



MitoCAMB Patient Newsletter

Issue 15
September 2026



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WELCOME to a special edition of the **Cambridge Clinical Mitochondrial Research Group** patient newsletter in honour of World Mitochondrial Disease Week 2026!

You are receiving this because you have previously signed up to help with research into **mitochondrial disease**. 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 2026

14th – 20th September

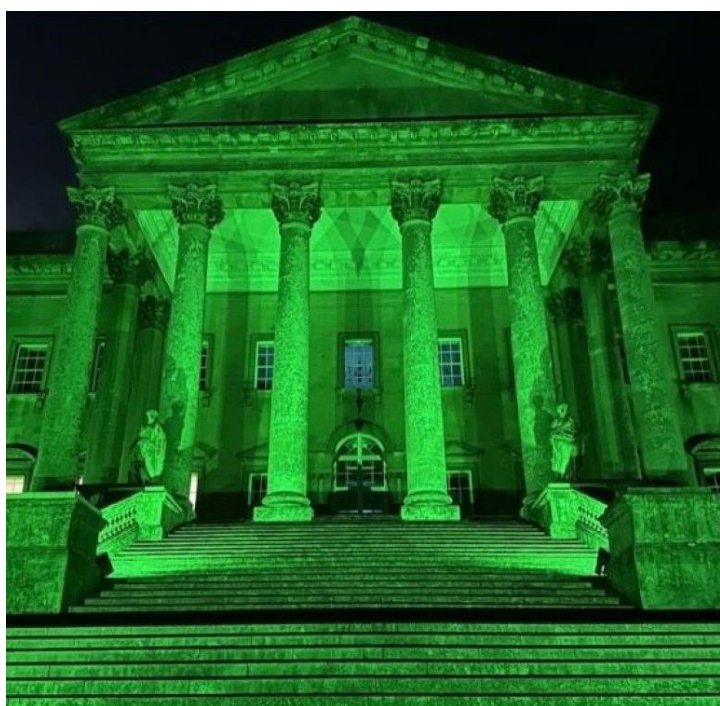
This year's campaign is **"Putting Mito on the Map"** which focuses on making mitochondrial disease more visible worldwide by amplifying patient voices, strengthening awareness, and connecting communities across borders.

The campaign features a new **Global Mito Map** - an interactive tool showcasing patient stories, organisations, research projects, events, and Light Up for Mito locations. As more pins are added, the map will grow into a powerful visual of the worldwide mito community. If you would like to add a pin, please visit: <https://mitopatients.org/add-your-pin/>



The Lily Foundation have lots of exciting ways you can [get involved in World Mitochondrial Disease Week](#):

- Update your profile pic on socials with their special [Twibbon](#)
- Repost mito-based facts and videos shared across their social channels during the week
- Host an awareness stand somewhere in your local area. The Lily Foundation will send out a [free pack](#) with everything needed to spread the word



On **Saturday 19th September**, landmarks around the world will light up green for mitochondrial disease awareness. Be part of **Light Up for Mito** by swapping a light bulb to green, adding green LED lights in your garden, or by asking your local monuments to light up green!

If you would like to approach a local building or landmark to light up green, the Lily Foundation have an email template to download and easily adapt on their [website](#).



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zoom ROOMS

This September, The Lily Foundation and [Rareminds](#) will be hosting two **Zoom Room Webinars**. These webinars are hosted by the Lily team and other healthcare professionals. They are designed to **offer advice and guidance** for patients and families affected by mitochondrial disease. There will be a 20-minute presentation followed by a 40-minute Q&A.

- How To Talk To Your Child About Mito - An online workshop for parents of children with mito- **Tuesday 22nd September 2026 7.00pm - 8.00pm**
- How To Talk About Mito- An online workshop for adults with mito- **Tuesday 29th September 2026 7.00pm - 8.00pm**

If you are interested in attending these webinars, please email katie@thelilyfoundation.org.uk to reserve your place!

To watch the previous zoom seminars, please visit: [Mitochondrial disease Zoom Room webinars: The Lily Foundation](#)

LILY FOUNDATION FAMILY SUPPORT WEEKENDS

This year, seven researchers from the Cambridge Clinical Mitochondrial Research Group attended the **Lily Foundation's Family Support weekend** (19th June-21st June 2026). This was a valuable opportunity to meet people affected by mitochondrial disease and share the team's research!

SAVE THE DATE: The dates for the next Lily Family Support weekend are **16th-18th July 2027**. Keep an eye out on the [Lily Foundation website](#) for more updates or if you would like to know more about the upcoming programmes.



MARATHON FOR MITO!

MitoCAMB's very own data manager, **Emma Harrison** will be taking on the 2026 Chicago Marathon in aid of The Lily Foundation. With less than a month to go and now going into the tapering phase of her first ever marathon training block, please see the following link if you would like to support Emma's fundraising efforts: [I'm running the Chicago Marathon to help The Lily Foundation beat Mito!](#)

NEW STUDY OPEN TO RECRUITMENT: STARS-MITO

Do you have a child with a mitochondrial aminoacyl-tRNA synthetase disease?

What is the goal of the study?

We are running a new study to find out whether giving **amino acids as a supplement** to patients with mitochondrial aminoacyl-tRNA synthetase diseases increases the availability of amino acids in the body and therefore improves mitochondrial function.

Who can take part?

Individuals (aged 5-18 years old) with a confirmed genetic diagnosis of a mitochondrial disease caused by changes in one of the genes **AARS2, EARS2 or DARS2**, and who experience symptoms of their disease.

What does the study involve?

- 9 visits (6 onsite & 3 telephone calls), across a period of up to 42 weeks (just under 10 months)
- 24 weeks of an amino acid supplement
- Completion of diaries for the duration of the study, to keep a record of symptoms, dietary intake, seizures and infections
- Physical, neurological and cognitive assessments, and clinical rating scales (used to look at how severe a person's condition is, their symptoms, and how their condition impacts their daily life).
- Blood and urine samples, physiological assessments, questionnaires about quality of life, and short scans using either x-rays or ultrasound.

The onsite study visits will be held at a dedicated clinical research facility at Cambridge University Hospitals and will last approx. 6-8 hours. Reasonable travel costs will be reimbursed.

For more information, please contact:

Telephone: 01223 331506

Email: add-tr.mitoteam@nhs.net

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UNDERSTANDING MITOCHONDRIAL NEUROGASTROINTESTINAL ENCEPHALOMYOPATHY (MNGIE)

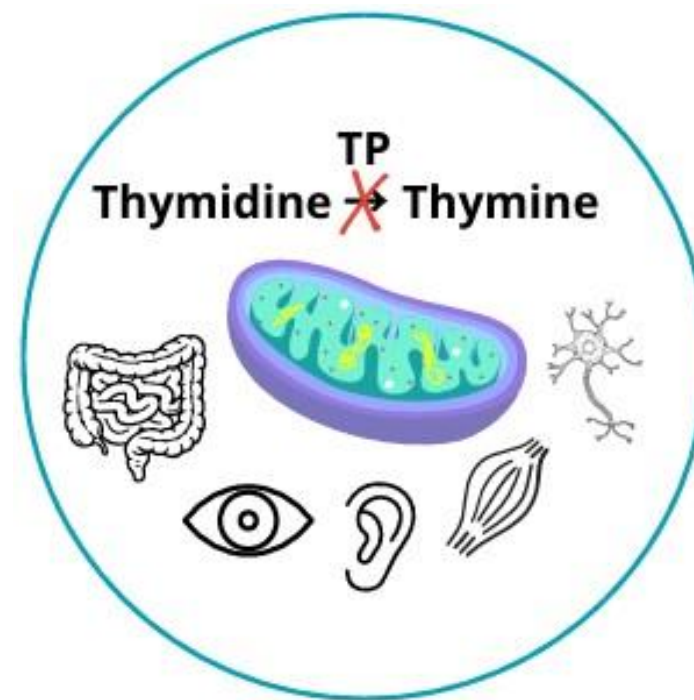
A new international research initiative to better understand the rare disease, **MNGIE** (**Mitochondrial Neurogastrointestinal Encephalomyopathy**), is looking for help from doctors, patients, and families. This research study aims to be the most detailed natural history study ever carried out for the rare mitochondrial disorder MNGIE.

This study is led by researchers from the **University of Cambridge** and the **University of Bologna**. It is funded by Pierrepont Therapeutics. The goal of the study is to better **understand MNGIE** and **guide future treatments**.

What is MNGIE?

MNGIE is a very rare inherited mitochondrial disease, caused by mutations in a gene called 'TYMP'. It affects only about 1 in every million people.

New therapies, including novel gene therapies and enzyme replacement strategies, are being developed by researchers and companies across the world. A better understanding of the course of the disease and how it affects patients is essential for guiding future clinical trials and improving patient care.



Why does this study matter?

Knowledge is still limited and each patient's data is very important. This study aims to collect clinical data from as many patients as possible from all over the world.

The research team hopes to:

- Better describe the **disease course** of people diagnosed with MNGIE.
- Identify the best **biomarkers** that measure how severely a patient is affected.
- Better understand how good the **current treatments** are and how to best **measure** this.
- Build a large network of doctors and researchers looking after people with MNGIE, to **exchange knowledge** and **improve care**.

Who can participate?

Doctors who care for or have cared for people who were diagnosed with MNGIE will be invited to complete a secure, anonymous online survey. Data will be collected from clinical records held by doctors and hospitals.

Patients and family members who wish for their data to be included should speak with their doctor, who can contact the study team directly. The study also welcomes information about patients who are deceased, and those who previously had treatments such as liver or haematopoietic stem cell transplantation.

Interested in learning more or taking part?

For more information or to participate, please contact:

add-tr.mitoteam@nhs.net. Alternatively, please visit our [website](#).



POLG SEMINARS

The Lily Foundation are running a series of seminars on current research on POLG led by University College London (UCL). POLG is a rare type of mitochondrial disease where cells cannot make enough energy to support organs. This is caused by changes in a gene called POLG.



To register your interest in POLG seminars organised by the Lily Foundation, please email Lindsay Wilson and Katie Waller. Please state your interest in “POLG SEMINARS”. You can email Lindsay and Katie at Lindsay.wilson@ucl.ac.uk; katie@thelilyfoundation.org.uk

SOON TO BE CLOSING TO RECRUITMENT: FALCON KL1333 STUDY (PHARMING)



WHAT IS THE TRIAL ABOUT?

This trial is investigating whether the medicine, **KL1333** (Napazimone), 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 is complete 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 Pharming Technologies.



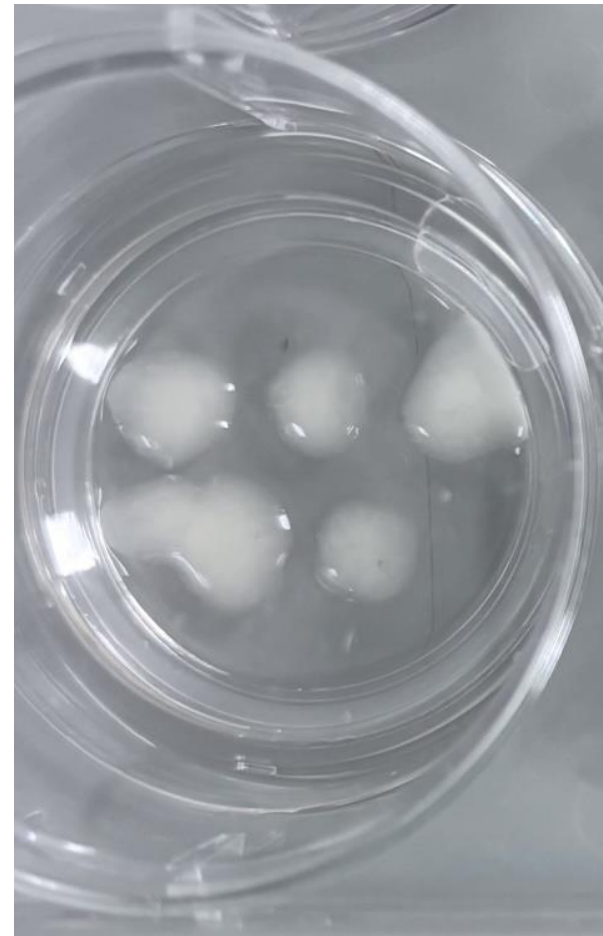
Recruitment to this trial will be **closing** at the **end of October 2026!** If you would like to find out more about the trial, please get in touch with us using the details on the last page of this newsletter.



VIEW FROM THE LAB

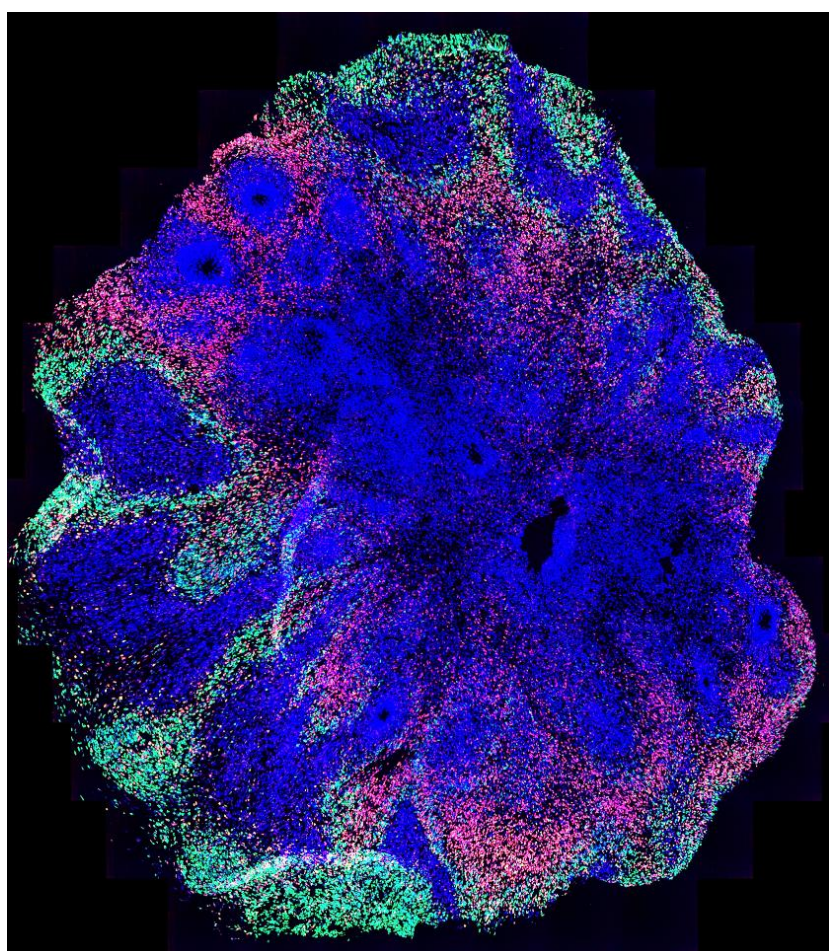
Using ‘mini brains’ to study mitochondrial disease

In a new study by Professor Rita Horvath’s team at the University of Cambridge, laboratory-grown **brain organoids**, or “mini brains,” were used to investigate how the common mitochondrial DNA mutation m.3243A>G affects the brain. This mutation is associated with **MELAS**, a mitochondrial disease that can cause seizures, stroke-like episodes, hearing loss, diabetes, and muscle problems.



The organoids are **small clusters of human brain cells** that are created from cells donated by people carrying the mutation. By generating organoids with low, medium, and high levels of mutated mitochondrial DNA, the researchers examined **how increasing mutation levels influence brain cell function**. They found that cells could adapt to low and moderate mutation levels by changing how they produce energy. However, at **high mutation levels** these mechanisms failed, and the **mitochondria could not function properly**, leading to energy shortages and cellular stress.

Importantly, **not all brain cells were equally affected**. The most vulnerable were nerve cells (neurons) that are involved in **long-distance communication**. These neurons have high energy demands due to their extensive connections. In organoids with high mutation levels, these neurons showed damage, stress, and increased cell death. Similar neuron loss was also observed in postmortem brain tissue from a person with MELAS, showing how relevant the organoids are at modelling human disease.



Although organoids cannot fully replicate the **complexity of the human brain**, the study provides valuable insight into how mitochondrial mutations can affect the brain and offers a **promising model** for developing future therapies.

If you are interested to find out more, you can read the full article about the study on the [Lily Foundation’s Research Zone](#)

Immunofluorescence image of a brain organoid, where the green and red show different types of nerve cells and the blue shows the nuclei (the control centres of the cells).



STAFF UPDATES

Nikoletta Nikolenko is a Postdoctoral Clinical Research Associate within the Department of Clinical Neurosciences at the University of Cambridge. Her role focuses on clinical and translational research, helping to translate scientific discoveries into new treatments for people living with rare mitochondrial disorders. Nikoletta is particularly interested in clinical trial development and delivery, biomarkers and outcome measures and in helping to build the evidence needed to bring promising new therapies into clinical trials.



Charlotte Russell recently joined the MitoCAMP research group as a Research Coordinator for the LifeArc Centre for Rare Mitochondrial Diseases. Charlotte holds a degree in Microbiology with an Industrial Placement from the University of Manchester and began her career as a Research Assistant at Queen Mary, University of London. She has since worked on several clinical trials, including leading patient and public involvement and engagement (PPIE) groups, establishing an inclusive environment to support meaningful participant contributions.

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

If you would like to donate a gift to support the research that we are doing, please follow this link: [Give online](#)

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