MitoCAMB Patient Newsletter

Issue 13
September 2025

UNIVERSITY OF CAMBRIDGE

MITO CAMB

WELCOME

...to our Cambridge Clinical Mitochondrial Research Group patient newsletter.

You are receiving this because you have previously signed up to help with research into mitochondrial disease or neurodegenerative disorders. Our aim is to provide you with the latest news from the research group, giving you more information on who we are, what we do, the science behind our work, and how we can support you, our patients.

WORLD MITOCHONDRIAL DISEASE WEEK

15th – 21st September 2025

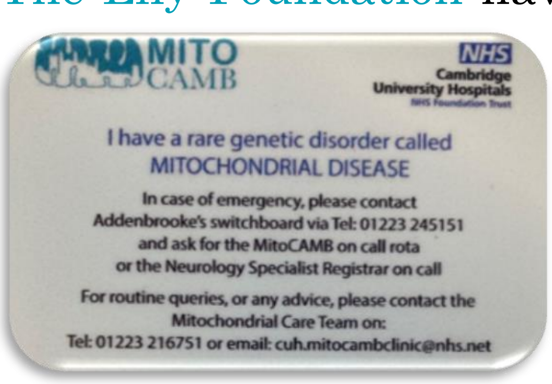
This week marks World Mitochondrial Disease Week! The theme this year is 'Decode the Mito Puzzle' and aims to raise awareness of the important role that genetics plays in mito for testing and research. Check out the [Lily Foundation's page](#) for some great ideas for how to get involved via social media or local events. Join others in shining a green light in your porch, garden or living room on 20th Sep to help spark a conversation!

We have now fully launched the new LifeArc Centre for Rare Mitochondrial Diseases website! Take a look at the website here: [LifeArc Centre for Rare Mitochondrial Diseases website](#). The Centre is the first national mitochondrial disease platform, aiming to connect specialists, patients, scientists, knowledge and infrastructure. There is also now a [LinkedIn page](#) that you can follow where the latest news from the centre will be posted.

LifeArc Centre
Rare Mitochondrial Diseases

COMMUNICATING YOUR MEDICAL NEEDS

The Lily Foundation have advice about some great tools to help communicate your medical needs if you're living with mitochondrial disease.



Medical alert cards developed alongside the MitoCAMB team include emergency contact details for the team at Addenbrooke's Hospital. These cards are perfect to carry with you in a wallet!

'This is Me' Rare Patient Passport created by The Lily Foundation and CamRARE for your wallet or lanyard. These documents outline medical and care details for people with mitochondrial disease and can include medical specialist and emergency contact details. The passport can even be personalised to highlight the key information to know about you.

© Cambridge Rare Disease Network

Medical alert wristbands created by Mito Mission to help people with mitochondrial disease communicate their health needs. The wristbands include a QR code for the clinical guidelines developed by the specialist mitochondrial centre in Newcastle. They can be personalised to include emergency contact and personal information.

For more information about these medical alert tools, please visit [The Lily Foundation website!](#)

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RESEARCH STUDY NEWSFLASH

ORION STATUS: Open to recruitment

Sharing data generated from our studies is incredibly valuable and means the research can have a greater impact! We are engaged in some exciting collaborations for the ORION study, which involve sharing the images collected of the back of the eyes along with some de-identified clinical information including age, sex, diagnosis and vision history.

One of these partners is OcuSciences, a US-based medical device company, who have developed the OcuMet Beacon device that we use in the study to take photos of the back of the eyes.

This device is currently only approved for use in research. Sharing the images and data with OcuSciences will not only help the study team with their analysis but also help to improve the technology!

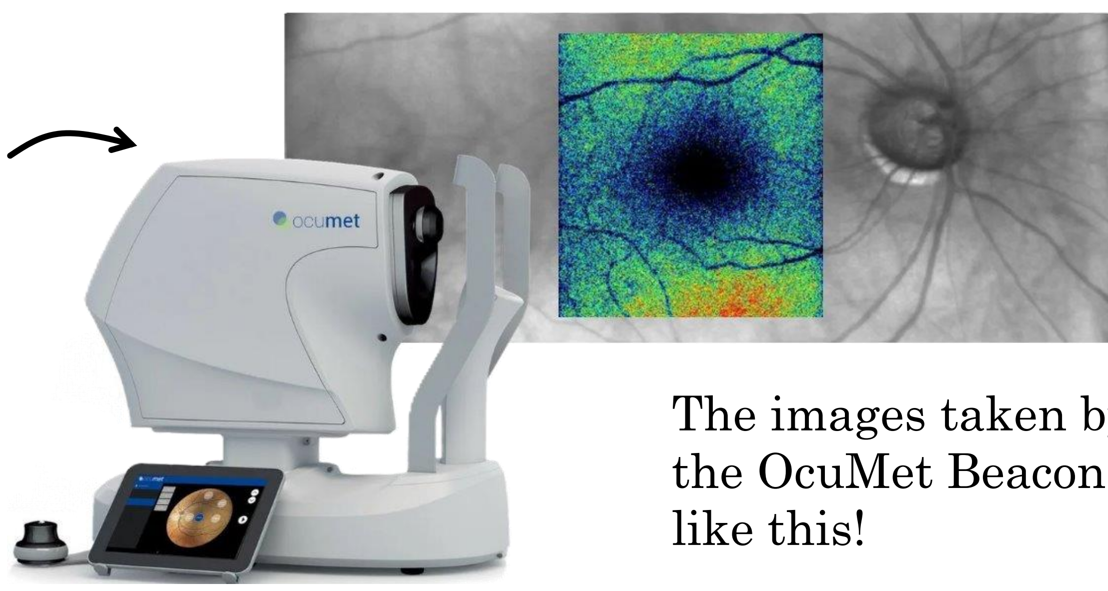

Image: Courtesy of OcuSciences

The images taken by the OcuMet Beacon look like this!

We have also partnered with Hong Kong University to perform a technique called Retinal Nerve Fibre Layer (RNFL) Optical Texture Analysis (ROTA) on the images collected from Optical Coherence Tomography (OCT). This is to help the study team analyse the nerves and identify changes in texture that may indicate defects or damage.

We may also make the study datasets available to researchers via a public online database. This is known as 'open data'. All data would be de-identified before they are uploaded to an open data platform. Sharing data in this way helps maximise the benefit of conducting research and allows other researchers to verify results and avoid duplicating projects.

Thank you to all the patients who are part of this vital research into inherited optic neuropathies! If you have any questions about the way that your data is used or shared, please do not hesitate to be in touch with the study team add-tr.mitoteam@nhs.net.

If you are interested in taking part, please visit the Website link for more information and application form.

OSPREY STATUS: Open to recruitment

A trial to test whether the drug STK-002 is safe and well tolerated in people with Autosomal Dominant Optic Atrophy (ADOA) is now open! This trial is open to people aged 18-54 who have a heterozygous optic atrophy type 1 (*OPA1*) gene variant and meet additional eligibility criteria. This study is sponsored by Stoke Therapeutics.

Participation in this trial will last approximately 1 year, during which there will be 10 in-person visits and 1 telephone call. These study visits will include eye exams/visual assessments, questionnaires, and blood samples. The medication is injected into the white of the eye with a small fine needle.

Check out the Website link for more information and application form.

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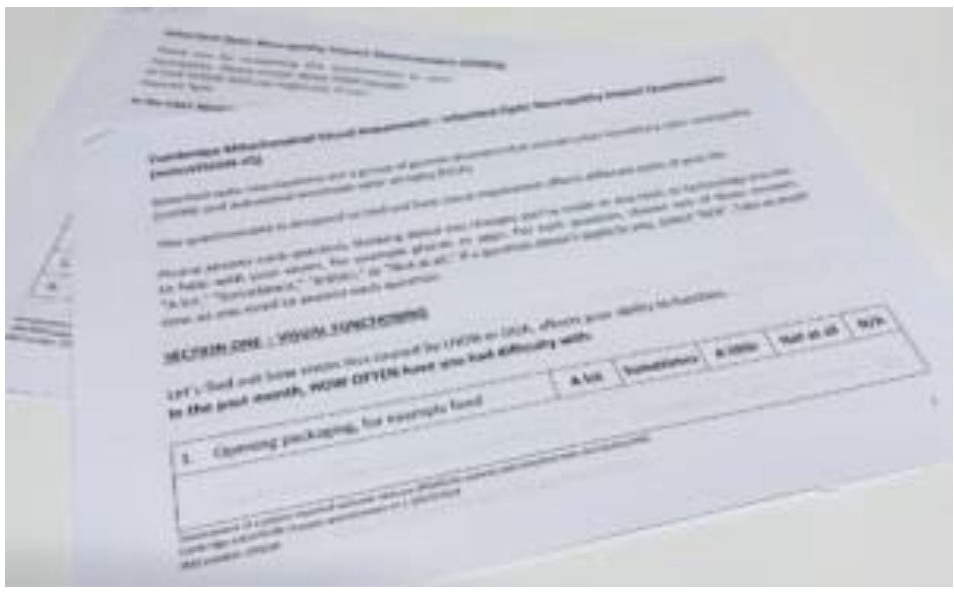
RESEARCH STUDY NEWSFLASH 2

ION PROM   STATUS: Open to recruitment

With the input of people affected by ADOA and LHON in the United Kingdom, we have developed a simple multi-choice questionnaire that assesses the impact of inherited optic neuropathies on visual functioning, social functioning, and mental wellbeing.



We are looking for people with a genetically confirmed diagnosis of Leber Hereditary Optic Neuropathy (LHON) or Autosomal Dominant Optic Atrophy (ADOA), aged 16 years and older and with symptoms of their condition to help test this questionnaire. The questionnaire is anonymous and takes around 15 minutes to complete.

The questionnaire can be found here.

Thank you to everyone who has completed it already, online or in clinic.

THANK YOU

FALCON (ABLIVA) STATUS: Open to recruitment

WHAT IS THE TRIAL ABOUT?
This trial is investigating whether the medicine, **KL1333**, is effective in improving symptoms of fatigue and daily living in people with mitochondrial disease. The safety and appropriate dose level of the drug will be evaluated. Wave 1 of the study involving 40 patients globally was completed last year and the interim analysis showed positive results. Therefore, the study is continuing into wave 2 with the goal to randomise 180 total patients.

WHAT IS INVOLVED?
We will recruit patients with a diagnosis of primary mitochondrial disease (m.3243A>G MELAS/MIDD or single large scale mitochondrial (mt)DNA deletions) who will be randomised to receive either KL1333 or placebo. The trial will involve at least 8 visits to Cambridge over a 16-month period. Telephone visits will also be conducted. Study visits will involve completing a number of self-reported questionnaires, blood samples, and a range of physiological and clinical assessments.

WHO IS RUNNING THIS TRIAL?
This is a global trial sponsored by Abliva AB.

ABLIVA

FALCON

If you would like to find out more about the trial, please get in touch with us using the details on the last page of this newsletter. We will also be contacting patients who could be screened.

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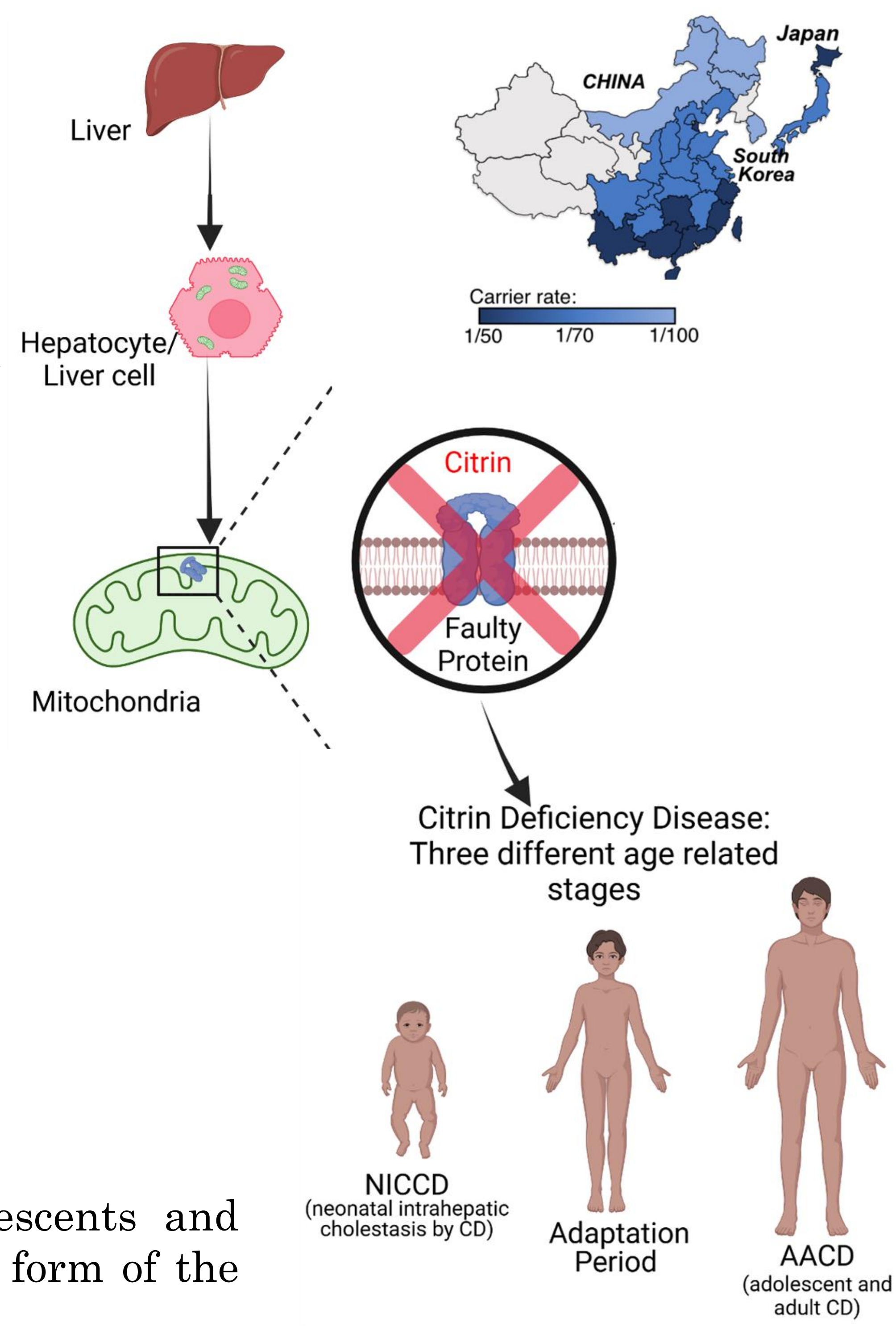
VIEW FROM THE LAB

Citrin Deficiency is a mitochondrial disease that many people in the UK have not heard about. It is a disease that is most common in Asia, but may affect people all across the world, of all ethnic backgrounds. In many patients with Citrin Deficiency, the disease often goes undiagnosed for a very long time.

Citrin Deficiency will affect people in different ways, depending on their age. The first stage of the disease occurs in newborns and is called NICCD (or 'Neonatal Intrahepatic Cholestasis caused by Citrin Deficiency'). Newborns with NICCD will have problems with their liver, causing jaundice (yellowing of the skin), and high levels of amino acids (a type of molecule that our cells use to make proteins) in their blood. Luckily, most babies improve after their first year of life and then go through a milder stage, called the adaptation period. During this milder stage, as they grow up, children will appear mostly healthy, but develop strong food preferences. They will typically avoid sugars and prefer eating food that contains more fats and proteins. Some children may also experience fatigue, stomach problems, and growth problems, and they may have low blood sugars.



As children grow up and become adolescents and adults, some may develop a more severe form of the disease, called AACD ('Adolescent and Adult Citrin Deficiency'). This severe stage involves symptoms affecting the liver and the brain, leading to mental health problems, behavioural changes, seizures and eventually loss of consciousness and death. The onset of AACD can be gradual or sudden, caused by triggers such as surgery, infections, alcohol consumption or eating lots of sugar. There is currently no cure for Citrin Deficiency, so doctors manage the condition with careful monitoring, dietary advice, and food supplements.

Citrin Deficiency is caused by problems with a protein called 'Citrin'. This protein is found in the mitochondria, which is why Citrin Deficiency is also considered a mitochondrial disease. Citrin is important for the liver's ability to process our body's waste products and toxins. It does so by transporting nutrients in and out of the mitochondria. When Citrin doesn't work properly or is absent, it causes issues with the liver mitochondria, leading to the problems and symptoms described earlier.

Very little is known about how Citrin really works. Professor Edmund Kunji and his team at the MRC Mitochondrial Biology Unit, University of Cambridge, are studying how exactly Citrin transports nutrients in and out of the mitochondria, and how its absence or malfunction leads to Citrin Deficiency. Eventually, this will lead to the development of treatments for this disease. The Kunji group works very closely together with the Citrin Foundation, a non-profit organization focused on tackling Citrin deficiency worldwide.

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MitoCAMB TEAM UPDATES

We have welcomed several new people to the MitoCAMB team over the last few months! Rithika, Nisha and Kanav are excited to meet you in clinic or during research visits, and you can read more information about them on the Our Staff page on our website!

Rithika Bose

Nisha Nixon

Raoul Kanav Khanna

THE LILY FOUNDATION ADULT WEEKEND

Some of the MitoCAMB team attended The Lily Foundation Adult Support Weekend 16th-17th August 2025! The weekend offered a unique opportunity for us to hear from a large number of patients and families directly impacted by mitochondrial disease. The Lily Foundation also informed everyone about the LifeArc Centre, challenging some of the key misconceptions about research, and inviting participants to design their perfect clinical trial. One of the key take aways was that people feel that symptoms are important to investigate, many of which focus on improving Quality of Life and Activities of Daily Living, rather than necessarily purely curative treatments.

CONTACT US

Thank you for your continuing participation in our research programme. If you have any queries relating to research studies that you have taken part in, or if you would like further information on any of our studies, please contact the team on:

add-tr.mitoteam@nhs.net 01223 335106 @mitocamb.bsky.social

https://www-neurosciences.medschl.cam.ac.uk/mitocamb/

For queries regarding routine NHS clinic appointments, please contact Katrina Dedman: cuh.mitocambclinic@nhs.net or 01223 216751