



CITRIN FOUNDATION QUARTERLY NEWSLETTER

CITRIN FOUNDATION GLOBAL SYMPOSIUM 2023

We recently held our second in-person Symposium, 18th to 20th Sep 2023, hosted at Magdalene College, University of Cambridge, UK. It was attended by over 50 researchers, clinicians, and biotech representatives from all over the world. The event was a resounding success and demonstrated the new, global and holistic approach the Foundation has adopted to act as a nexus and bring together professionals with multidisciplinary backgrounds to work collaboratively.



The event included a two-day scientific program and a half-day patient-oriented session. The scientific sessions provided our dedicated professional network the opportunity to reflect on the progress made thus far and to address the challenges that lie ahead.

In the scientific program, the Foundation dedicated a space for patient stories at the poster session. Three storyboards had been adapted from interviews with three of our members. We hope that they can shed light on the ways in which this condition can affect patients and their families in various ways. These are now available to read on our patient website. We would like to thank those who took the time to share their experiences.

- [Interview with the Chairman of the Citrullinemia Patient Association in Japan](#)
- ["My brother, who died 11 years ago, had CTLN2."](#)
- ["We lost our new-born twins to CD."](#)

The half-day patient session was both memorable and heart-warming. Over 150 patients and family members joined us in person and online from Japan, Korea, Bulgaria, United States, United Kingdom, Indonesia, Malaysia, India, Pakistan, Austria, Vietnam, Greece and Taiwan.

We are incredibly grateful to siblings Carson and Akemi Kawabata who gave live presentations about their experiences growing up with citrin deficiency (CD). **We also debuted our patient video, which told the diverse stories of our membership.** [The video](#) is now available for viewing on YouTube.



We let our patients understand what we do by showing them a video comprised of the researchers and clinicians we work with, along with having presentations by Barbara Yu, John Walker, Johannes Häberle and Kimitoshi Nakamura, who specifically talked about current CD management and guidelines. Sami Qadri also presented on the Adult Metabolic Study; a planned clinical study slated to begin next year.

Finally, our Patient Engagement and Clinical Program Manager, Shaima Alterkawi, spoke about what we have accomplished over the last year as well as the future of our support initiatives. The session ended with a Q and A session with our Global Expert Panel, offering those in attendance an opportunity to voice their concerns and have their questions answered.

We hope that this Symposium has succeeded in providing an excellent opportunity for our patients to have their voices heard, have a better understanding of what we do and be more engaged in our community. As we move forward, we are committed to continuing the spirit of collaboration and the sense of unity that this symposium has instilled in us, leaving us confident in our ability to one day solve CD holistically.



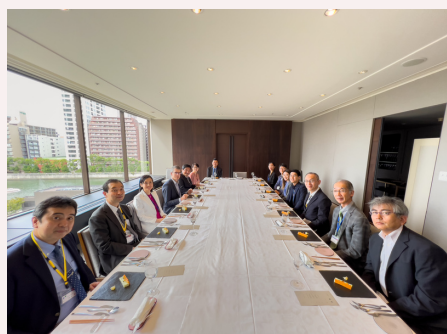
Thank you once again to all who attended and contributed to this remarkable event. We look forward to what lies ahead. Please visit our YouTube channel to view our [symposium highlights reel](#).



CONFERENCE UPDATES

64th Annual Meeting of the Japanese Society for Inherited Metabolic Diseases (JSIMD) – Oct 2023, Osaka, Japan

Earlier this month, the Foundation sponsored a Citrin Deficiency (CD) symposium at the 64th JSIMD Annual Meeting in Osaka. The CD symposium was co-chaired by Johannes Häberle and Kimitoshi Nakamura. Our Co-founder, Barbara Yu, delivered a presentation that highlighted the new CD research initiatives and progress made by the Foundation. Johannes Häberle discussed the pre-clinical requirements for improving the management of CD. Kimitoshi Nakamura presented on the role of the CD Center of Excellence. Finally, Ituro Inoue closed the session with a presentation about the dysregulation of gene expression in liver samples from CTLN2 patients. Additionally, the Foundation co-hosted a CD luncheon with Kimitoshi Nakamura, where many prominent regional clinicians with an interest in CD attended, further advancing our goals.



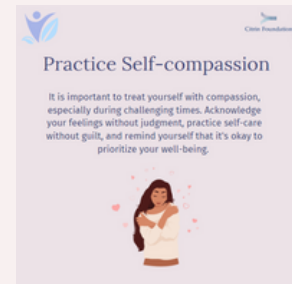
International Patient Committee



We are excited to invite passionate adult patients and parents of patients from our global community to join our Citrin Deficiency Patient Committee. As part of this committee, you will serve as regional patient representatives, contributing your unique insights and experiences to a global board. Together, we will hold regular feedback meetings to review ongoing initiatives, ensuring that the voices of our diverse community are heard and valued. Furthermore, you will have the opportunity to engage in brainstorming sessions and participate in strategic planning for the future. By joining us, you will act as a leader within your regional patient community, fostering connections, providing support, and making a real difference in the lives of those affected by CD. To apply to join us for this initiative please email patients@citrinfoundation.org.

Wellbeing Wednesdays

One of the Foundation's objectives is to improve the daily life of our members. One of the ways in which we are working to do that is by supporting your wellbeing. Prioritising self-care improves the overall quality of life, sustains caregiving efforts, and prevents burnout. It is essential for rare disease patients and their families to prioritise their wellbeing. By doing so, they can maintain their physical and mental health, develop healthy coping strategies, and build resilience in the face of the unique challenges they may face. Therefore, we have started our "Wellbeing Wednesdays" series on our social media to bring you wellbeing tips and help support you holistically.



1-1 support calls

Further to the above, we wanted to reiterate that 1-1 calls with our Patient Engagement Manager are now available to book via email. These calls are designed to provide you with a dedicated space to share your thoughts and experiences, positive or negative. During these conversations, we encourage you to express any feedback you may have. Your insights are invaluable to us. Additionally, these calls serve as a practical platform to connect you with the resources you might need. Whether you're looking for support or information, our Patient Engagement Manager is here to assist you.



AUTUMN RECIPES

Embrace the cozy flavours of fall with our new specially curated seasonal recipes. We have put together an array of low-carb recipes that not only capture the essence of the season but also focus on convenience and ease for family meals.



Featured Recipe

Autumn Stew

Ingredients:

- 1 pound (450g) beef stew meat, cubed
- 2 tablespoons olive oil
- 1 large onion, chopped
- 2 cloves garlic, minced
- 1 cup green beans, trimmed and cut into bite-sized pieces
- 2 carrots, peeled and sliced
- 2 parsnips, peeled and sliced
- 2 potatoes, peeled and cubed
- 2 cups butternut squash, peeled and cubed
- 4 cups beef or vegetable broth
- 1 can (14.5 oz) diced tomatoes
- 2 bay leaves
- 1 teaspoon dried thyme
- Salt and pepper to taste
- Chopped fresh parsley for garnish (optional)



Instructions:

- Heat the olive oil in a large soup pot over medium-high heat.
- Add the cubed beef stew meat and brown on all sides, about 5-7 minutes. Remove the meat from the pot and set it aside.
- In the same pot, add the chopped onion and garlic. Cook for 2-3 minutes until they become fragrant and translucent.
- Return the browned beef to the pot, and add the carrots, parsnips, potatoes, butternut squash, and green beans. Stir to combine.
- Pour in the beef or vegetable broth and diced tomatoes (including their juice). Add the bay leaves and dried thyme. Season with salt and pepper to taste.
- Bring the mixture to a boil, then reduce the heat to low. Cover the pot and let it simmer for 1.5 to 2 hours, or until the beef and vegetables are tender and the flavors meld together.
- Remove the bay leaves before serving.
- Taste the stew and adjust the seasoning, if necessary.
- Ladle the autumn stew into bowls, garnish with chopped fresh parsley if desired, and serve hot.



Halloween flyer

In our ongoing mission to ignite a passion for food among children, we have crafted a Halloween flyer that embodies creativity, spookiness, and, most importantly, fun. We believe that involving kids in the process of preparing food not only makes them appreciate the effort that goes into it but also fosters a lifelong love for cooking and good nutrition. These recipes are not just about nourishment; they are about sparking creativity and encouraging children to explore the wonders of the kitchen. Our recipes are tailored to transform ordinary ingredients into spooky delights.

Beyond the culinary adventures, our flyer aims to relieve the burden on parents. We understand the challenges of juggling busy schedules, work commitments, and the desire to create memorable experiences for your children. We've taken care of the planning, so you can focus on the joyous moments spent together in the kitchen, crafting delicious treats that will be remembered long after the Halloween night is over.

UPCOMING EVENTS

Adult Metabolic Study Patient Information Session

This proposed landmark study will for the first time precisely determine the metabolic status in patients and in doing so, provide answers to many outstanding questions and contribute towards the development of effective treatments for the condition. The study will be taking place in the United Kingdom, and we will be recruiting eligible patients next year.

Without willing participants, research toward effective treatments cannot move forward. That is why we strongly believe in involving patients in the design and planning of such studies. This is your chance to be a part of something bigger. The study results will not only make a difference to yourself and other patients living with CD, but also for wider metabolic research. Would you be interested in this study? If not, why? What adjustments could be made to the study protocol and procedures to change your mind? We would love to hear feedback from you to make our studies better. We will be hosting a patient information session next month to discuss this. Please email patients@citrinfoundation.org if you would like to learn 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@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 January.