PhD, MPH, MA
Assistant Professor, Department of Population Health,
NYU Grossman School of Medicine |
| | Jeremy Snyder, PhD
Professor of Health Sciences, Simon Fraser University |
| | Timothy Yu, MD, PhD
Associate Professor of Pediatrics, Harvard Medical School |



Wednesday, February 24, 2021 Day 1

12:30 pm - 12:45 pm EST
(5:30 pm - 5:45 pm GMT)

BREAK

12:45 pm - 12:50 pm EST
(5:45 pm - 5:50 pm GMT)

Keynote Speaker Introduction

Jayne Gershkowitz

Chief Patient Advocate, Amicus Therapeutics

12:50 pm - 1:20 pm EST
(5:50 pm - 6:20 pm GMT)

Patient Voice

Lynn Barghout Jafar

Co-Founder, Loulou Foundation

1:20 pm - 2:20 pm EST
(6:20 pm - 7:20 pm GMT)

Panel Discussion | Global Access to Treatments

Panel Chair

Glen N. Gaulton, PhD

Vice Dean and Faculty Director, Center for Global Health
Professor of Pathology and Laboratory Medicine,
Perelman School of Medicine, University of Pennsylvania

Panelists

Luiz Henrique Mandetta, MD

Former Minister of Health Brazil

Khaled Otaifi, MD

Director of the Metabolic Disease Service at the
Egyptian Ministry of Health

Joan Benson, MD, MPH

Executive Director of Global Health Partnerships, Merck

2:20 pm - 2:30 pm EST
(7:20 pm - 7:30 pm GMT)

Closing Remarks

James M. Wilson, MD, PhD

Completion of Day 1



Thursday, February 25, 2021 Day 2

10:00 am - 10:10am EST
(3:00 pm - 3:10 pm GMT)

Opening Remarks

J. Larry Jameson, MD, PhD

Executive Vice President, University of Pennsylvania
for the Health System; Dean, Perelman School of Medicine

10:10 am - 10:15 am EST
(3:10 pm - 3:15 pm GMT)

Moderator and Featured Speaker Introduction

Majid Jafar

Co-Founder, Loulou Foundation

10:15 am - 11:00 am EST
(3:15 pm - 4:00 pm GMT)

Fireside Chat | Genomics in Rare Disease Diagnostics

Moderator

Tachi Yamada, MD

Former President of R&D, GSK and Takeda

Featured Speaker

David Cameron

Former UK Prime Minister

11:00 am - 12:00 pm EST
(4:00 pm - 5:00 pm GMT)

Panel Discussion | Patient Registries

Panel Chair

Pat Furlong

Founding President and CEO,
Parent Project Muscular Dystrophy (PPMD)

Panelists

Alicyn Campbell, MPH

Head of Digital Health, Oncology R&D, AstraZeneca

David Fajgenbaum, MD, MBA, MSc

Assistant Professor of Medicine, Perelman School of Medicine,
University of Pennsylvania

Founder, Castleman Disease Collaborative Network

Betsy Bogard, MS

Head of Program and Alliance Management
5 AM Ventures, 4:59 Initiative

12:00 pm - 12:10 pm EST
(5:00 pm - 5:10 pm GMT)

BREAK



Thursday, February 25, 2021

Day 2

12:10 pm - 12:15 pm EST
(5:10 pm - 5:15 pm GMT)

Keynote Speaker Introduction
Emma A. Meagher, MD
Vice Dean and Chief Clinical Research Officer
Perelman School of Medicine, University of Pennsylvania

12:15 pm - 1:00 pm EST
(5:15 pm - 6:00 pm GMT)

Keynote Speaker | Title TBD
Janet Woodcock, MD
Acting Commissioner, US Food & Drug Administration

1:00 pm - 2:00 pm EST
(6:00 pm - 7:00 pm GMT)

Panel Discussion | Industry Perspective and Incentives

Panel Chair

Annalisa Jenkins, MBBS, FRCP
Biotech Executive

Panelists

John Crowley, JD, MBA
CEO, Amicus

Emil Kakkis, MD, PhD
CEO, Ultragenyx

2:00 pm - 2:05 pm EST
(7:00 pm - 7:05 pm GMT)

Keynote Speaker Introduction
Daniel J. Rader, MD
Seymour Gray Professor of Molecular Medicine, Perelman School of
Medicine, University of Pennsylvania
Chair, Department of Genetics

2:05 pm - 2:50 pm EST
(7:05 pm - 7:50 pm GMT)

**Keynote Speaker | Homozygous Hypercholesterolemia:
An Orphan That Saved Millions**
Michael S. Brown, MD
Regental Professor, Molecular Genetics and Internal Medicine
UT Southwestern

2:50 pm - 3:00 pm EST
(7:50 pm - 8:00 pm GMT)

Closing Remarks
James M. Wilson, MD, PhD

Completion of Day 2