



# CITRIN FOUNDATION

## QUARTERLY NEWSLETTER

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### PATIENT ENGAGEMENT

#### International Patient Committee

This year, we launched our International Patient Committee. Alongside acting as regional representatives for the CD community, the committee members meet quarterly to contribute valuable input to drive the Foundation's initiatives and develop our patient engagement strategy. You can learn more about our committee members below.



#### Anita Dimitrova

Anita lives in Sofia, Bulgaria, with her two children: a 15-year-old daughter and an 11-year-old son diagnosed with CD. With a robust background in telecommunications engineering spanning over 15 years, Anita has spent the last 7 years as a Business Analyst at IBM.

#### Jeff Tsao

Based in Boston, Massachusetts, Jeff is father to a daughter with citrin deficiency. Jeff is a scientist-entrepreneur with 20 years of biotech/pharma experience from Flagship Pioneering, Novartis, McKinsey, and Millipore Sigma in new venture creation, R&D, strategy, BD, operations, and commercial. His work has contributed to 3 commercial drugs and several medical imaging technologies.

#### Shirlene Lim

Shirlene is mother to a 6-year-old son who has citrin deficiency and lives with her family in Seattle, Washington. Shirlene has over 20 years of experience in technology. She is currently a Lead Product Manager at Reality Labs and drives the roadmap for Virtual Reality capabilities. Prior to joining Meta, she led strategy and development across consumer and enterprise products at Microsoft and Google.

#### Jennifer Nakao

Jennifer and her family live in Salt Lake City, Utah. She has a daughter born in 2011 and a son born in 2005 both diagnosed with Citrin Deficiency. Jennifer has over 25 years of accounting and operations experience working with multiple industries and locations. Jennifer is currently the CFO and Managing Director of Administration for the World Trade Center of Utah. Prior to her current position, she worked in public accounting for 13 years and a start-up Intellectual Property law firm.

#### Seiji Sugimura

Based in Osaka, Japan, Seiji serves as the president of the Japanese patient association group, Citrin Deficiency Patient Association. Since 2008, he has coped with adolescent and adult citrin deficiency (AACD). He is working with members of the patient group to provide information about the condition and to organize family social gatherings. He joined the committee because he believes that working with the Citrin Foundation will advance clinical research and establish a cure for citrin deficiency.

## Manaho Otsuka

Manaho lives with her family in Ibaraki, Japan, and her daughter, born in 2022, has citrin deficiency. Because she has been able to access various information and connect with patients and their families through the Citrin Foundation, Manaho joined the International Patient Committee in the hope that she can help the Foundation's activities, even if only in a small way. As she has only known the condition for a short period of time, she would like to learn more every day and work on what she wants to achieve for the world based on her own experience.

## Himene Asatsuma

Himene lives in Tokyo, Japan. She graduated from nursing school, but her own experiences led her to become a Child Life Specialist and she now works in a café while volunteering and raising awareness of citrin deficiency. She was diagnosed with CD soon after birth and recovered her health through diet and treatment. She is now able to manage her own health. She joined the International Patient Committee to interact and build supportive relationships with other citrin deficiency patients around the world. She aims to use her experience as a patient and family member to be an approachable and reachable person.

# RESOURCES AND EVENTS

## Rare Disease Day

For this year's Rare Disease Day, we invited our members to make a submission that celebrates an aspect of who they are through something that they love. We wanted to celebrate the remarkable individuals we support for the kaleidoscope of qualities that make them who they are. Beyond the confines of a diagnosis, they are artists, athletes, students, siblings, and dreamers.



## Online Event on Hypoglycemia in CD (in Japanese)

We were thrilled to host our online event this month, inviting Prof. Jun Kido from Kumamoto University to delve into the topic of hypoglycemia in citrin deficiency, a symptom presents quite frequently among our Japanese patient community. The event offered valuable insights and practical advice for patients and families navigating this condition. It also provided a unique platform for patients to have direct interaction with a renowned expert, allowing attendees to ask questions, share experiences, and gain a deeper understanding of hypoglycemia in citrin deficiency. We had over 30 patients and family members attended the talk. We would like to sincerely thank Prof. Jun Kido for his efforts.

## Singapore Patient Gathering

In March, we hosted an in-person social gathering in Singapore for our patients and family members, providing an opportunity to reconnect with familiar faces and welcome new families affected by the condition. Discussions covered a range of topics including patient wellbeing, clinical symptoms, food preferences, treatments, supplements, and strategies for managing the condition.

Participants particularly appreciated resources like the Emergency Card provided by the Foundation, noting its usefulness during international travel. Overall, it was a very good gathering and was well received by our patient community. We look forward to organizing similar events for members in other parts of the world soon.

## Spring Recipes

The Foundation has curated a selection of Spring-inspired seasonal recipes to help our members with meal-planning and keeping variety in their diets.



## RESEARCH UPDATES

### New review publication: “Clinical landscape of citrin deficiency: A global perspective on a multifaceted condition”

We are pleased to announce the publication of “Clinical Landscape of Citrin Deficiency: A Global Perspective on a Multifaceted Condition” in the Journal of Inherited Metabolic Disease (JIMD). This review offers a comprehensive update on citrin deficiency (CD), covering its genetics, diverse clinical presentations, and diagnostic processes, including updated insights from the UK patient cohort by Dr. Saikat Santra. It also introduces the term adolescent and adult citrin deficiency (AACD) to replace citrullinemia type 2 (CTLN2). Importantly, the paper fills a significant gap in the literature aimed at enhancing the clinical understanding of CD. The work is a result of a collaboration between Prof. Jun Kido and Prof. Johannes Häberle, supported by the Citrin Foundation Study Abroad Award to Prof. Kido, and you may read it [here](#).

### Appointment of Prof. Cary O. Harding as an Advisor

We are delighted to announce the appointment of Prof. Cary O. Harding as an Advisor. Prof. Harding is a Professor of Molecular and Medical Genetics and Paediatrics at Oregon Health & Science University (OHSU), and serves as an attending physician in the metabolic clinic at Doernbecher Children’s Hospital and the Medical Director of the Biochemical Genetics Lab.

## PROFESSIONAL EVENTS

### Citrin Foundation Eating Disorders Information Session

In March, the Foundation hosted an online information session for the eating disorder community in the United States, attended by over 25 dietitians, physicians, and clinicians from across the country. The Foundation presented on the critical issue of the misdiagnosis of citrin deficiency as an eating disorder due to patients’ unique food preferences. The presentation highlighted how eating disorder professionals can distinguish CD patients from those with typical eating disorders such as anorexia nervosa.

The session led to actionable strategies, including collaborating with the eating disorder community to develop a CD informational flyer for clinicians to raise awareness and hopefully uncover more patients. The Foundation extends special thanks to Dr. Michael Lutter for his assistance in notifying the eating disorder community about the session.

## 45th Society for Inherited Metabolic Disorders (SIMD) Annual Meeting, Charlotte, North Carolina

In April, we attended the SIMD annual meeting in Charlotte, North Carolina. The Foundation presented a poster titled "The Model of Citrin Deficiency: Adopting a New Approach to Rare Disease" and sponsored a booth offering informational materials on CD and the Foundation to both professionals and patients. Through our participation, we identified more patients across North America, a number we expect to increase as we continue to expand our outreach within the medical community.



Importantly, we also forged new relationships with major industry partners and patient advocacy groups and gained a better understanding of the drug development and regulatory approval process in the US. These crucial developments will undoubtedly contribute towards our goal of finding effective therapies for CD.

## UPCOMING CONFERENCES

### Society for the Study of Inborn Errors of Metabolism (SSIEM) Annual Symposium, September 2024, Porto, Portugal

The Foundation will host and sponsor a Roundtable Discussion titled "Multi-stakeholder Perspectives on the Development of Novel Therapies for Monogenic Metabolic Diseases" at the SSIEM Annual Symposium. The session will be held on 4 September 2024 from 09:00 to 10:15 (Portugal time). Despite advancements in technologies for novel therapies in IMD, significant challenges remain in delivering these therapies to clinics successfully. The session aims to bring together key stakeholders, including researchers, clinicians, regulators, and patient advocacy groups, to discuss critical issues such as the use of biomarkers as surrogate endpoints, translating preclinical findings into clinical trials, designing efficient clinical trials with strong patient and regulatory support, and the regulatory progress in accelerated drug approval programs for rare diseases.

We aim to foster a constructive dialogue between the key stakeholders to discuss potential solutions for these critical issues, ensuring the continued development and ultimate delivery of novel therapies to patients. We also hope that attendees will gain valuable insights and a comprehensive understanding of the successful strategies for developing therapies for patients in the IMD space.

### 65th Annual Meeting of the Japanese Society for Inherited Metabolic Diseases (JSIMD), November 2024, Tokyo, Japan

The Foundation will host a Citrin Deficiency Symposium during the 65th Annual Meeting of the JSIMD. The theme of the symposium is "Advancements in Novel Therapies and New Cellular Models for Citrin Deficiency." The symposium will take place on November 8th from 15:00 to 16:30 (JST) and will be co-chaired by Prof. Kimitoshi Nakamura from Kumamoto University and Prof. Kimihiko Oishi from Jikei University. Barbara Yu from the Foundation will give an overview presentation.

The session will feature esteemed speakers, including Prof. Joseph Baur from the University of Pennsylvania, who will discuss the exploration of novel redox-modulating compounds as potential therapies for CD. Prof. Julien Baruteau from Great Ormond Street Hospital for Children, University College London, will talk about the latest developments in mRNA therapies and their potential applications for CD. Finally, Prof. Jun Kido from Kumamoto University will present on new cellular models of CD, highlighting his collaborative work with Prof. Johannes Häberle to better understand therapeutic interventions.

## EXPERT TALKS

The Foundation regularly holds webinars featuring distinguished speakers who are leading experts in their respective fields. These seminars aim to educate and stimulate discussions, driving progress towards the Foundation's primary objective of developing effective treatments for CD. The details of all past talks are now listed chronologically on our website, which may be viewed [here](#).



There will be other talks taking place this year which are in the process of being finalised. Please stay tuned to upcoming news and updates from the Foundation to find out more.

Thank you for being a part of the Foundation's journey to finding a cure for citrin deficiency. We welcome suggestions on how we can improve our resources as well as ideas for new projects to explore and support you better. If you would like to get in touch, please email us at [patients@citrixfoundation.org](mailto:patients@citrixfoundation.org). We look forward to continuing to support you and your family.

*You can look forward to the next edition of our newsletter in Summer.*

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<https://patient.citrixfoundation.org/en/>

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