REAGAN-UDALL FOUNDATION FOR THE FDA

Qualifying Biomarkers to Support Rare Disease Regulatory Pathways
Case example: Heparan sulfate in neuronopathic lysosomal storage diseases

Hybrid Public Meeting Agenda
February 21, 2024; 10AM - 4PM (eastern time)
1333 New Hampshire Avenue NW; Rooftop Meeting Room
Washington, DC, 20036

Meeting Description: A Foundation-convened hybrid public meeting to explore primary disease activity biomarkers in rare genetic diseases using heparan sulfate in neuronopathic mucopolysaccharidoses as a case study for a biomarker to support accelerated approval.

10am **Welcome & Opening Remarks**
Susan C. Winckler, RPh, Esq., Reagan-Udall Foundation for the FDA

10:10am **Biomarkers in Rare Genetic Diseases**
- Regulatory perspective on criteria for qualifying a reliable biomarker in rare genetic diseases/ Consistency in Assay Development - Peter Marks, MD, PhD, Center for Biologics Evaluation and Research (CBER)

10:40am **Case Study: Understanding Neuronopathic Mucopolysaccharidoses (MPS)**
- Caregiver perspective: Impact of MPS on Patients - Mark Dant, Ryan Foundation
- Overview of MPS - Joseph Muenzer, MD, PhD, Univ. of North Carolina at Chapel Hill

11:20am **Case Study: Measuring Glycosaminoglycans (GAGs), including Heparan Sulfate (HS)**
- Optimization of GAG/HS quantification - Maria Fuller, PhD, University of Adelaide

11:50am **Q & A Session with morning Case Study presenters**

12:10pm **Lunch Break (Lunch provided)**

12:40pm **Case Study: Animal Model Translation to Human Application**
- Nidal Boulos, PhD, Regenxbio
- Patricia Dickson, MD, Washington University School of Medicine, St. Louis
- Matthew Ellinwood, DVM, PhD, National MPS Society

1:20pm **Case Study: Relationship Between Cerebrospinal HS Levels and Clinical Outcomes**
- Simon Jones, MBChB, University of Manchester
- Heather Lau, MD, MS, Ultragenyx
- Eric Zanelli, PhD, Allievex

2:05pm **Q & A Session with afternoon presenters**

2:35pm **10-minute break**

2:45pm **Panel Discussion: Challenges in Qualifying Biomarkers to Support Rare Disease Approvals**
Susan C. Winckler, moderator

- John Crowley, JD, MBA, Amicus Therapeutics, Inc.
  Biotechnology Innovation Organization (*incoming*)
- Cherie Fathy, MD, MPH, Center for Biologics Evaluation and Research, FDA
- Carole Ho, MD, Denali Therapeutics
- Gavin Imperato, MD, PhD, Center for Biologics Evaluation and Research, FDA
- Edward Neilan,MD, PhD, National Organization of Rare Diseases
- Cara O'Neill, MD, Cure Sanfilippo Foundation
- James Wilson, MD, PhD, University of Pennsylvania

3:55 - 4pm **Closing Remarks & Adjourn**

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