WINTER, 2024

CITRIN FOUNDATION QUARTERLY NEWSLETTER

2023 brought great strides for the Foundation on all fronts including research, raising awareness, and patient support for citrin deficiency (CD), culminating in our second in-person global symposium in September.

Throughout the past year, the Foundation has made good progress in our research efforts by deepening collaborations with our existing consortium and working with new collaborators. Our clinical strategy has also yielded good results where we have identified global clinical cohorts of over 1,100 patients. Furthermore, we have redoubled our efforts to provide holistic support to our patients through a range of new resources and opportunities for patients and families.

As we embark on the new year, our commitment to making a lasting difference in the lives of those affected by citrin deficiency remains unwavering. We look forward to making further progress in the development of novel therapies and laying the important groundwork for future clinical trials, including the establishment of a global patient registry. Your continued support is the driving force behind our achievements, and we look forward to the challenges and triumphs that lie ahead.

RECAP OF 2023

GLOBAL IN-PERSON SYMPOSIUM

The highlight of 2023 was our Second In-person Global Symposium in Cambridge, UK, where some of you were present. It was a great gathering for our research, clinical and patient community where many great ideas were discussed, and friendships were formed as we were united by our shared goals and visions. Please click here if you are interested to learn more about it.

ESTABLISHMENT OF NEW CENTERS

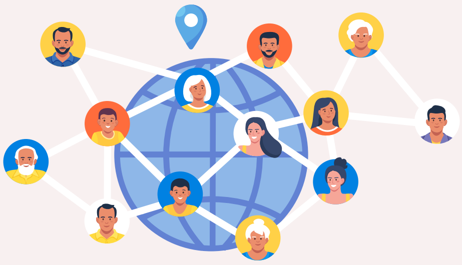
CD Center of Excellence at Kumamoto

The Citrin Deficiency Center of Excellence at Kumamoto University led by Prof. Kimitoshi Nakamura and sponsored by our Foundation have made great progress towards the goal of uncovering more CD patients, improving newborn screening and genetic diagnosis, standardizing diagnosis and management guidelines for CD, and developing a CD patient registry. It has also acted as a leader for clinical studies in Japan and Asia.

Urea Cycle Disorders (UCD) Translational Research Center at Zürich

The Foundation has announced the significant initiative of the establishment of the UCD Translational Research Center at University Children's Hospital Zürich, University of Zürich, to be led by Prof. Johannes Häberle. This Center will focus on translational research and clinical studies for UCD, with CD as a model disease.

CLINICAL COHORTS



The Foundation has significantly expanded our clinical cohort in the past year to over 1,100 patients, by identifying more than 450 patients from various cohorts globally in countries such as Taiwan, Korea and Vietnam. One such cohort of over 300 patients is at the National Children Hospital Hanoi under the care of A/Prof. Nguyen Pham Anh Hoa, our close collaborator.

UREAGENESIS TEST

After successful pilot studies assessing the safety and diagnostic utility of the ureagenesis test in some CD patients, conducted in collaboration with Prof. Johannes Häberle, who developed the test with his team, we are expanding the scope of these studies to larger patient cohorts.

ADULT METABOLIC STUDY

This clinical study, a collaborative effort involving the Foundation, the University of Helsinki (Prof. Hannele Yki-Järvinen), the University of California Berkely (Prof. Marc Hellerstein), and the University of Oxford (Prof. Leanne Hodson), will use state-of-the-art stable isotope tracer methods to precisely determine the metabolic status of adult CD patients. If you are interested to learn more about this please contact us at patients@citrinfoundation.org.

RAISING GLOBAL AWARENESS

Throughout 2023, the Foundation actively engaged and presented in numerous international meetings to raise awareness of CD within the IEM community:

- 44th Society for Inherited Metabolic Disorders (SIMD) Annual Meeting in Salt Lake City, Utah
- 6th Asian Congress on Inherited Metabolic Disease in Bangkok, Thailand
- 64th Annual Meeting of the Japanese Society for Inherited Metabolic Diseases in Osaka, Japan

In August last year, the Foundation also organized a talk in partnership with Boston Children's Hospital's Metabolism Grand Rounds, inviting Prof. Johannes Häberle to speak about citrin deficiency.

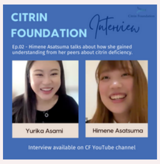
More recently, the Foundation released a global awareness video that has been shared widely on our various social media channels. You may watch the full video on our YouTube Channel.

PATIENT ENGAGEMENT

Resources and support

Last year also marked a surge in the Foundation's capabilities to support our patients as we dedicated efforts to create a number of valuable resources for our membership:

- Food-related resources including seasonal recipes, practical snack ideas and special event flyers
- Emergency card template
- Age-specific resources i.e. alcohol and peer pressure resource, revamped workplace flyer, resource for primary school children



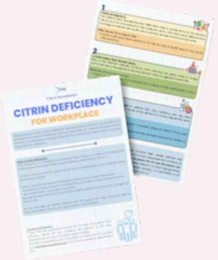
CITRIN FOUNDATION Interview

Ep.02 - Himene Asatsuma talks about how she gained understanding from her peers about citrin deficiency.

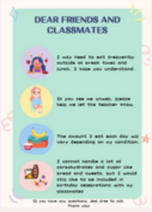
Yurika Asami

Himene Asatsuma

Interview available on CF YouTube channel



CITRIN DEFICIENCY FOR WORKPLACE



DEAR FRIENDS AND CLASSMATES

Membership Update

In the past year, our patient global membership has grown significantly to over 300 patients worldwide, with 96 new patients joining our community. Our professional network has also grown with professionals joining from Turkey, Malaysia, Japan, Indonesia, Vietnam, the UK and the US.

WHAT'S NEW THIS JANUARY?

ADVISORY BOARD

The Foundation has appointed four new advisors who are the leading experts in their fields, and whom we believe will help us to further advance our goals. We are grateful to them for joining our efforts, and we look forward to their future contributions.

Dr. Nicholas Ah Mew
Dr. Nicholas Ah Mew is Director of the Inherited Metabolic Disorders Program at Children's National Hospital and is Associate Professor of Paediatrics at The George Washington University.

Prof. Marc Hellerstein
Prof. Marc Hellerstein is a Professor of Human Nutrition at the University of California, Berkeley, where he occupies an Endowed Chair (Dr. Robert C. and Veronica Atkins Chair). He is also a Professor of Endocrinology, Metabolism and Nutrition in the Department of Medicine at the University of California in San Francisco

Dr. Robin Lachmann
Dr. Robin Lachmann leads the Charles Dent Metabolic Unit at University College London Hospitals, one of the largest units in the world looking after adults with inherited metabolic diseases.

Stephen Yang
Stephen Yang is Head of Strategy for Immunology at Novartis where he is responsible for coordinating company-wide research, commercialisation and business development activities within the therapeutic area.

ADULT PATIENT SUPPORT GROUP

We are excited to announce the launch of our adult patient support group. This group will provide a safe space for our adult patients to connect and offer mutual support.

If you would like to join our adult patient peer support group, please visit our patient website and fill out the registration form.

INTERNATIONAL PATIENT COMMITTEE

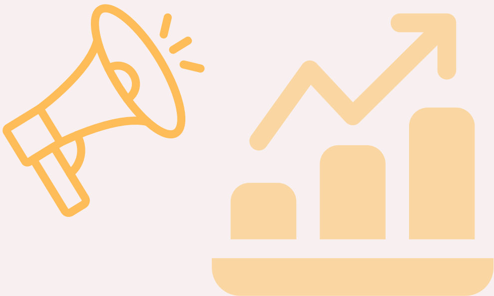
This month we officially launched our International Patient Committee and held our first committee meeting. The committee will be working to meet the following objectives:

1. Acting as ambassadors for the Foundation and the work we do
2. Raising awareness about CD
3. Acting as regional representatives for the CD community
4. Contributing valuable input to drive the Foundation's initiatives

If you or someone you know is interested in learning more about this initiative, please email patients@citrinfoundation.org

LOOKING AHEAD FOR THIS YEAR:

The Foundation will be working towards a number of goals in 2024, including:



Uncovering more patients and increasing awareness of CD

The Foundation has developed a comprehensive strategy to uncovering more CD patients in the coming year, with a particular focus on the United States. We will be participating in and hosting a number of events as well as utilising social media and continuing with direct outreach.

Setting up a patient registry

The next big step in navigating the research landscape and furthering our goals is to set up a patient registry. Having a global CD patient registry would allow us to design more targeted and effective interventions, accelerate the development of new therapies and improve the health outcomes of our patients.

Continuing to develop our support initiatives and increasing patient engagement

The patient engagement team is also working on a number of helpful resources for our patients and families. While we always welcome suggestions and new ideas, we also encourage a co-production approach to the things we are working on and will be inviting our members to get involved in various projects throughout the year.

Currently, we are looking for patients, family members and professionals who would like to share their stories or research in our interview series. These can be done in written or video form. If you are interested in signing up, please email patients@citrinfoundation.org. You can read or watch our other patient stories and interviews on YouTube and 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 and support you better. If you would like to get in touch, please email us at patients@citrinfoundation.org. We look forward to continuing to support you and your family.

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