Venue: Hyatt Regency Orlando

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Heme Biosynthesis and the Porphyrias: Recent Advances

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Friday, January 12, 2018
Latest Advances in the Diagnosis and Management of the Porphyrias

9:00 am Welcome & Course Goals– R.J. Desnick, PhD, MD
Co-Chairs: J.-C. Deybach, MD, PhD & E. Minder, 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 Porphyrias (AHPs): Metabolic Defects, Pathophysiology & Clinical Manifestations– R.L. Bissell, MD
10:45 am AHPs: Diagnosis and Current Management– K.E. Anderson, MD
11:30 am AIP Patient Interview– D.M. Bisello, MD

12:00 pm Expert Panel: Critical Issues to Resolve & Future Directions
Moderator: J.-C. Deybach, MD, PhD
Panel: J.-C. Deybach, MD, PhD, K.E. Anderson, MD, D.M. Bisello, MD, E. Minder, 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– R.J. Desnick, PhD, 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: J.-C. Deybach, MD, PhD, K.E. Anderson, MD, J.D. Phillips, PhD, J.-C. Deybach, MD, PhD

Clinical Session 3: The Erythropoietic Porphyrias

Co-Chairs: E. Minder, MD & M.P. Poh-Fitzpatrick, MD
3:30 pm Congenital Erythropoietic Porphyria (CEP): Metabolic Defect, Pathophysiology & Treatment– R.J. Desnick, PhD, MD
4:15 pm CEP Patient Interview– R.J. Desnick, PhD, MD
4:45 pm Erythropoietic Protoporphyrina (EPP) & X-Linked Porphyrinemia (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. Brummer, 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, MD, PhD & 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
9:30 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: Porphyria and Porphyrin Precursors in Health and Disease

Co-Chairs: H.L. Rockovsky, MD & S. Ferrone, PhD
1:00 pm ALA or PBG: Which is the Culprit?– H. Wang, MD
1:30 pm Murine Models of Porphyria– M. Yacoub, MD, PhD
2:10 pm Mitochondrial Iron and Heme Metabolism– B. Paw, MD, PhD
2:45 pm Transport of Porphyrins– J. Hanca, PhD
3:45 pm Elevated Precursors and/or Porphyrins and No Heme Biosynthetic Gene Mutation– M. Bastien, MD, PhD
4:20 pm New Methods of Heme Quantitation– A.R. Redd, 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

Sunday, January 14, 2018
Research Session 2: Underlying Mechanisms for Clinical Complications of the Porphyrias

Co-Chairs: D.M. Bissell, MD & E. Minder, MD
8:00 am Neurologic Mechanisms Responsible for Pain in AHPs– H.L. Birksanky, MD
8:35 am Complications of the AHPs: Renal Disease, HCC, etc.– J.-C. Deybach, MD, PhD
9:15 am Psychosocial Aspects of the AHPs– H. Kirk, MD
10:00 am PCT: Basis of Complications Including Hepatitis, Iron Overload, etc.– J.-C. Deybach, MD, PhD
10:35 am Pathogenesis of Phototoxicity & Pain in the Erythropoietic Porphyrias– 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 Porphyrias

Co-Chairs: J.D. Phillips, PhD & P. Wilson, MD
1:00 pm RNAi Therapy for Acute Hepatic Porphyrias– E. Sardh, MD, PhD
1:35 pm Gene and Molecular Therapies for AIP– A. Fontenelles
2:10 pm Molecular Approaches to Treat Erythropoietic Porphyrias– J.-C. Deybach, MD, PhD
3:00 pm Porphyria Genetics: Penetration and the Search for Modifying Genes– R.L. Bissell, MD, PhD
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 Porphyrias
Including Interactive Q & A Sessions with Experts
Sunday, January 13, 2018, 10:00 am - 4:45 pm