AGENDA

Monday, 29 November 2010

8:00 AM	**Registration/Breakfast**
8:30 AM	**What are we here for?** *Sharon Terry, PXE International*
8:45 AM	**Keynote** Therapeutic Development for Rare Diseases: The Plan of Action *Christopher Austin, NCGC, NHGRI, NIH*
9:15 AM	**Basic Science of ABC Transporters** *Chair: Irwin Arias, NICHD, NIH* ABC family - *Michael Dean, NCI, NIH* The genetics of ABCC6 and creation of NLM rare disease database - *Timothy Hefferon, NHGRI, NIH* Mechanisms of drug conjugate transport - *Irwin Arias, NICHD, NIH* The ABCC6 protein: modeling and structure-function studies - *András Váradi, Hungarian Academy of Science, Budapest, Hungary* Transcriptional regulation of the gene - *Tamás Arányi, Hungarian Academy of Science, Budapest, Hungary*
11:00 AM	**Break**
11:15 AM	**PXE Basic Science** *Chair: Jouni Uitto, Jefferson Medical College* Genotype/phenotype correlations - *Anne DePaepe, University of Ghent, Ghent, Belgium* Pathology of elastic structures: primary or secondary effect? - *Jeffrey Davidson, Vanderbilt University, Nashville, TN* Therapeutic approaches to inhibit elastic fiber calcification in PXE: New perspectives derived from treatments of PXE fibroblasts - *Daniela Quaglino, Unviersity of Modena and Reggio, Modena, Italy* TBD - *Astrid Plomp, Netherlands Institute for Neuroscience, University of Amsterdam, The Netherlands (invited)*
1:00 PM	**Lunch**
2:00 PM	**Human Disease Associated with ABC Transporters** *Chair: Michael Gottesman, NCI, NIH* ABC transporters and human disease: an overview - *Michael Gottesman, NCI, NIH* CFTR - *Patrick Thibedeau, University of Pennsylvania, PA* Retinitis Pigmentosa, Stargardt's disease and age-related macular degeneration - *Rando Allikmets, Columbia University, NY*

3:30 PM	**Break**
3:45 PM	**Animal Models** *Chair: Olivier LeSaux, University of Hawaii* The PXE mouse - *Qiujie Jiang, Jefferson Medical College, Philadelphia, PA* PXE zebra fish model - *Qiaoli Li, Jefferson Medical College, Philadelphia, PA* In vivo expression and maturation of the human ABCC6 and its PXE-mutants in mice liver - *Olivier LeSaux, University of Hawaii*
5:15 PM	**Summary of the Day** *Jouni Uitto*
5:45 PM	**Poster Session and Reception**
6:45 PM	**Dinner** **Presentation of PXE International Award to Jouni Uitto, MD, PhD**

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8:00 AM	**Breakfast**
8:30 AM	**Hypotheses** *Chair: TBD* Vitamin K-dependent g-glutamyl carboxylation reactions - *Kathleen Berkner, Case Western Reserve University, Cleveland, OH* Dietary magnesium as a modifier of mineralization - *Jennifer LaRusso, Jefferson Medical College, Philadelphia, PA (invited)* Role of serum fetuin-A in PXE - *Doris Hendig, Herz- und Diabeteszentrum Nordrhein-Westfalen, Institut Für Laboratoriums- und Transfusionsmedizin, Germany* Modifier genes involved in PXE - *Olivier Vannaker, Ghent University Hospital, Ghent, Belgium*
10:30 AM	**Break**
10:45 AM	**Clinical Studies** *Chair: Lionel Bercovitch, Brown University, Providence, RI* Treatments for the eye - *Emily Chew, NEI, NIH* Renagel study - *Mark Lebwohl, Mt. Sinai School of Medicine* Pediatric study - *Ludovic Martin, Angers University Hospital, Angers, France* Ethical issues in predictive genetic testing for children - *Lionel Bercovitch, Brown University, Providence, RI*

12:15 PM	**Lunch**
1:15 PM	**Consensus: Diagnostic Criteria Proposal** *Chair: Lionel Bercovitch, Brown University, Providence, RI* *Anne DePaepe* *Mark Lebwohl* *Martin Ludovic* *Astrid Plomp* *Jouni Uitto*
3:00 PM	**Break**
3:15 PM	**Panel Discussion: Towards Treatment** *Moderator: Jouni Uitto, Jefferson Medical College* *Irwin Arias* *Chris Austin* *Lionel Bercovitch* *Patrick Terry*
5:00 PM	**Summary Charge** *Patrick Terry, PXE International*