



Sunday September 3rd & Monday September 4th
preceding the 13th ICIEM in Rio de Janeiro, Brazil

Sunday September 3, 2017

Session 1: Welcome and Keynote Speaker

- 15:30-16:30 Registration
- 16:30-16:40 **Welcome and Overview to INFORM 2017 & Keynote Speaker Introduction**
Jerry Vockley, Pittsburgh PA, USA
- 16:45-17:45 **Keynote, The Impact of NBS of FAODs**
Nicola Longo, Salt Lake City, Utah, USA
- 17:45-18:15 Discussion
- 19:00-21:30 Networking Reception and Opening Dinner

Monday Morning September 4, 2017

Session 2: Basic Science

- 7:30-8:30 Registration / Breakfast
- 8:30-9:00 **Myotubes from CPT2 Deficient Patients as a Disease Model**
Fatima Djouadi, Paris, France
- 9:00-9:30 **Free Fatty Acid Receptors and the Gut Microbiome: Questions in relation to FAD Defects**
Niels Gregersen, Aarhus Denmark
- 9:30-9:45 **Endoplasmic reticulum-mitochondria crosstalk and redox homeostasis disruption in very long-chain acyl-CoA dehydrogenase deficient fibroblasts**
Bianca Seminotti, Pittsburgh, PA, USA
- 9:45-10:00 **Development and characterization of patient-specific iPSC-derived retinal pigmentary epithelia (RPE)-like cells as a model of LCHAD-associated retinopathy**
Tiffany DeVine, Portland, Oregon, USA
- 10:00-10:30 Break
- 10:30-11:00 **NAD⁺ Therapy Improves Cardiac Function and Bioenergetics in a Mouse Model of Heart Failure**
Matt Hirschey, Duke University, USA
- 11:00-11:30 **Lessons Learned from Animal Models of VLCAD Deficiency**
Ute Spiekeroeter, Freiburg, Germany
- 11:30-11:45 **VLCAD deficiency related chronic inflammation pattern is suggestive of systemic mediators**
Megan Beck, Memphis, TN, USA
- 11:45-12:00 **Experimental evidence that fatty acids accumulating in VLCAD deficiency disrupt mitochondrial respiration in heart, liver and brain of young rats**
Cristiane Cecatto, Porto Alegre, RS, Brazil
- 12:00-12:30 Panel Discussion
- 12:30-14:00 Lunch

Monday Afternoon September 4, 2017

Session 3: Clinical session

- 14:00-14:30 **Nutritional Ketosis**
Kieran Clarke, Oxford, UK
- 14:30-15:00 **Ketone Body Treatment in FAODs**
Gepke Visser, Utrecht/Amsterdam, Netherlands
- 15:00-15:15 **3-hydroxybutyrate (3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency: a systematic literature review and international retrospective cohort study**
Willemijn J. van Rijt, Groningen, the Netherlands
- 15:15-15:30 **Results from a 78-week Single-arm, Open-label Phase 2 Study to Evaluate UX007 in Pediatric and Adult Patients with Moderate to Severe Long-Chain Fatty Acid Oxidation Disorders (LC-FAOD)**
Jerry Vockley, Pittsburgh, PA, USA
- 15:30-15:45 ... Break
- 15:45-16:15 **An overview of FAODs and unmet needs in Latin America**
Ida Schwartz, Porto Alegre, Brazil
- 16:15-16:30 **Health-related Quality of Life in Patients with long-chain Fatty Acid Oxidation Disorders**
Suzan JG Knottnerus, Amsterdam, The Netherlands
- 16:30-16:45 ... **Intravenous Sources of Medium Chain Triglycerides for Critically Ill Patients with Fatty Acid Oxidation Disorders**
Heather Bausell, Chicago, IL, USA
- 16:45-17:15 Panel Discussion
- 17:15-17:30 **Summary / Closing remarks**
- 17:30-18:45 ... **Cocktail Reception & Poster Session with Oral Presentations for Jr. Investigators interested in presenting**
- 19:00 INFORM Advisory Committee Meeting

INFORM SYMPOSIUM at ICIEM / September 7, 2017

FATTY ACID OXIDATION DISORDERS

Chair: Mike Bennett (Philadelphia, PA, USA) / Co-Chair: Eric Goetzman (Pittsburgh, PA, USA)

Anti-oxidant therapy as an adjunct for treatment of long chain fatty acid oxidation disorders *Guilhan Leipnitz, Brazil*

Novel mechanisms of pathogenesis in mitochondrial trifunctional protein deficiency: implications for clinical outcome and treatment : *Areeg El-Gharbawy, USA*

Rhabdomyolysis and inflammation in metabolic muscle disease: *Yamina Hamel, France*