

HEME BIOSYNTHESIS AND THE PORPHYRIAS: RECENT ADVANCES

Meeting Highlights

Expert international faculty

Latest clinical and basic research advances

Talk tables for trainees during breakfast and lunch

Separate clinical session for patients

Friday, January 12, 2018

Latest Advances in the Diagnosis and Management of the Porphyrrias

8:00 AM Welcome & Course Goals— R.J. Desnick, PhD, MD

Co-Chairs: J.-C. Deybach MD, PhD & E. Sardh, MD, PhD

8:15 AM Heme Biosynthesis & Degradation for Clinicians— J.D. Phillips, PhD

9:00 AM Porphyria Genetics: Inheritance and Molecular Diagnosis— R.J. Desnick, PhD, MD

10:00 AM The Acute Hepatic Porphyrrias (AHPs): Metabolic Defects, Pathophysiology & Clinical Manifestations— H.L. Bonkovsky, MD

10:45 AM AHPs: Diagnosis and Current Management— K.E. Anderson, MD

11:30 AM AIP Patient Interview— D.M. Bissell, MD

12:00 PM Expert Panel: Critical Issues to Resolve & Future Directions

Moderator: J.-C. Deybach, MD, PhD

Panel: H.L. Bonkovsky, MD, K.E. Anderson, MD, D.M. Bissell, MD, E. Sardh, MD, PhD

Clinical Session 2- Porphyria Cutanea Tarda

Co-Chairs: P. Wilson, MD & H. Puy, MD, PhD

1:30 PM Porphyria Cutanea Tarda: Metabolic Defect, Predisposing Factors, Pathophysiology & Treatment— A.K. Singal, MD

2:15 PM PCT Patient Interview— K.E. Anderson, MD

2:45 PM Expert Panel: Critical Issues to Resolve & Future Directions

Moderator: P. Wilson, MD

Panel: A.K. Singal, MD, K.E. Anderson, MD, J.D. Phillips, PhD, J. To-Figueras, PhD

Clinical Session 3- The Erythropoietic Porphyrrias

Co-Chairs: E. Minder, MD & M. Poh-Fitzpatrick, MD

3:30 PM Congenital Erythropoietic Porphyria (CEP): Metabolic Defect, Pathophysiology & Treatments— R.J. Desnick, PhD, MD

4:15 PM CEP Patient Interview— R.J. Desnick, PhD, MD

4:45 PM Erythropoietic Protoporphyrria (EPP) & X-Linked Protoporphyrria (XLP): Metabolic Defects, Pathophysiology & Treatment— M. Balwani, MD, MS

5:30 PM EPP/XLP Patient Interview— M. Poh-Fitzpatrick, MD

6:00 PM Expert Panel: Critical Issues to Resolve & Future Directions

Moderator: M. Poh-Fitzpatrick, MD

Panel: M. Balwani, MD, MS, J.R. Bloomer, MD, P. Wilson, MD, E. Minder, MD

7:00 PM Reception & Posters

Saturday, January 13, 2018

Research Session 1- Chemistry, Biology, and Genetics of Human Heme Biosynthesis

Co-Chairs: R.J. Desnick, PhD, MD & B. Paw, MD, PhD

8:00 AM Regulation and Tissue-Specific Expression of ALAS— H. Puy, MD, PhD

8:45 AM Structure and Assembly of ALAS— T. Baker, PhD

9:20 AM Human ALAS1 and ALAS2 Catalysis— G. Ferreira, PhD

10:30 AM Mitochondrial Heme Biosynthesis Complex— H. Dailey, PhD

11:00 AM Selected Abstract Talks 1 & 2

11:30 AM Expert Panel: Critical Issues to Resolve and Future Directions— Co-Chairs to Moderate

Research Session 2- Porphyrins and Porphyrin Precursors in Health and Disease

Co-Chairs: H.L. Bonkovsky, MD & G. Ferreira, PhD

1:00 PM ALA or PBG: Which is the Culprit?— B. Wang, MD

1:35 PM Murine Models of Porphyria— M. Yasuda, MD, PhD

2:10 PM Mitochondrial Iron and Heme Metabolism— B. Paw, MD, PhD

2:45 PM Transport of Porphyrins— I. Hamza, PhD

3:45 PM Elevated Precursors and/or Porphyrins and No Heme Biosynthetic Gene Mutation— M. Badminton, MChB, PhD

4:20 PM New Methods of Heme Quantitation— A.R. Reddi, PhD

5:00 PM Trainee Abstract Talks 3 & 4

5:30 PM Expert Panel: Critical Issues to Resolve and Future Directions— Co-Chairs to Moderate

SAVE THE DATE

January 12-14, 2018

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Orlando, FL

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Sunday, January 14, 2018

Research Session 3- Underlying Mechanisms for Clinical Complications of the Porphyrrias

Co-Chairs: D.M. Bissell, MD & E. Minder, MD

8:00 AM Neurologic Mechanisms Responsible for Pain in AHPs— H.L. Bonkovsky, MD

8:35 AM Complications of the AHPs: Renal Disease, HCC, etc.— J.-C. Deybach, MD, PhD

9:10 AM Psychosocial Aspects of the AHPs— H. Naik, MS

10:00 AM PCT: Basis of Complications Including Hepatitis, Iron Overload, etc.— J. To-Figueras, PhD

10:35 AM Pathogenesis of Phototoxicity & Pain in the Erythropoietic Porphyrrias— M. Poh-Fitzpatrick, MD

11:10 AM Trainee Abstract Talks 5 & 6

11:40 AM Expert Panel: Critical Issues to Resolve and Future Directions— Co-Chairs to Moderate

Research Session 4- Emerging Therapies for Acute & Erythropoietic Porphyrrias

Co-Chairs: J.D. Phillips, PhD & P. Wilson, MD

1:00 PM RNAi Therapy for Acute Hepatic Porphyrrias— E. Sardh, MD, PhD

1:35 PM Gene and Molecular Therapies for AIP— A. Fontanellas, MD

2:10 PM Molecular Approaches to Treat Erythropoietic Protoporphyrrias— J.-C. Deybach, MD, PhD

3:00 PM Porphyria Genetics: Penetrance and the Search for Modifying Genes— R.J. Desnick, PhD, MD

3:35 PM Trainee Abstract Talks 7 & 8

4:10 PM Expert Panel: Critical Issues to Resolve and Future Directions— Co-Chairs to Moderate

4:30 PM Summation and Closing Remarks— P. Wilson, MD

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SPECIAL PATIENT MEETING

Discussing the Erythropoietic
& Acute Hepatic Porphyrrias

Including Interactive Q & A Sessions with Experts

Saturday, January 13, 2018, 10:00 AM - 4:45 PM