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Friday, January 12, 2018
Latest Advances in the Diagnosis and Management of the Porphyrias
8:05 am Welcome & Course Goals—R.J. Desnick, PhD, MD
8:15 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—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
8:00 am Co-Chairs: P. Wilson, MD & H. Puy, MD, PhD
8:30 am Congenital Erythropoietic Porphyria (CEP): Metabolic Defect, Predisposing Factors, Pathophysiology & Treatments—R.J. Desnick, PhD, MD
8:45 am CEP Patient Interview—R.J. Desnick, PhD, MD
9:15 am Expert Panel: Critical Issues to Resolve & Future Directions—Moderator: P. Wilson, MD
9:30 am Panel: A.K. Singal, MD, K.E. Anderson, MD, J.D. Phillips, PhD, J. To-Figueras, PhD
Saturday, January 13, 2018
Research Session 1- Chemistry, Biology, and Genetics of Human Heme Biosynthesis
8:00 am 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. Bailer, PhD
9:30 am Human ALAS1 and ALAS2 Catalysis—G. Ferrero, PhD
10:30 am Mitochondrial Heme Biosynthesis Complex—K. Dalkey, 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 Porphyria Precursors in Health and Disease
8:00 am Co-Chairs: H.L. Bankovsky, MD & G. Ferrero, PhD
8:00 am ALA or PBG: Which is the Culprit?—B. Wang, MD
8:15 am Murine Models of Porphyria—M. Yacoud, MD, PhD
8:30 am Mitochondrial Iron and Heme Metabolism—B. Pan, MD, PhD
8:45 am Transport of Porphyrins—J. Hanco, PhD
9:00 am Elevated Precursors and/or Porphyrins and No Heme Biosynthetic Gene Mutation—M. Batori, MDCM, PhD
9:15 am New Methods of Heme Quantitation—A.R. Reddi, PhD
9:30 am Trainee Abstract Talks 3 & 4
10:30 am Expert Panel: Critical Issues to Resolve and Future Directions—Co-Chairs to Moderate
Sunday, January 14, 2018
Research Session 3- Underlying Mechanisms for Clinical Complications of the Porphyrias
8:00 am Co-Chairs: D.M. Bissell, MD & E. Minder, MD
8:30 am Neurologic Mechanisms Responsible for Pain in AHPs—H.L. Bonkovsky, MD
9:15 am Complications of the AHPs: Renal Disease, HCC, etc.—J.-C. Deybach, MD, PhD
10:00 am Psychosocial Aspects of the AHPs—J. To-Figueras, PhD
10:30 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
8:00 am Co-Chairs: J.D. Phillips, PhD & P. Wilson, MD
8:30 am RNAi Therapy for Acute Hepatic Porphyrias—E. Sardh, MD, PhD
9:15 am Gene and Molecular Approaches for AIP—A. Fontenelle, MD
9:30 am Molecular Approaches to Treat Erythropoietic Porphyrias—J.-C. Deybach, MD, PhD
10:00 am 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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