APRIL 2023

Citrin Foundation

# QUARTERLY NEWSLETTER

We are very excited to be rolling out our new quarterly newsletter. This first issue will cover updates from the last year including membership cohort updates. Future issues will then cover quarterly updates. This newsletter will cover updates about ongoing patient engagement initiatives as well as new projects.

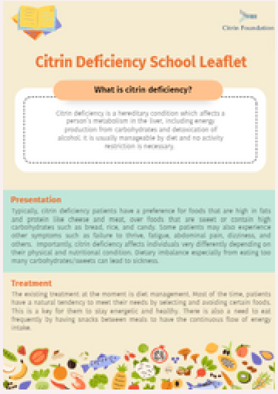
## MEMBERSHIP COHORT UPDATES

In the past year, our global membership has grown significantly, with patients and their family members from Australia, Austria, Canada, China, Hong Kong, Indonesia, Japan, Malaysia, Philippines, the US and the UK joining our global patient community which now have more than 150 members

Our Foundation also established a global physician registry for clinicians with CD patients which currently consists of more than 60 clinicians internationally, from countries and regions such as Europe, Japan, Korea, Vietnam, the Middle East, the US, and the UK.

## PATIENT LEAFLETS

We created a comprehensive resource covering an overview of the condition, current treatment and management, available support, and ways in which patients could get involved with the Foundation. We have also revamped our school leaflet with tips on how teachers can support students living with citrin deficiency and created a resource for school children to give to their classmates. Furthermore, we have created a snack suggestion sheet with easy food and drink ideas. All leaflets are available to download from our patient website and have been distributed to our membership and professional network.



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Citrin Deficiency School Leaflet

What is citrin deficiency?

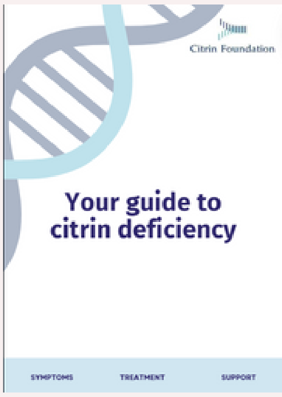
Citrin deficiency is a hereditary condition which affects a person's metabolism in the liver, including energy production from carbohydrates and detoxication of alcohol. It is usually manageable by diet and no activity restriction is necessary.

Presentation

Typically, citrin deficiency patients have a preference for foods that are high in fats and protein like cheese and meat, over foods that are sweet or contain high carbohydrates such as bread, rice, and candy. Some patients may also experience other symptoms such as failure to thrive, fatigue, abdominal pain, dizziness, and others. Importantly, citrin deficiency affects individuals very differently depending on their physical and nutritional condition. Dietary imbalance especially from eating too many carbohydrates/sweets can lead to sickness.

Treatment

The existing treatment at the moment is diet management. Most of the time, patients have a natural tendency to meet their needs by selecting and avoiding certain foods. This is a key for them to stay energetic and healthy. There is also a need to eat frequently by having snacks between meals to have the continuous flow of energy intake.



Citrin Foundation

Your guide to citrin deficiency

SYMPTOMS TREATMENT SUPPORT

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Snack ideas

At home
- Green peppers filled with cheese
- Boiled eggs
- Strawberries and citrus fruits
- Cucumber and carrot sticks with hummus or yoghurt dip
- Roasted chickpeas with spices
- Edamame beans
- Smoked salmon
- Avocado
- Tuna salad with cucumber, tomato and boiled egg

On the go
- Peanuts
- Pistachios
- Macadamia nuts
- Walnuts
- Beef jerky
- Cheese sticks
- Flavoured rice crackers

Beverages
- Unsweetened soy milk, peanut butter and MCT smoothie
- Unsweetened almond milk
- Lemon and sparkling water
- Lemon water with cucumber and mint

What are your favourite snacks?

Submit your suggestions or recipes to patients@citrinfoundation.org

DEAR FRIENDS AND CLASSMATES

I may need to eat frequently outside of break times and lunch. I hope you understand.

If you see me unwell, please help me let the teacher know.

The amount I eat each day will vary depending on my condition.

I cannot handle a lot of carbohydrates and sugar like bread and sweets, but I would still like to be included in birthday celebrations with my classmates

If you have any questions, feel free to ask. Thank you!

# RECIPES

Many of our members have expressed a need for more food-related resources. Finding recipes that are tasty, varied and meet the dietary requirements of individuals living with citrin deficiency can be difficult and often makes navigating meal planning and grocery shopping quite stressful. Therefore, in January, we launched a new initiative focused on recipe gathering and development. Our membership has been wonderful in their contributions to helping us build our recipe log on our patient website as well as other food-related resources like our snack suggestion sheet. We look forward to continuing to expand this resource. If you would like to contribute, please submit your recipe and photo of the dish to patients@citrinfoundation.org

## Featured Recipe

### Healthy bun-less poultry burgers [Serves 4]

### Ingredients

- 400g turkey or chicken mince
- 1 egg
- 1/2 tsp Paprika
- 1 pinch Salt
- 2 cloves of minced garlic
- 1/2 red onion minced
- Handful of fresh basil and coriander chopped
- Dash of chilli flakes
- 40g breadcrumbs
- Optional toppings: lettuce, avocado, and your favourite sauces.

### Instructions

1. In a large bowl, mix all the burger ingredients using your hands, to form a large ball which has all the ingredients evenly distributed.
2. Split the mixture into 4 smaller round balls, and use your hand to flatten into burger shapes
3. Heat olive oil in a large, non-stick pan over a medium heat
4. Add the burgers to the oil, and cook for 5 minutes on one side, until browned
5. Turn using a spatula and cook for a further 5 minutes on the other side - until browned and cooked through
6. Serve with your favourite toppings and a side-dish of your choice.

## EVENTS

**English-speaking patient and family webinar December 2022**
In December 2022, we hosted our first-ever online patient and family gathering for our English-speaking members. Those who joined us spanned four continents, coming from Singapore, Canada, Malaysia, Bulgaria, Pakistan, the United Kingdom, the United States, Vietnam, China, Hong Kong, Australia, Indonesia, and Greece.

The program consisted of a presentation segment, a member's experience segment, and an open discussion. Our Co-founder, Barbara Yu, first gave a formal introduction to the Foundation and the work we do and summarised our upcoming studies. Our Director of Scientific and Medical Development, Li Eon Kuek, then gave a stellar presentation about our latest understanding of the condition and how that affects diet management and supplementation. A recording of his presentation can be viewed on our main website. Lastly, Shaima Alterkawi, our Patient Engagement and Clinical Program Manager spoke about our ongoing patient engagement initiatives as well as our plans for 2023.

Two of our members kindly volunteered to contribute their stories. Carson Kawabata spoke about his experience living with citrin deficiency as an active teenager who refuses to let it hold him back. Anita Dimitrova shared her story of being a mother learning to manage her son's condition.

**Japanese-speaking patient and family webinar March 2023**
In March 2023, Citrin Foundation ran the Japanese iteration of the above patient and family webinar. Our Foundation Associate, Yurika Asami, presented on the Foundation's background and research initiatives, the latest understanding of citrin deficiency and its implications for patients, covering the energy deficit theory and the direct implications for patients, and the Foundation's patient engagement and support initiatives to our Japanese patient community.

## PEER SUPPORT GROUP

We also launched our global peer support community in January 2023. This takes the form of an informal chat group of other parents and adult patients where you can exchange ideas and build a support network as well as digital peer support meetings facilitated by the Foundation. Members also have a chance to have a regular point of contact with the patient engagement team. Discussion topics that have been covered in these groups have ranged from travel tips and snack suggestions to questions revolving round childcare and explaining CD to extended family. The frequently asked questions and tips that are shared within the group are used to inform the development of further resources for our membership. Results from an anonymous survey conducted at the end of February showed that members of our peer support community find the group helpful for both informational and emotional support.

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

## GLOBAL DIGITAL COMMUNITY

Last October, we launched our digital community on social media. We share once a week and this includes diet tips, patient stories, and research updates and showcases our involvement with conferences.

# RESEARCH AND PROFESSIONAL UPDATES

Over the past year, Citrin Foundation has continued to provide funding to research projects to further our aims at better understanding citrin deficiency and to develop promising treatments for the condition. Compared to 2021, we have since doubled the number of research projects funded. These projects are being carried out by world-leading clinicians and scientists from renowned academic institutions worldwide. You can read more about our research projects on our website.

## GLOBAL OMICS STUDY

In order to find a good biomarker to track disease progress and evaluate treatments, Citrin Foundation has initiated a global omics study that aims to analyze blood samples from at least 100 citrin deficiency patients by applying a combination of transcriptomics, proteomics, metabolomics, and lipidomics. This landmark study involves multiple international study sites (JP, TW, GS, KR, UK, USA). The study is slated to begin in 2023. Please participate when you get a chance.

## GLOBAL CONFERENCE UPDATES

We presented at various conferences among the relevant professional scientists, clinicians to raise the awareness of citrin deficiency and share the work that our Foundation has been doing.

### Ureagenesis Conference in Spain

In October 2022, Citrin Foundation sponsored the first-ever spotlight program for citrin deficiency at the International Conference on Ureagenesis Defects and Allied Conditions held in Valencia. Our co-founder, Barbara Yu, introduced the Foundation and our unique approach to rare disease. John Walker, Nobel Laureate and the chairman of our Scientific Supervisory Board, gave the keynote speech. This was followed by a series of lectures in which we discussed various aspects of the condition and highlighted our collaborative approach to solving citrin deficiency.

### JSIMD in Kumamoto

In November 2022, we attended the 63rd Japan Society for Inherited Metabolic Diseases in Kumamoto, Japan. , Barbara Yu, presented to a large audience of clinicians and scientists with a strong interest in our work during the citrin deficiency symposium. The Foundation also co-hosted a citrin deficiency luncheon prior to the CD symposium where clinicians with interest in CD were invited. Many of them showed interest in our global omics study which we will be rolling out later this year.

### SIMD in the United States

This year, Citrin Foundation sponsored an exhibition booth at the SIMD annual meeting in Salt Lake City, Utah from March 18th to March 21st. Alongside attending the impressive program of talks, our patient engagement team focused on networking and expanding our reach in the United States and had the pleasure of meeting some of our US-based members face-to-face.

### ACIMD in Bangkok

The Foundation attended the 6th Asian Congress on Inherited Metabolic Diseases (ACIMD) in Bangkok in March. Barbara Yu gave an engaging presentation at the conference, on the Foundation's global approach towards solving citrin deficiency and highlighted the importance of working together with scientists and clinicians in Asia.

The first Citrin Deficiency Consortium for Asia was established during the meeting with the Foundation as the Sponsor. The Consortium aims to meet regularly to work on initiatives such as to uncover more patients, creation of a CD patient registry and potential clinical studies on CD.

## UPCOMING EVENTS

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### Cambridge Symposium

The Foundation is pleased to be hosting the second in-person Global Citrin Deficiency Symposium this autumn. The last half day of the event, September 20th, is dedicated to our patients. Prominent clinicians in the field will talk about citrin deficiency and and is a great opportunity to participate in fruitful discussions and connect with other patients and families in our community.

This event will be held at Magdalene College, a beautiful venue with a rich history in the academic city of Cambridge, UK. If you would like to know more or are interested to attend, please contact: patients@citrinfoundation.org

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 July.***