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MitoCAMB Patient Newsletter

Issue 14
February 2026

UNIVERSITY OF CAMBRIDGE

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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.

RARE DISEASE DAY 2026

SHARE YOUR COLOURS

Rare Disease Day is taking place this year on **28th February**. This global movement, which started in 2008, aims to encourage everyone, whether patient, professional or member of the public, to raise awareness and take action for the 300 million people worldwide living with a rare disease. You can get involved by spreading the word and sharing your experience on social media, taking part in local events, lighting up buildings and calling on policymakers.

CamRARE are organising an event on **27th February** at the Discovery Centre, AstraZeneca, Cambridge, where individuals and families from the rare community will share first-hand stories and experiences to highlight how rare conditions impact lifelong human experiences. There will also be reflections from individuals in healthcare, research, industry and support organisations.

cam RARE

If you are interested in attending this event, you can get tickets here: https://www.eventbrite.co.uk/e/living-rare-reflections-across-a-lifetime-tickets-1977502495196?aff=oddtdtcreator

THE LILY FOUNDATION GUIDE TO MITOCHONDRIAL DISEASE

The Lily Foundation has developed a *Guide to Mitochondrial Disease* which has been put together by patients, their families and medical experts. The guide offers information about mitochondrial disease, the science behind mitochondrial conditions, and guidance on finding support. The guide is aimed at people living with a mitochondrial disease, newly diagnosed patients, families and carers, and healthcare professionals.

If you are interested in getting a physical copy of the guide, please ask your doctor at your next clinic visit who will be able to provide you with one.

If you would like to access a digital version, please access this link: The Lily Foundation Guide to Mitochondrial Disease

Lily

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RESEARCH STUDY NEWSFLASH

STARS-MITO

In March 2026, we will be starting a new study, funded by Muscular Dystrophy UK, 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.

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 will be eligible to take part.

What will 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 the research team via telephone: 01223 331506 or email: add-tr.mitoteam@nhs.net

A RETROSPECTIVE NATURAL HISTORY STUDY OF SUBJECTS AFFECTED BY MNGIE

A study we currently have in set-up is *A retrospective natural history study of subjects affected by MNGIE.* MNGIE (Mitochondrial Neuro-Gastro-Intestinal Encephalomyopathy) is an ultra-rare mitochondrial disease, caused by mutations in the TYMP gene.

This natural history study aims to collect existing patient data from clinicians worldwide, to understand how the rare mitochondrial disease, MNGIE, affects individuals, how their disease progresses over time, and how current treatments affect them.

What does taking part involve?

Anonymous data will be collected in a secure REDCap database through an online questionnaire/survey. The survey should be completed by a clinician or their delegates. Completing the survey will take approximately 30 minutes to 1 hour.

If you or someone in your family, are affected by MNGIE, and you would like your clinical data to be included in this study, feel free to share this information with your treating clinician. They can then contact the study team, who will share a link to the survey with them. For more information, please visit our website!

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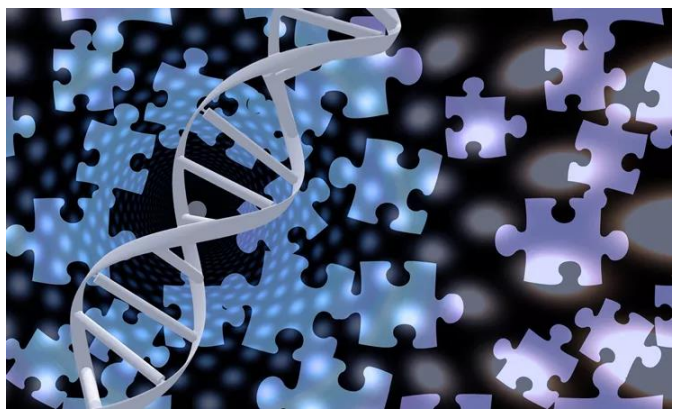
RESEARCH STUDY NEWSFLASH 2

BIORESOURCE LONG READ SEQUENCING PROJECT

Professor Patrick Chinnery, Professor Patrick Yu Wai Man and the MitoCAMB research team are supporting a project led by the NIHR BioResource, which is using a new sequencing technology called Long Read Sequencing (LRS).



Other methods of DNA sequencing generate many small sections of DNA which are pieced together like a big jigsaw to look for any changes. The LRS technology produces longer sections of DNA which makes putting the pieces of the jigsaw together much easier. The goal is to identify changes in the DNA that may have been missed in previous sequencing.

It is hoped that this study will lead to:

- Improved diagnosis for Rare Disease patients
- The development of new treatments
- Potential new research avenues

What would taking part involve?

- Signing a consent form
- Donating a blood sample (at Addenbrookes Hospital)
- Some clinical data would be collected about you

Please see their website for further information.

Recruitment to the BioResource Long Read Sequencing project will be closing in March 2026, so if you are interested in taking part in this project, or would like more information, please get in touch using the contact details at the end of this newsletter!

GENOTYPE AND PHENOTYPE IN INHERITED NEURODEGENERATIVE DISEASES – TEARS COLLECTION SUB-STUDY

A new sub-study of the Genotype and Phenotype in Inherited Neurodegenerative Diseases study, called the tears collection sub-study is recruiting patients with genetic conditions that affect the eye and cause vision loss, in particular, Dominant Optic Atrophy and Leber Hereditary Optic Neuropathy (LHON).

We are looking for biological markers (such as metabolites and fats) in tears and plasma (the liquid part of the blood) of patients affected by these diseases. The goal of this study is to improve diagnosis, gain a better understanding of why these diseases present differently among patients, and discover more about how vision loss happens.

Tears are collected using a micropipette which is a non-invasive method to collect a small volume of tears, usually in under 5 minutes. There is also the option to provide a fasting blood sample as part of this study.

You may be offered the tears collection sub-study in line with your clinic appointment or during other research study visits, such as for the ORION study (Outcome Research in Inherited Optic Neuropathies).

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THE LILY FOUNDATION - SUPPORT WEEKENDS

The Lily Foundation is hosting a range of support weekends this summer, offering those affected with mitochondrial disease the opportunity to come together, socialise, and access support and practical information about their conditions. Dates have now been confirmed for the family, young adult, and adult support weekends, and there is still a chance to sign up to the Adult and Young Adult support weekends.

Lily Adult Support Weekend 2026

Location: Chesford Grange Hotel, Warwick, 14th Aug 2026 - 16th Aug 2026

Invitations will be emailed out in spring 2026. Please note: you will need to sign up to the Lily database or you won't receive an invite – so be sure to **sign up here**!
For further information, please visit: Lily Adult Support Weekend 2026 – Save the Date - The Lily Foundation

Lily Young Adult Support Weekend 2026

Location: Devon, 25th Sep 2026- 27th Sep 2026

Invitations will be emailed out in 2026 You will need to sign up to the Lily database, or you won't receive an invite – so be sure to **sign up here**.
For further information, please visit: Lily Young Adult Support Weekend 2026 – Save the D… - The Lily Foundation

Lily Family Support Weekend 2026

Location: Chesford Grange Hotel, Warwick, 19th Jun 2026- 21st June 2026

Invites have now been sent out for this year's event, and all places have been allocated. If you would like to be added to the waiting list, or considered for a future event, please email lauren@thelilyfoundation.org.uk.
For further information, please visit: Lily Family Support Weekend 2026 - Save the Date - The Lily Foundation

THE LILY FOUNDATION- IMPACT COMMITTEE

IMPACT, **The Lily Foundation's Mitochondrial Patient Advisory Committee**, is a virtual committee of individuals affected by mitochondrial disease. By sharing your experiences, you can help shape research, clinical care and treatments that truly reflect the needs of the mito community.

The MitoCAMB team have worked closely with the IMPACT group to deliver focus groups for patients affected by mitochondrial disease to share their experiences of receiving feedback from research studies. We are always keen to collaborate with IMPACT members to help shape our research and future projects. To find out more about IMPACT and how to join, please visit: Make an IMPACT - The Lily Foundation.

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TEAM UPDATES

We are pleased to welcome new members to the MitoCAMB team! Aneta and Shamini are excited to meet you in clinic or during research visits, and you can read more information about them on the Our Staff page of our website!

Aneta Gordani

Shamini Saravanabavan

Aneta Gordani is currently a Clinical Research Fellow in the Cambridge Clinical Mitochondrial Research group within the Department of Clinical Neurosciences at the University of Cambridge, where she is developing her clinical research and trial experience.

Shamini Saravanabavan is a Neurology speciality registrar at Cambridge University Hospital. She is joining MitoCAMB as a LifeArc Clinical Fellow, identifying biomarkers to aid monitoring and treatment effectiveness.

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

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