

PRELIMINARY PROGRAM

Heme Biosynthesis & the Porphyrins: 2021 Program

Thursday October 14, 2021

8:00 pm Faculty Dinner

Friday October 15, 2021

Session 1 - Introduction to Heme Biosynthesis and the Porphyrins

Chairs: Robert J. Desnick, PhD, MD and Sverre Sandberg, MD, PhD

1:00 pm **Welcome and Symposium Overview**
Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC

1:15 pm **Heme Biosynthesis: Overview, Latest Advances and COVID-19**
John D. Phillips, PhD, University of Utah, Salt Lake City

1:45 pm **Overview of the Hepatic and Erythropoietic Porphyrins**
Janneke Langendonk, MD, PhD, Erasmus Medical Center, Rotterdam, Netherlands

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, Manifestations, & Variants**
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**

7:30 pm **Reception and Attended Poster Session**
Moderated by Simon Beaven, M.D, University of Los Angeles, Los Angeles

8:30 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

Chairs: John Phillips, PhD and Antonio Fontanellas, PhD

8:00 am **Regulation of Heme Synthesis During Erythropoiesis by the Immunometabolite Itaconate**
Harry Dailey, PhD, University of Georgia, Athens

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- 8:35 am **Structure-Based Drug Design to Aminolevulinate Synthase 2**
Wyatt Yue, PhD, University of Oxford, Oxford
- 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**
Chairs: Michael Badminton, MB, ChB, PhD and 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

Chairs: Manisha Balwani, MD, MS and Jean-Charles Deybach, MD, PhD

- 3:00 pm **Congenital Erythropoietic Porphyria (CEP): Manifestations & Current Treatment**
Angelika Erwin, MD, PhD, Cleveland Clinic, Cleveland
- 3:35 pm **CEP Patient Interview**
Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC
- 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, Netherlands
- 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,

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8:00 pm	<i>Dinner</i>
Sunday October 17, 2021	
7:30 am	Breakfast—Talk Tables for Mentorship and Networking
Session 6 - Emerging Treatments	
Chairs: Bruce Wang, MD and Janneka Langendonk, MD	
8:30 am	Enzyme and Gene Therapy for Acute Intermittent Porphyria <i>Antonio Fontanellas, PhD, Center for Applied Medical Research (CIMA), University of Navarra, Pamplona</i>
9:10 am	Ciclopirox for CEP <i>Oscar Millet, PhD, CICbioGUNE and Atlas Molecular Pharmaceuticals, Bilbao and Madrid</i>
9:50 am	Emerging Treatments for Erythropoietic Protoporphyrria <i>J.C Deybach, MD, PhD, Paris Diderot University, Paris</i>
10:30 am	Break and Poster Viewing
10:45 am	Preclinical Studies on the Role of ABCG2 in Erythropoietic Protoporphyrria <i>Xiaochao Ma, PhD, University of Pittsburgh, Pittsburgh</i>
11:25 am	Late Breaking Research
12:00 pm	Adjourn
12:00 pm	Lunch—Talk Tables for Networking and Mentoring
Session 7- Porphyria Biochemical and Molecular Diagnostics	
Chairs: Karl Anderson, MD and Robert J. Desnick, PhD, MD	
1:00 pm	Porphyria Diagnostics: International Issues and Goals <i>Michael Badminton, MB, ChB, PhD, Cardiff University, Cardiff</i>
1:15 pm	Improving Porphyria Diagnostics: Pre-analytical, Analytical and Post-analytical Aspects <i>Sverre Sandberg, MD, PhD, University of Bergen, Bergen, Norway</i>
1:45 pm	Diagnosis of PCT and Erythropoietic Porphyria <i>Karl E. Anderson, MD, University of Texas Medical Branch, Galveston</i>
2:00 pm	Diagnostic Guidelines and International Molecular Database <i>Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC</i>
2:30 pm	Need for Harmonization in Diagnostic Testing and Reporting <i>Moderator: Discussion: Diagnostic Lab Participation</i>
3:00 pm	Closing Remarks <i>Robert J. Desnick, PhD, MD, Icahn School of Medicine at Mount Sinai, NYC</i>

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Sunday October 17, 2021 Session 8: Concurrent Patient Session

Chairs: Cynthia Levy, MD and Charles Parker, MD

- 10:00 am **Welcome and American Porphyria Foundation (APF) Update**
Kristen Wheeden, American Porphyria Foundation, Executive Director
- 10:05 am **APF Goes Global**
Desiree Lyon, American Porphyria Foundation, Global Director
- 10:15 am **Overview and What's New in the Porphyrias**
Diagnosis, management, and treatment
- Guidelines and Long-Term Follow-Up for all Porphyrias**
Manisha Balwani, MD, MS, Icahn School of Medicine at Mount Sinai, NYC
- AHP**
Michael Badminton, MB, ChB, PhD, Cardiff University, Cardiff
- PCT**
Karl Anderson, MD, Karl Anderson, University of Texas Medical Branch, Galveston
- EPP/XLP**
Manisha Balwani, MD, MS, Icahn School of Medicine at Mount Sinai, NYC
- CEP**
Angelika Erwin, MD, PhD, Cleveland Clinic, Cleveland
- 11:30 am **Emerging Treatments in the Porphyrias**
Discussion about new and potential treatments in the porphyrias
Jean-Charles Deybach, MD, PhD, Paris Diderot University, Paris, France
- 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 **Pain in the Porphyrias**
Explain and discuss pain in the Acute (Hepatic) and Cutaneous (Erythropoietic) Porphyrias
Elena Pischik, MD, PhD, Consultative and Diagnostic Center with Polyclinics, St. Petersburg
Mohamed Kazamel, MD, University of Alabama, Birmingham
- 12:45 pm **Lunch**
- Chairs: Sean Rudnick, MD and Gregory Vercellotti, 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**
Bruce Wang, Sioban Keel, Cynthia Levy, and Brendan McGuire
- 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, Executive Director, APF
- 3:00 pm **Adjourn**