



Citric Foundation  
**CITRIN FOUNDATION IN-PERSON GLOBAL SYMPOSIUM 2023**  
 SCIENTIFIC PROGRAM

Day 1 Program - Monday, Sept 18th 2023			Duration (mins)		
<b>Session 1 – Overview (Co-Chairs: Barbara Yu, Yen How Tai)</b>					
Time	Speaker	Description	Presentation	Q&A	Total
9:00	<b>Barbara Yu &amp; Yen How Tai</b> <i>Citric Foundation</i>	Opening Speech	30	10	40
9:40	<b>John Walker (Nobel Laureate)</b> <i>The University of Cambridge, UK</i>	Keynote Speech – Citrin Deficiency: the Questions and the Paths to Answers	30	10	40
10:20	<b>Johannes Häberle</b> <i>University Children’s Hospital Zürich, University of Zürich, Switzerland</i>	The linkage of biochemistry and pathophysiology of citrin deficiency to clinical presentations, current research landscape and unanswered questions	20	10	30
10:50	<b>Morning Break</b>		30		30
11:20	<b>Hannele Yki-Järvinen</b> <i>University of Helsinki, Finland</i>	Citrin Deficiency Associated Liver Disease (CDALD) in Adults, Pathophysiology and Potential Therapeutic Implications	20	10	30
<b>Session 2 – Developments in the Basic Scientific Understanding of Citrin Deficiency (Chair: John Walker)</b>					
11:50	<b>Sotiria Tavoulari</b> <i>The University of Cambridge, UK</i>	Development of cellular models to elucidate pathogenic mechanisms in citrin deficiency	30	10	40
12:30	<b>Diana Stojanovski</b> <i>The University of Melbourne, Australia</i>	The molecular mechanisms of Citrin biogenesis in health and disease	20	10	30
13:00	<b>Lunch + Photoshoot</b>		75		75
14:15	<b>Georgios Makris</b> <i>University Children’s Hospital Zürich, University of Zürich, Switzerland</i>	Biochemical, cellular and functional methods for studies of citrin deficiency	15	5	20
<b>Session 3 – Novel Therapies I – Protein Replacement &amp; Gene Therapies (Chair: Johannes Häberle)</b>					
14:35	<b>Laura Contreras</b> <i>Universidad Autónoma de Madrid, Spain</i>	Evaluation of aralar as citrin replacement: exogenous aralar partially recovers MAS activity in citrin KO mitochondria	30	10	40
15:15	<b>Gerald Schwank</b> <i>University of Zürich, Switzerland</i>	Correcting metabolic liver diseases by in vivo base- and prime editing	20	10	30
15:45	<b>Julien Baruteau</b> <i>Great Ormond Street Institute of Child Health, University College London, UK</i>	Messenger RNA therapy for liver inherited metabolic diseases	15	5	20
16:05	<b>Poster Session</b>		120		120
18:05	<b>Evening Break</b>		85		85
19:30	<b>Conference Dinner</b>				

Day 2 Program – Tuesday, Sept 19th 2023			Duration (mins)		
<b>Session 4 - Novel Diagnostic Tools &amp; Center Initiatives (Chair: Barbara Yu)</b>					
Time	Speaker	Description	Presentation	Q&A	Total
9:00	<b>Kimitoshi Nakamura</b> <i>Kumamoto University, Japan</i>	Establishing early diagnosis and long-term follow-up for CD with management guidelines	30	10	40
9:40	<b>Johannes Häberle</b> <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	<b>Nicola Longo</b> <i>University of Utah, USA</i>	Identification of Citrin Deficiency by Newborn Screening in Utah	15	5	20
10:30	<b>Morning Break</b>		30		30
<b>Session 5 - Emerging Clinical Evidence on Citrin Deficiency (Chair: Robin Lachmann)</b>					
11:00	<b>Ituro Inoue</b> <i>National Institute of Genetics, Japan</i>	Lessons from liver tissues of CTLN2 patients	20	10	30
11:30	<b>Masahide Yazaki</b> <i>Shinshu University, Japan</i>	Therapeutic Experiences including LOLA for CTLN2 Patients	20	10	30
12:00	<b>Kimihiko Oishi</b> <i>The Jikei University School of Medicine, Japan</i>	Clinical Biochemical Characteristics of Citrin Deficiency in Japan	20	10	30
12:30	<b>Shirou Matsumoto</b> <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	<b>Lunch</b>		60		60
14:00	<b>Marc Hellerstein</b> <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
<b>Session 6 – Novel Therapies II – Development of Therapeutic Interventions for Citrin Deficiency (Chair: Edmund Kunji)</b>					
14:30	<b>Paolo Martini</b> <i>Moderna, Inc., USA</i>	Messenger RNA therapy is a platform therapeutic for the treatment of Rare Genetic Disorders	20	10	30
15:00	<b>Joseph Baur</b> <i>University of Pennsylvania, USA</i>	Modulating NAD+ Availability in Mice with Citrin Deficiency	20	10	30
15:30	<b>Short Break</b>		10		10
15:40	<b>Yukio Nagasaki</b> <i>University of Tsukuba, Japan</i>	Innovations in Amino Acid donors: Design and implementation using amphiphilic block copolymer micelles	20	10	30
16:10	<b>Marc Prentki</b> <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	<b>Afternoon Break</b>		30		30

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

**POSTER PRESENTATIONS (ALPHABETISED ACCORDING TO LAST NAME)**

Name	Title
<b>Araceli del ARCO</b> <i>Centro de Biología Molecular Severo Ochoa CSIC-UAM, Spain</i>	Citrin replacement with Aralar: dissecting Citrin-Aralar interactions
<b>Eri IMAGAWA</b> <i>The Jikei University School of Medicine, Japan</i>	A new drug trial for citrin deficiency with triheptanoin, a medium-odd-chain heptanoic acids (C7), using a mouse model
<b>Bosco JOSE</b> <i>The University of Cambridge, UK</i>	Development and characterization of cellular Citrin Deficiency models
<b>Jun KIDO</b> <i>Kumamoto University, Japan</i>	Selected amino acids and acylcarnitines may effectively detect newborns with citrin deficiency in the newborn screening
<b>Yuta KODA</b> <i>University of Tsukuba, Japan</i>	Poly(amino acid)-Based Self-Assembling Polymer Drugs Enhancing the Efficacy of Chemotherapy by Continuous Release of Amino Acids
<b>Li Eon KUEK</b> <i>Citrin Foundation, Singapore</i>	From Challenges to Solutions: Advancing Pre-clinical Models in Citrin Deficiency Research
<b>NGUYỄN Thanh Phuong</b> <i>National Institute of Genetics, Japan</i>	Molecular relevance of citrullinemia type II and liver cancer
<b>NGUYEN Thi Mai Huong</b> <i>National Children's Hospital Hanoi, Vietnam</i>	The mutation spectrum of the SLC25A13 gene in pediatric cohort in Vietnam
<b>Catherine PALMER</b> <i>The University of Melbourne, Australia</i>	The molecular mechanisms of Citrin biogenesis in health and disease
<b>Alice SOWTON</b> <i>The University of Cambridge, UK</i>	Bioenergetic Profiling of Cellular Models of Citrin Deficiency
<b>Toni VUKOVIC</b> <i>University Children's Hospital Zürich, University of Zürich, Switzerland</i>	Citrin knock-out HepaRG cells as a model for citrin deficiency