

Day 2 Program – Tuesday, Sept 19th 2023			Duration (mins)		
Session 4 - Novel Diagnostic Tools & Center Initiatives (Chair: Barbara Yu)					
Time	Speaker	Description	Presentation	Q&A	Total
9:00	Kimitoshi Nakamura <i>Kumamoto University, Japan</i>	Establishing early diagnosis and long-term follow-up for CD with management guidelines	30	10	40
9:40	Johannes Häberle <i>University Children's Hospital Zürich, University of Zürich, Switzerland</i>	Quantification of the urea cycle flux and biomarker detection as novel diagnostic tools for citrin deficiency	20	10	30
10:10	Nicola Longo <i>University of Utah, USA</i>	Identification of Citrin Deficiency by Newborn Screening in Utah	15	5	20
10:30	Morning Break		30		30
Session 5 - Emerging Clinical Evidence on Citrin Deficiency (Chair: Robin Lachmann)					
11:00	Ituro Inoue <i>National Institute of Genetics, Japan</i>	Lessons from liver tissues of CTLN2 patients	20	10	30
11:30	Masahide Yazaki <i>Shinshu University, Japan</i>	Therapeutic Experiences including LOLA for CTLN2 Patients	20	10	30
12:00	Kimihiko Oishi <i>The Jikei University School of Medicine, Japan</i>	Clinical Biochemical Characteristics of Citrin Deficiency in Japan	20	10	30
12:30	Shirou Matsumoto <i>Kumamoto University, Japan</i>	Clinical trial in single center: Treatment with L-carnitine, MCT oil and Amino acids for Citrin Deficiency	15	5	20
12:50		Announcement	10		10
13:00	Lunch		60		60
14:00	Marc Hellerstein <i>University of California at Berkeley, USA</i>	Metabolic Pathogenesis of CD: Applying New Stable Isotope-Mass Spectrometric Flux Measurements Combined with Innovative Tools to Manipulate Cytosolic Redox	20	10	30
Session 6 – Novel Therapies II – Development of Therapeutic Interventions for Citrin Deficiency (Chair: Edmund Kunji)					
14:30	Paolo Martini <i>Moderna, Inc., USA</i>	Messenger RNA therapy is a platform therapeutic for the treatment of Rare Genetic Disorders	20	10	30
15:00	Joseph Baur <i>University of Pennsylvania, USA</i>	Modulating NAD+ Availability in Mice with Citrin Deficiency	20	10	30
15:30	Short Break		10		10
15:40	Yukio Nagasaki <i>University of Tsukuba, Japan</i>	Innovations in Amino Acid donors: Design and implementation using amphiphilic block copolymer micelles	20	10	30
16:10	Marc Prentki <i>Montreal Diabetes Research Center, CRCHUM, Canada</i>	Targeting liver glycerol-3-phosphate phosphatase and the glycerol shunt for citrin deficiency	15	5	20
16:30	Afternoon Break		30		30

Session 7 - Global insights on Citrin Deficiency and Other Relevant Inborn Errors of Metabolism (Chair: Nicola Longo)					
17:00	Saikat Santra <i>Birmingham Women's and Children's NHS Foundation Trust, UK</i>	Variation in the FTTDCD Phase of Citrin Deficiency in Two Ethnic Groups in the UK	15	5	20
17:20	Robin Lachmann <i>University College London Hospitals NHS Foundation Trust, UK</i>	Citrin Deficiency and other UCDs in Adults	15	5	20
17:40	Ljubica Caldovic <i>Children's National Hospital, USA</i>	Transcriptional regulation of citrin gene: A datamining approach	15	5	20
18:00	Alice Sowton <i>The University of Cambridge, UK</i>	Presentation by winner of the Poster Session	10	5	15
18:15	Nguyen Thi Mai Huong <i>National Children's Hospital Hanoi, Vietnam</i>	Presentation by winner of the Poster Session	10	5	15
18:30	John Walker <i>The University of Cambridge, UK</i>	Closing remarks	5		5
18:35	Evening Break		55		55
19:30	Gala Dinner (Speeches: Barbara Yu & Yen How Tai)				