

APBD

MAY 11-12, 2022

SCIENTIFIC & COMMUNITY CONFERENCE

AGENDA AT A GLANCE

The Conference will feature:

Scientific and Medical Track

- Sessions oriented towards a scientific and medical audience.
- All are welcome to attend!

Community Track

- Sessions tailored especially for people with APBD and their loved ones.

Wednesday, May 11

ET	Scientific and Medical Track
10:00 AM	Welcome and Opening
10:15 AM	APBD – The Lived Experience
10:30 AM	Keynote Address
10:50 AM	Substrate and Polyglucosan Body Reduction Therapeutic Strategies
12:30 PM	Break
1:00 PM	Facilitated Meet and Greet
1:30 PM	Gene and Antisense Oligonucleotide Therapy Approaches for GBE1-deficient Diseases
2:45 PM	Break
3:00 PM	Updates from the Recent "Million Dollar Bike Ride" Pilot Grant Awardees
3:30 PM	Strategies for Advancing Therapies for Rare Neurological Disorders
4:00 PM	Closing Remarks

Thursday, May 12

ET	Scientific and Medical Track	ET	Community Track
10:00 AM	Welcome Back and Group Photo	1:00 PM	APBD – The Lived Experience
10:15 AM	APBD – The Lived Experience	1:20 PM	Patient-Focused Discussion of Clinical Practice Guidelines for APBD & GSD IV
10:30 AM	Meet and Greet	2:00 PM	Break
10:45 AM	Clinical Practice Guidelines for APBD & GSD IV	2:15 PM	APBD Therapeutic Development: Lay Overview of Day 1
11:30 AM	Prevalence, Natural History, Quality of Life and Registry	3:00 PM	Break
12:30 PM	Break	3:15 PM	Return to Scientific and Medical Track for "APBD – The Caregiver's Perspective" and Closing Remarks
1:00 PM	Biomarkers – Continuing the Conversation		
1:45 PM	Networking Session		
2:15 PM	Break		
2:30 PM	Addressing the Challenges of Diagnosing APBD		
3:15 PM	APBD – The Caregiver's Perspective		
3:45 PM	Closing Remarks		

AGENDA

Wednesday, May 11: Scientific and Medical Track

Time (Eastern Time)	Session
10:00 AM - 10:15 AM	Welcome and Opening Jeff Levenson, DDS, APBD Research Foundation
10:15 AM - 10:30 AM	APBD – The Lived Experience Sarah Williams, Person Living with APBD
10:30 AM - 10:50 AM	Keynote: A Fireside Chat with Dr. Ethan Perlstein Ethan Perlstein, PhD, Perlara PBC
10:50 AM - 12:30 PM	Substrate and Polyglucosan Body Reduction Therapeutic Strategies Moderator: Wyatt Yue, PhD, Newcastle University Stop polyglucosans to stop polyglucosan body disease Berge A. Minassian, MD, UT Southwestern Medical Center Silvia Nitschke, PhD, UT Southwestern Medical Center MZE001 GYS1 substrate reduction therapy for Pompe disease Julie Ullman, PhD, Maze Therapeutics Pharmacological approaches for treating APBD Or Kakhlon, PhD, Hadassah-Hebrew University Medical Center Alexander Lossos, MD, Hadassah-Hebrew University Medical Center Pre-clinical development of antibody-enzyme fusions for treatment of glycogen storage diseases Matthew S. Gentry, PhD, University of Kentucky Targeting glycogen clearance in the CNS with a clinical antibody-enzyme therapeutic Dustin Armstrong, PhD, Parasail LLC Q&A
12:30 PM - 1:00 PM	Break
1:00 PM - 1:30 PM	Facilitated Meet and Greet
1:30 PM - 2:45 PM	Gene and Antisense Oligonucleotide Therapy Approaches for GBE1-deficient Diseases Moderator: Nicolas J. Abreu, MD, NYU Langone Health Strategies for adeno-associated virus based gene therapy for APBD Mayank Verma, MD, PhD, UT Southwestern Medical Center

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Amelioration of GBE1 deficiency by intronic mutation-specific antisense oligonucleotide therapy

H. Orhan Akman, PhD, Columbia University Irving Medical Center

Michio Hirano, MD, Columbia University Irving Medical Center

AAV-mediated GBE1 gene replacement therapy for GSD IV/APBD

Rebecca L. Koch, PhD, RDN, LDN, Duke University Medical Center

Q&A

2:45 PM - 3:00 PM

Break

3:00 PM - 3:30 PM

Updates from the Recent "Million Dollar Bike Ride" Pilot Grant Awardees

Moderator: Edwin H. Kolodny, MD, NYU Grossman School of Medicine

Structure-guided discovery of glycogen synthase inhibitors

Wyatt Yue, PhD, Newcastle University

Conquer from Within – Treating APBD by viral delivery of cross-correction-enabled amylase

Felix Nitschke, PhD, UT Southwestern Medical Center

Q&A

3:30 PM - 4:00 PM

Strategies for Advancing Therapies for Rare Neurological Disorders

Jill A. Morris, PhD, National Institute of Neurological Disorders and Stroke

4:00 PM

Closing Remarks

Emil Weiss, MS, APBD Research Foundation

Thursday, May 12: Scientific and Medical Track

Time (Eastern Time)

Session

10:00 AM - 10:15 AM

Welcome Back and Group Photo

Catherine Fanning, APBD Research Foundation

Caliopy Glaros, Philanthropy Without Borders

10:15 AM - 10:30 AM

APBD – The Lived Experience

Paul, Person Living with APBD

10:30 AM - 10:45 AM

Meet and Greet

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10:45 AM - 11:30 AM	Clinical Practice Guidelines for APBD and GSD IV Introduction: Kim Burney, Association for Glycogen Storage Diseases Moderator: Rebecca L. Koch, PhD, RDN, LDN, Duke University Medical Center Deeksha Bali, PhD, FACMG, Duke University Medical Center Bridget Kiely, BS, Duke University Medical Center Andrew Landstrom, MD, PhD, Duke University Medical Center Jennifer L. Orthmann-Murphy, MD, PhD, University of Pennsylvania
11:30 AM - 12:30 PM	Study Updates: Prevalence, Natural History, Quality of Life and Registry Moderator: Olivia Montaña, CCDM, BS, PROS Foundation GBE1-related disease prevalence study Samantha Baxter, MS, CGC, Broad Institute GBE deficiency (GSD IV and APBD) natural history study Rebecca L. Koch, PhD, RDN, LDN, Duke University Medical Center Symptom burden and health-related quality of life survey for APBD Genevieve Wilson, BS, NYU Langone Health First patient-reported natural history study of APBD Francesco E. Michelassi, MD, PhD, Columbia University Jacy Sparks, BS, Columbia University Q&A
12:30 PM - 1:00 PM	Break
1:00 PM - 1:45 PM	Biomarkers – Continuing the Conversation ¹³C-MR as a diagnostic and non-invasive tool for quantification of brain glycogen in APBD Or Kakhlon, PhD, Hadassah-Hebrew University Medical Center The human and mouse metabolic signatures of Lafora disease Matthew S. Gentry, PhD, University of Kentucky
1:45 PM - 2:15 PM	Networking Session
2:15 PM - 2:30 PM	Break
2:30 PM - 3:15 PM	Addressing the Challenges of Diagnosing APBD Moderator: Jennifer L. Orthmann-Murphy, MD, PhD, University of Pennsylvania Aaron Baldwin, MS, LCGC, University of Pennsylvania Joseph R. Berger, MD, University of Pennsylvania Christyn A. Edmundson, MD, Swedish Medical Center
3:15 PM - 3:45 PM	APBD – The Caregiver’s Perspective Moderator: Linda Cedarbaum, APBD Research Foundation

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Sandra Gartner, Caregiver, Advocate, and Spouse
Jerry Leeds, Caregiver, Advocate, and Spouse
Bruce Rosky, Caregiver, Advocate, and Spouse

Q&A

3:45 PM - 4:00 PM

Closing Remarks

Harriet Saxe, APBD Research Foundation

Thursday, May 12: Community Track

Time (Eastern Time)

Session

1:00 PM - 1:20 PM

APBD – The Lived Experience

Moderator: Clyde Partin, MD, Emory University School of Medicine

Sarah Williams, Person Living with APBD

Paul, Person Living with APBD

1:20 PM - 2:00 PM

Patient-Focused Discussion of Clinical Practice Guidelines for APBD and GSD IV

Introduction: Kim Burney, Association for Glycogen Storage Diseases

Moderator: Rebecca L. Koch, PhD, RDN, LDN, Duke University Medical Center

Jennifer L. Orthmann-Murphy, MD, PhD, University of Pennsylvania

Raphael Schiffman, MD, MHSc, Texas Neurology

Ariana L. Smith, MD, University of Pennsylvania

Interactive discussion

2:00 PM - 2:15 PM

Break

2:15 PM - 3:00 PM

APBD Therapeutic Development: Lay Overview of Day 1

Moderator: Berge A. Minassian, MD, UT Southwestern Medical Center

Michio Hirano, MD, Columbia University Irving Medical Center

Alexander Lossos, MD, Hadassah-Hebrew University Medical Center

Anat Mordechai, MSc, Hadassah-Hebrew University Medical Center

Q&A

3:00 PM - 3:15 PM

Break

3:15 PM

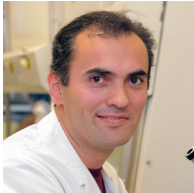
Return to Scientific and Medical Track for session:
APBD – The Caregiver’s Perspective

SPEAKER PROFILES



[Nicolas J. Abreu, MD](#)

Assistant Professor of Neurology
Director, Lysosomal Storage Disorders Program
Associate Director, Division of Neurogenetics
NYU Langone Health



[H. Orhan Akman, PhD](#)

Associate Professor of Neurology
Columbia University Irving Medical Center



[Dustin Armstrong, PhD](#)

President
Parasail LLC



[Aaron Baldwin, MS, LCGC](#)

Genetic Counselor
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[Deeksha Bali, PhD, FACMG](#)

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[Samantha Baxter, MS, CGC](#)

Associate Director
Genetic and Genomic Data Sharing
Translational Genomics Group
Broad Institute of MIT and Harvard

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[Joseph R. Berger, MD](#)

Associate Chief, Multiple Sclerosis Division
Professor of Neurology
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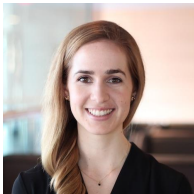


Kim Burney
President
Association for Glycogen Storage Disease



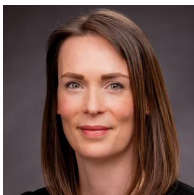
[Linda Cedarbaum](#)

Caregiver/Family Chat Group Facilitator
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Research Coordinator
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[Catherine Fanning](#)

Pharmaceutical Development Committee Chair
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Sandra Gartner
Caregiver, Advocate, and Spouse

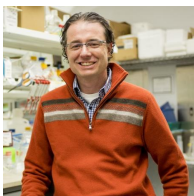
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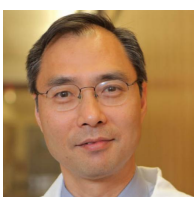
[Caliopy Glaros](#)

Facilitator & Consultant
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[Matthew S. Gentry, PhD](#)

Antonio S. Turco Endowed Professor
Department of Molecular and Cellular Biochemistry
University of Kentucky



[Michio Hirano, MD](#)

Chief of the Neuromuscular Division
Co-Director of the CUMC Muscular Dystrophy Association Clinic
Director of the H. Houston Merritt Center for Muscular Dystrophy and
Related Diseases
Columbia University



[Or Kakhlon, PhD](#)

Assistant Professor
Department of Neurology
Hadassah-Hebrew University Medical Center



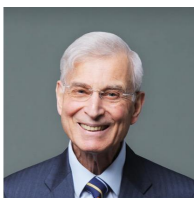
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Research Professor of Neurology
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Jerry Leeds

Caregiver, Advocate, and Spouse



[Jeff Levenson, DDS](#)

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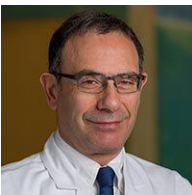
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Neurology Resident

Columbia University



[Berge A. Minassian, MD](#)

Division Chief

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[Olivia Montañó, CCDM, BS](#)

Advisor, Clinical Data Management, Rare Disease Pharma

Senior Director, Program Management, PROS Foundation

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Caregiver, Advocate, and Spouse



Harriet Saxe
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Professor in the Department of Internal Medicine, Texas Christian University
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Director of Pelvic Medicine and Reconstructive Surgery UPENN Chief of Urology Pennsylvania Hospital
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Resident Physician
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Emil Weiss, MS
Co-President
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Sarah Williams
Person Living with APBD



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[Wyatt Yue, PhD](#)
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CONFERENCE PLANNING COMMITTEE

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