



CITRIN FOUNDATION

QUARTERLY NEWSLETTER

We hope you enjoy the Summer Edition of our professional newsletter where you will find updates on our research initiatives, awards, and upcoming events and expert talks. Also included is a summary of our recent patient initiatives.

THE 72ND LINDAU NOBEL LAUREATE MEETING, JUN 2023

The prestigious Lindau Nobel Laureate Meetings are annual scientific conferences held in Lindau, Germany, since 1951 to bring together Nobel laureates and leading scientists around the globe to foster scientific exchange between different disciplines with over 600 participants. We are very pleased and honored to announce that Professor Sir John Walker, Chair of the Foundation's Scientific Supervisory Board, was one of the Nobel laureates to present at this year's physiology/medicine conference with a talk titled "Citrin Deficiency: A Defect in the Urea Cycle". The Foundation would like to express our utmost gratitude to Sir John Walker for his invaluable contribution in raising awareness of citrin deficiency and the Foundation at such a high profile international meeting. The full talk and abstract can be accessed [here](#).

NEWLY FUNDED RESEARCH PROJECTS

The Foundation is pleased to announce that Prof. Marc Hellerstein (University of California, Berkeley) has recently been awarded a research grant to investigate the metabolic alterations in animal models of citrin deficiency (CD). The titles of these projects are appended below:

- Metabolic alterations in primary hepatocytes from animal models and effects of redox manipulations – pilot studies
- Metabolic alterations *in vivo* in liver of animal models and effects of redox manipulations

CITRIN DEFICIENCY ASIA CONSORTIUM

The CD Asia Consortium that was formed at the 6th ACIMD in Bangkok with Prof. Kimitoshi Nakamura as the leader is actively exploring potential clinical studies across CD cohorts in the Asia region. We hope to provide you with exciting developments in future editions of our newsletter.

SOCIETY FOR THE STUDY OF INBORN ERRORS OF METABOLISM (SSIEM). ARCHIBALD GARROD AWARD

We would like to extend our sincere congratulations to Dr. Jun Kido for receiving the prestigious Archibald Garrod Award from the SSIEM for his recent publication "Clinical manifestation and long-term outcome of citrin deficiency: Report from a nationwide study in Japan" Kido et al. *JIMD* 2022. Dr. Kido is the first Asian clinician scientist to receive this award. This highlights the wide-open field in citrin deficiency research where much can still be done. This also is a testimony to the outstanding work that Dr. Kido and the team at Kumamoto University have achieved under the leadership of Prof. Kimitoshi Nakamura, as well as a showcase of the successful collaboration between Kumamoto University and Prof. Johannes Häberle, who was also a meaningful co-author of the paper. You may find more information about the award [here](#).

CITRIN FOUNDATION STUDY ABROAD AWARD

Citrin Foundation is committed to supporting talented individuals dedicated to improving our understanding of citrin deficiency and the lives of those affected by the condition. To facilitate this, the Foundation has created the Citrin Foundation Study Abroad Award and we are pleased to announce Dr. Jun Kido (Kumamoto University) as the first recipient of this award for his exceptional clinical research contributions and dedication to citrin deficiency. This award grants Dr. Kido a unique opportunity to work exclusively on citrin deficiency research with Prof. Johannes Häberle at the University Children's Hospital Zürich for 8 months. We congratulate Dr. Kido for receiving this award and hope that more groundbreaking results will come from his research efforts.

EXPERT TALK SERIES

Our Foundation regularly holds seminars by leading experts in their respective fields that are relevant to our aims. The following are some of our upcoming talks:

- **Prof. Uğur Şahin**, Founder and CEO of BioNTech. Date: September 5th, 2023
- **Prof. Patrick Chinnery**, Professor of Neurology and Head of the Department of Clinical Neurosciences, University of Cambridge. Date: November 16th, 2023

BOSTON CHILDREN'S HOSPITAL METABOLISM GRAND ROUNDS, AUG 2023

We are pleased to announce that Prof. Johannes Häberle will be giving an online lecture on citrin deficiency at the Boston Children's Hospital Metabolism Grand Rounds. This will be the first dedicated presentation on citrin deficiency in the history of the Grand Rounds, making it a significant milestone in raising awareness for the condition in North America. Prof. Häberle's talk is scheduled for 22nd August 2023.

PARTNERSHIP WITH NATIONAL UREA CYCLE DISORDERS FOUNDATION (NUCDF)

The Foundation is delighted to announce a collaborative partnership with NUCDF. NUCDF is the driving force behind critical research to improve the understanding and management of urea cycle disorders (UCD), find new treatments, and ultimately a cure. NUCDF serves as a lifeline to UCD patients, families, and medical professionals worldwide seeking information, support, and HOPE.

UPCOMING CONFERENCES

SSIEM Annual Symposium Aug 2023, Jerusalem, Israel

The SSIEM annual symposium is renowned for gathering global experts on inborn errors of metabolism (IEM) to discuss the latest research progress in the field. This year, there will be a special focus on citrin deficiency. Our Co-Founder, Barbara Yu, will give a talk about our Foundation and our unique approach to rare diseases. Prof. Johannes Häberle will deliver a plenary lecture on citrin deficiency. Dr. Jun Kido will present his work from the publication on the nationwide study of citrin deficiency in Japan (Kido et al. 2022, *JIMD*) as a recipient of the prestigious Archibald Garrod Award. He will also present on his recent research that is supported by the Foundation on a new method of newborn screening (NBS) for citrin deficiency. Dr. Kido worked closely with Prof. Kimitoshi Nakamura on these projects. Finally, Prof. Kimihiko Oishi and his team will present the results of their work on the CD knock-out mouse model, which was supported by the Foundation. We hope that these cumulative efforts will help to raise awareness on the condition and on our work, encouraging more professionals to join our efforts to find a cure and provide better care for patients.

64th Japanese Society for Inherited Metabolic Diseases (JSIMD) Annual Symposium Oct 2023, Osaka, Japan

The Foundation will attend this year's JSIMD meeting in Osaka to host a dedicated session on citrin deficiency, where progress on new research initiatives on CD will be presented. We will update you with more details in our next newsletter.

GLOBAL CITRIN DEFICIENCY SYMPOSIUM

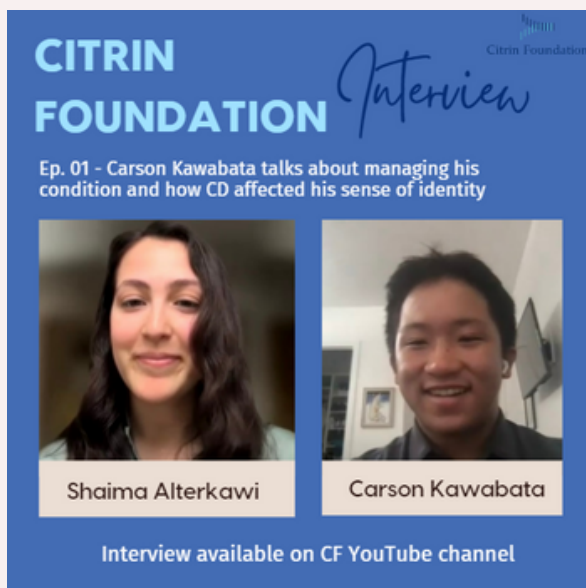


We would like to provide you with exciting updates about the upcoming Citrin Foundation Global Symposium. Preparations are proceeding well for the event, and we are pleased to share that approximately 50 attendees have already confirmed their participation, making it an exceptional gathering of experts dedicated to advancing citrin deficiency research.

The symposium's program will cover a broad range of topics, including cutting-edge research in basic science and clinical studies, new developments in potential therapies, and a dedicated session for patients, amongst other exciting topics. We anticipate in-depth discussions and collaborations that will lead to breakthroughs in citrin deficiency research. If you would like further information about the symposium, please reach out to us at symposium2023@citrinfoundation.org

PATIENT RESOURCES

MEMBER INTERVIEW SERIES



We also launched our interview series. In the first episode, our Patient Engagement Manager speaks to Carson Kawabata, an inspirational citrin deficiency patient and very active member of our community. Carson talks about managing his condition and how CD has affected his sense of identity and his relationships with others. An article version of the interview will soon be made available in Japanese.

For our second episode, we welcomed Himene Asatsuma, a citrin deficiency patient living in Tokyo, Japan who illustrated our primary school resource. Himene talks about how she gained understanding from her peers in her own way and her thoughts on living with citrin deficiency. The episode ends with a powerful message from Himene to children with CD. An article version of the interview will soon be made available in English.

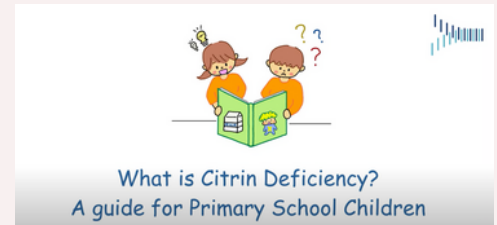
YOUTUBE CHANNEL LAUNCH

In April, the Foundation launched a YouTube channel to share resources in a visually engaging and easily digestible format which we believe would be valuable to our members as well as professionals or members of the general public who would like to learn more about CD.



GUIDE FOR PRIMARY SCHOOL CHILDREN

As part of our drive towards building more age-specific resources, we created a short video guide aimed at primary school children to help explain citrin deficiency. We partnered with one of our Japanese members, Himene Asatsuma to illustrate this resource.





MULTI-LANGUAGE RESOURCES

We are delighted to announce the launch of our Multi-language Resource Hub on our website, now offering our citrin deficiency patient flyer in German and Korean. We have plans to expand this initiative to include the following languages: Arabic, Vietnamese, Thai, and Chinese.



EMERGENCY CARD


The patient engagement team has designed a wallet-sized patient emergency card. This card provides crucial information about the patient and their condition to ensure rapid and accurate medical attention during emergencies.



**EMERGENCY CARD**
I have a condition called: CITRIN DEFICIENCY
(CD: Inherited metabolic disorder)

PATIENT INFORMATION:
Name: _____ Date of Birth: _____
Blood Type: A / B / O / AB / Unknown Allergies (if any): _____
Current CD stage: NICCD / FTTDCD / Compensatory period / CTLN2
Hospital/Name of doctor: _____

MEDICATIONS/SUPPLEMENTS (IF ANY):


CONTACT EMERGENCY CONTACT(S):
Name: _____ Relationship: _____
Phone: _____

<https://citrinfoundation.org/>

**EMERGENCY CARD**
I have a condition called: CITRIN DEFICIENCY
(CD: Inherited metabolic disorder)

IMPORTANT INFORMATION:

- **Please do not treat CD patient with high glucose infusion and GLYCEROL®.** There are no issues with general infusion to treat hypoglycemia, diarrhea, or vomiting, but in order to promote faster recovery, it is advisable to let patient consume what they wish after their appetite returns.
- **Avoid fasting** or prolonged periods without eating.
- Monitor for signs of **hyperammonemia** (elevated ammonia levels) and **hypoglycemia** (low blood sugar).
- Please be aware of **metabolic decompensation**.

<https://citrinfoundation.org/>

Available to download and print from our patient website.

LEAFLETS

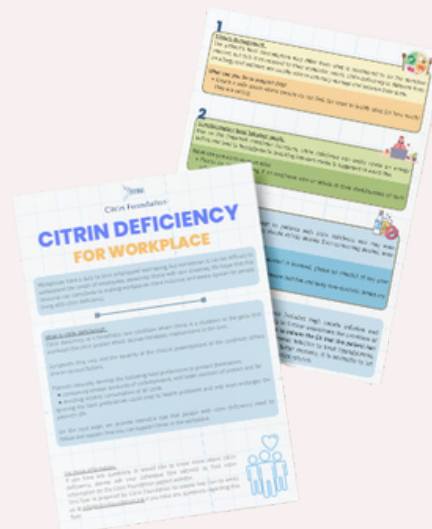
Birthday food flyer

Building upon the launch of our recipe section at the beginning of this year, we wanted to create a resource to help parents with planning birthdays by developing recipes that balance CD dietary requirements, flavour, and ease.

Workplace flyer

Workplaces have a duty to their employees' well-being, but sometimes it can be difficult to understand the needs of employees, especially those with rare diseases. As a contribution to helping to make workplaces more inclusive and aware spaces for people living with citrin deficiency, we created this resource for our members to share with their employers if they wish.

All leaflets are available to download from our patient website.



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.

If you would like to get in touch, please email us at info@citrinfoundation.org for general inquiries or grants@citrinfoundation.org for grant inquiries.

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