

## Thursday October 14, 2021

8:00 pm **Faculty Dinner**

## Friday October 15, 2021

### Session 1- Introduction to Heme Biosynthesis and the Porphyrins

**Chair: Robert J. Desnick, PhD, MD**

- 1:00 pm **Welcome and Symposium Overview**  
*Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC*  
*Kristen Wheeden MBA, American Porphyria Foundation, Bethesda*  
*Sverre Sandberg, MD, PhD, University of Bergen, Bergen*
- 1:15 pm **Heme Biosynthesis: Overview and Latest Advances**  
*John D. Phillips, PhD, University of Utah, Salt Lake City*
- 1:45 pm **Overview of the Hepatic and Erythropoietic Porphyrins**  
*Bruce Wang, M.D., University of California, San Francisco*
- 2:15 pm **Porphyria Genetics, Genomics and Modifying Genes**  
*Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC*
- 2:50 pm **Porphyria Diagnostics: First- and Second-line Testing**  
*Karl Anderson, MD, University of Texas Medical Branch, Galveston*
- 3:25 pm **Break and Poster Viewing**

### Session 2 – Acute Hepatic Porphyrins (AHPs)

**Chairs: Karl Anderson, MD and Hetanshi Naik, PhD**

- 3:40 pm **Acute Hepatic Porphyrins (AHP): Pathogenesis and Manifestations**  
*Eliane Sardh MD, PhD, Karolinska Institute, Stockholm*
- 4:15 pm **AHPs – Neurologic Involvement**  
*Elena Pischik, MD, PhD, Consultative and Diagnostic Center with Polyclinics, St. Petersburg*
- 4:50 pm **AHPs: Current Management & Treatment**  
*Herbert Bonkovsky, MD, Wake Forest Baptist Medical Center, Winston-Salem*
- 5:25 pm **Acute Intermittent Porphyria (AIP) Patient Interview**  
*Sioban Keel, MD, University of Washington, Seattle*
- 5:45 pm **ALAD Deficient Porphyria**  
*Karl Anderson, MD, University of Texas Medical Branch, Galveston*
- 6:15 pm **Adjourn**
- 7:00 pm **Reception and Attended Poster Session**  
*Moderated by Bruce Wang, M.D., University of California, San Francisco*
- 8:00 pm **Dinner—Talk Tables for Mentorship and Networking**

## Saturday October 16, 2021

7:00 am **Breakfast—Talk Tables for Mentorship and Networking**

### Session 3 - Heme Biosynthesis: Recent Research

**Chair: John Phillips, PhD**

- 8:00 am **The Immunometabolite Itaconate Inhibits Heme Synthesis and Remodels Cellular Metabolism in Erythroid Precursors**  
*Harry Dailey, PhD, University of Georgia, Athens*
- 8:35 am **Structure-Based Drug Design to Aminolevulinic Synthase 2**  
*Wyatt Yue, PhD, University of New Castle, New Castle*

- 9:10 am **ALAD Is an Iron Sulfur Cluster Enzyme**  
Tracey A. Rouault, MD, NIH Division of Intramural Research Program, Bethesda
- 9:45 am **Heme Biosynthesis: Single Cell Hepatocyte Studies**  
Bruce Wang, MD, University of California at San Francisco, San Francisco
- 10:20 am **Break and Poster Viewing**  
**Chair: John Quigley, MD**
- 10:35 am **Exploring the Complex(ity) of Heme Biosynthesis & Distribution**  
Amy Medlock, PhD, University of Georgia, Athens
- 11:10 am **Heme Tolerance and Metabolism**  
Iqbal Hamza, PhD, University of Maryland, College Park
- 11:45 am **Investigation of Hepatic Heme Homeostasis Using an Inducible Hepatocyte-Specific Alas1 Knockout Mouse**  
Makiko Yasuda, MD, PhD, Icahn School of Medicine at Mount Sinai, NYC
- 12:20 am **Intracellular Heme Dynamics in Adipocytes**  
Enrique Saez, PhD, Scripps Research, La Jolla
- 12:55 pm **Lunch—Talk Tables for Mentorship and Networking**

### Session 4 - Porphyria Cutanea Tarda (PCT)

**Chairs: Herbert Bonkovsky, MD and Manish Thapar, MD**

- 1:45 pm **Porphyria Cutanea Tarda (PCT): Variants & Current Treatment**  
John Phillips, PhD, University of Utah, Salt Lake City
- 2:20 pm **Porphyria Cutanea Tarda (PCT) Patient Interview and Discussion**  
Herbert Bonkovsky, MD, Wake Forest Baptist Medical Center, Winston-Salem
- 2:40 pm **Break and Poster Viewing**

### Session 5 - Erythropoietic Porphyrias

**Chair: Manisha Balwani, MD, MS**

- 3:00 pm **Congenital Erythropoietic Porphyria (CEP): Manifestations & Current Treatment**  
Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC
- 3:35 pm **CEP Patient Interview**  
Angelika Erwin, MD, PhD, Cleveland Clinic, Cleveland
- 3:55 pm **Erythropoietic Protoporphyrin (EPP) & X-Linked Protoporphyrin (XLP): Variants & Current Treatment**  
Manisha Balwani, MD, MS, Icahn School of Medicine at Mount Sinai, NYC
- 4:30 pm **EPP/XLP Patient Interview**  
Cynthia Levy, MD, University of Miami, Miami
- 4:50 pm **EPP: A Physician-Patient's Perspective and Patient Survey**  
Amy Dickey, MD, Massachusetts General Hospital, Boston
- 5:10 pm **EPP: Late Breaking Research**  
Janneke Langendonk, MD, PhD, Erasmus Medical Center, Rotterdam
- 5:30 pm **Mechanism of Phototoxicity and the Erythropoietic Porphyrias**  
Bishr Omary, MD, PhD, Rutgers Biomedical and Health Sciences, Piscataway
- 5:50 pm **Adjourn**
- 7:00 pm **Reception and Attended Poster Session**  
Moderated by Brendan McGuire, MD, University of Alabama, Birmingham,
- 8:00 pm **Dinner**

Sunday October 17, 2021

## Session 6 - Emerging Treatments

Chair: Bruce Wang, MD

- 8:30 am **Innovative Therapies for Acute Intermittent Porphyrria**  
*Antonio Fontanellas, PhD, CIMA, University of Navarra, Pamplona*
- 9:10 am **Ciclopirox as a Pharmacological Chaperone for CEP**  
*Oscar Millet, PhD, CICbioGUNE & Atlas Molecular Pharmaceuticals, Bilbao & Madrid*
- 9:50 am **Emerging Therapies for Erythropoietic Protoporphyrria**  
*J.C Deybach, MD, PhD, Paris Diderot University, Paris*
- 10:30 am **Break and Poster Viewing**
- 10:45 am **Preclinical Studies on the Role of ABCG2 in Erythropoietic Protoporphyrria**  
*Xiaochao Ma, PhD, University of Pittsburgh, Pittsburgh*
- 11:25 am **Selected Poster Presentations**
- 12:00 pm **Lunch—Talk Tables for Networking and Mentoring**

## Session 7- Porphyrria Biochemical and Molecular Diagnostics

Chairs: Karl Anderson, MD and Robert J. Desnick, PhD, MD

- 1:00 pm **Porphyria Diagnostics: International Issues and Goals**  
*Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC*
- 1:15 pm **Improving Porphyria Diagnostics: Pre-analytical, Analytical and Post-analytical Aspects**  
*Sverre Sandberg, MD, PhD, University of Bergen, Bergen*
- 1:45 pm **Diagnosis of PCT and Erythropoietic Porphyria**  
*Karl E. Anderson, MD, University of Texas Medical Branch, Galveston*
- 2:00 pm **The International Porphyrias Gene Mutation Database**  
*Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC*
- 2:30 pm **Need for Harmonization in Diagnostic Testing and Reporting**  
*Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC*  
*Karl E. Anderson, MD, University of Texas Medical Branch, Galveston*
- 3:00 pm **Closing Remarks**  
*Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC*



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Sunday October 17, 2021

Session 8: Concurrent Patient Session

Chairs: Charles Parker, MD

- 10:00 am **Welcome and American Porphyria Foundation (APF) Update**  
*Kristen Wheeden, MBA American Porphyria Foundation, Bethesda*
- 10:05 am **APF Goes Global**  
*Desiree Lyon, American Porphyria Foundation, Global Director, Bethesda*
- Overview and What's New in the Porphyrias**  
*Diagnosis, management, and treatment*
- 10:15 am **Guidelines and Long-Term Follow-Up for all Porphyrias**  
*Manisha Balwani, MD, MS, Icahn School of Medicine at Mount Sinai, NYC*
- 10:30 am **AHP**  
*Sioban Keel, MD, University of Washington, Seattle*
- 10:45 am **PCT**  
*Karl Anderson, MD, University of Texas Medical Branch, Galveston*
- 11:00 am **EPP/XLP**  
*Cynthia Levy, MD, University of Miami, Miami*
- 11:15 am **CEP**  
*Angelika Erwin, MD, PhD, Cleveland Clinic, Cleveland*
- 11:30 am **Pain in the Porphyrias**  
Discussion about pain in the Acute (Hepatic) and Cutaneous (Erythropoietic) Porphyrias  
*Mohamed Kazamel, MD, University of Alabama, Birmingham*
- 12:00 pm **Research Is the Key to Your Cure!**  
Current research and how to get involved  
*Karli Hedstrom, MPH, Icahn School of Medicine at Mount Sinai, NYC*
- 12:15 pm **Emerging Treatments in the Porphyrias**  
Discussion about new and potential treatments in the porphyrias  
*Bruce Wang, M.D., University of California, San Francisco*
- 12:45 pm **Lunch**
- Chairs: Sean Rudnick, MD
- 1:15 pm **Quality of Life and Psychosocial Issues**  
Issues and impact beyond the physical symptoms  
*Hetanshi Naik, PhD, Icahn School of Medicine at Mount Sinai, NYC*
- 1:45 pm **Cannabis Use and Alternative Medicines**  
Results of a patient survey  
*Hetanshi Naik, PhD, Icahn School of Medicine at Mount Sinai, NYC*
- 2:00 pm **Ask the Porphyria Experts Panel**  
*Mohamed Kazamel, MD, Sioban Keel, MD, and Brendan McGuire, MD*
- 2:45 pm **Get Involved – what you can do as a rare disease patient**  
Highlights of ways to be involved in education, awareness, advocacy and support programs  
*Kristen Wheeden, MBA American Porphyria Foundation, Bethesda*
- 3:00 pm **Adjourn**