

# MitoCAMB Patient Newsletter

Issue 8  
September  
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UNIVERSITY OF  
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## WELCOME

...to our new-look **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.

## TEAM NEWS

In the last few months we have said 'hello' and 'goodbye' to a number of team members...here is a round-up of MitoCAMB staff news.



In April we said goodbye to **Chloe Seikus**, our Research Assistant. Chloe has moved on to become an assistant psychologist in Brighton. She will work closely with patients living with a variety of neurological conditions. Chloe's message to everyone was: *"It has been an incredible experience to be a part of the MitoCAMB team for the past 2.5 years and to be involved in such novel and important research. I have learned so much from my brilliant colleagues, and, of course, the patients, whose generosity and support allow the research to happen."*

In June we welcomed **Elizabeth Ashby** as our new Clinical Research Coordinator. Elizabeth previously worked as a Healthcare Assistant during the COVID-19 pandemic, and gained valuable patient-facing and clinical experience. This, combined with her interest in research, led her to pursue clinical research and she joined the GI and Lymphoma team at The Royal Marsden (London) as a Research Assistant Practitioner in 2021. We are sure you will get to know her well as part of the MitoCAMB team!



We also welcome **Riddhima Gautam**, Senior Ophthalmic Technician to the MitoCAMB team. Riddhima worked as a Clinical Optometrist in India before graduating with her MSc in Investigative Ophthalmology and Vision Research from Glasgow Caledonian University. Riddhima is based at the Cambridge Clinical Vision Laboratory (CCVL) where she brings her clinical experience in the field of Optometry and Vision Sciences to all of the clinical assessments taking place at the Vision Lab.

**Dr Katherine Schon & Dr May Yung Tiet** are both taking up new posts, in Cambridge and Nottingham respectively. However, never fear; they will continue their research with the MitoCAMB team and our patients. In addition, **Dr Elena Conci** (Neurology) and **Dr Rahul Makam** (Ophthalmology) are looking forward to meeting our patients in clinics and at study visits soon!



You may have seen in the news that **Professor Patrick Chinnery** has been appointed as Executive Chair of the MRC. This is a huge role and our congratulations go to Professor Chinnery! He will continue to run both his laboratory and clinical research programmes at the University of Cambridge, and remains committed to understanding the role of mitochondria in human disease, and investigating ways to develop new treatments for mitochondrial disorders. For more information on this story, visit: [https://www.cam.ac.uk/research/news/leading-cambridge-neuroscientist-appointed-as-executive-chair-of-medical-research-council?utm\\_campaign=research&utm\\_medium=social&utm\\_source=twitter](https://www.cam.ac.uk/research/news/leading-cambridge-neuroscientist-appointed-as-executive-chair-of-medical-research-council?utm_campaign=research&utm_medium=social&utm_source=twitter)



## STUDY UPDATES NEWSFLASH

Here's a quick overview of what's been going on in the world of MitoCAMB in the last few months...

The **DefiNe** trial looking at deferiprone for the treatment of neuroferritinopathy passed its first regulatory hurdle. We still hope to open this trial in late 2023 - look out for more news in the coming months!

"**The LHON and Short Of It**" is a new podcast written and produced by Dr Benson Chen from MitoCAMB, co-hosted by Chloe Seikus (former MitoCAMB research assistant) and James Ferguson (UK LHON Society), with support from the University of Cambridge Public Engagement Starter Fund. Benson and his co-hosts explore what it is like to live with LHON, delve into the science of mito diseases, & discuss the research happening in Cambridge. Episodes will be released monthly on all podcast streaming platforms and on the MitoCAMB website.

The **PROSPAX** Natural History study for patients with SPG7 & ARSACS is in its final year of data collection. Thank you to everyone who has completed their visits so far, & we look forward to welcoming the remainder of our patients in the coming months. The PROSPAX consortium has a new website, including a 'for patients' section: <https://www.prospax.net/>

We would like to thank everyone who took part in the **Nicotinamide Riboside** study. This study is now **closed** and we are busy analysing all the data. We will be in touch with an update on our findings in due course...

### ORION

Recruitment to the **ORION** study has now begun, so you may be contacted by the team. The aim of this study is to understand how inherited optic neuropathies progress over a 3 year period. We will assess a number of your visual parameters and analyse how these change. The study involves 4-6 study visits over the 3 years. Using this information, we hope to improve our knowledge of the natural history of inherited optic neuropathies. This information will be used for future clinical trials, helping progress research towards a therapy for inherited optic neuropathies.

Dr Benson Chen has developed a new questionnaire for individuals affected by autosomal dominant optic atrophy (**ADOA**) & Leber hereditary optic neuropathy (**LHON**) to report their **quality of life** and experiences living with their condition. The draft questionnaire is ready to be tested, & can be completed online:

<https://www.redcap-ide.cam.org.uk/surveys/?s=PAJWHNL9JPX3FYYH>

If you are completing it on your phone or tablet, the team have built in accessibility functions that you can use. The questionnaire is anonymous and takes about 15 minutes to complete.

### MITOX study

Despite the challenges of working through the COVID-19 pandemic, this study is close to completion. We are expecting our final patient & matched healthy volunteer to participate in the next 2 months, when we will start analysing the data to answer the important question around the effects of giving high flow **oxygen** to patients with mito disease.



## VIEW FROM THE LAB



In a change to the usual scientific programming from the lab, in this issue we wish to congratulate **Dr Dylan Ryan** and **Dr Stephen Burr** from the **MRC-MBU** on both completing Zigzag - Sun's on the Run in aid of the **Lily Foundation**. Dylan ran his first ultramarathon, while Stephen ran 65km in 9 hours! Stephen is pictured here with our Lily Bear (who also joined Stephen on his run, for some moral support!). Great work!



**Pint of Science** is a worldwide science festival which brings researchers to your local pub/cafe/space to share their scientific discoveries with you.

Earlier in the year **Professor Patrick Chinnery** and **Alannah King** (PhD student, MRC-MBU Kunji group), participated in the “**Rare Body**” event. Alannah introduced "The Mighty Mito." The only organelle to become an internet meme, mitochondria have found fame as the “powerhouse of the cell”. But is that accurate? Alannah did some mitochondria-myth-busting about the mitochondria in our cells – and what happens when they are damaged.

Patrick spoke about "A Global Energy Crisis: how do rare mitochondrial diseases arise and how can we treat them?" Patrick and his research group have been unravelling how rare mito diseases come about and using these groundbreaking discoveries to forge a path towards better treatments. Perhaps, one day, we can even hope for a cure.



## WORLD MITOCHONDRIAL DISEASE WEEK

18<sup>th</sup> – 24<sup>th</sup> September 2023

The theme of World Mitochondrial Disease Week 2023 is **Fatigue**. To help raise awareness of mitochondrial disease, the Lily Foundation are selling their **Lily Bears**. The MitoCAMB team has its very own Lily Bear, which has already been running to raise money (see above), and which will be visiting various places in Cambridge...and perhaps further afield...to spread the word. Look out for Lily Bear adventures on our Twitter/X page: [@cam\\_mito](https://twitter.com/cam_mito)



To kick us off, here is our Lily Bear getting their blood taken in clinic. Many of our studies involve collecting blood samples for research. These are precious donations for which we are very grateful, and

our Lily Bear is keen to contribute!

You can get hold of your own bear here:

<https://www.thelilyfoundation.org.uk/product/merchandise/gifts/new-lily-teddy-bear-chocolate/>



## CLINICAL TRIAL IN THE NETHERLANDS

Generate your muscle study: Phase IIa clinical trial with patient's own muscle stem cells in m.3243A>G mutation carriers.

We are writing to let you know about an opportunity to participate in a Phase IIa clinical trial being run by our colleagues in the Netherlands.

Here, researchers from Maastricht University tell us more about the trial, which is actively recruiting patients [now](#).



“More than half of m.3243A>G mutation carriers become quickly fatigued and have muscle weakness, and at present no treatment is available for this. In the Maastricht UMC+ (the Netherlands) we have already been investigating for some years whether the administration of muscle stem cells can lead to an improvement in muscle function. To prevent rejection reactions, we want to use the patient's own muscle stem cells for this. In our laboratory we have shown that around half of the m.3243A>G mutation carriers have mutation-free stem cells. Last year we carried out a safety study in which the patient's own muscle stem cells were administered once in the lower leg of m.3243A>G-mutation carriers. However, it is clear that 3 administrations are necessary to achieve an effective dose.

The purpose of this phase IIa clinical trial is to investigate whether 3x administration of patient's own muscle stem cells in the arm leads to an improvement in muscle strength and a reduction in fatigue. This therapeutic administration is at present only carried out in a study context.”

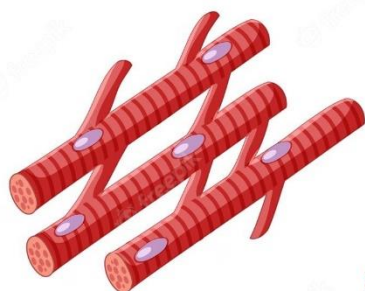
The study involves up to 9 visits to the Netherlands over a 9 month period. The study team will take a muscle biopsy from your upper arm – 3 times over the course of the study.

You will receive compensation if you participate in this study. €50 for visit 1 and €750 for participation in visits 2 to 9 (or part thereof if you stop earlier). You will also receive a reimbursement for your travel expenses.

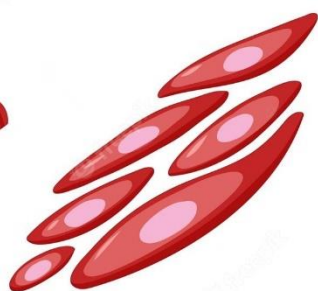
If you are an [m.3243A>G](#) mutation carrier and would like more information on this study, then please contact Florence van Tienen, +31 (0)43 3882918, [florence.vantienen@maastrichtuniversity.nl](mailto:florence.vantienen@maastrichtuniversity.nl)

For more information on the study visit the teams' website: [www.generateyourmuscle.com](http://www.generateyourmuscle.com) or visit The Lily Foundation page: <https://www.thelilyfoundation.org.uk/lily-research/uk-trials-research-studies/generate-your-muscle-gym-study/>

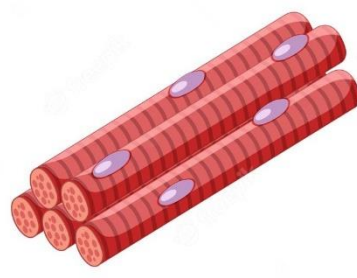
### TYPES OF MUSCLE CELLS



CARDIAC CELLS



SMOOTH CELLS



SKELETAL MUSCLE



## EMERGENCY CONTACT CARDS

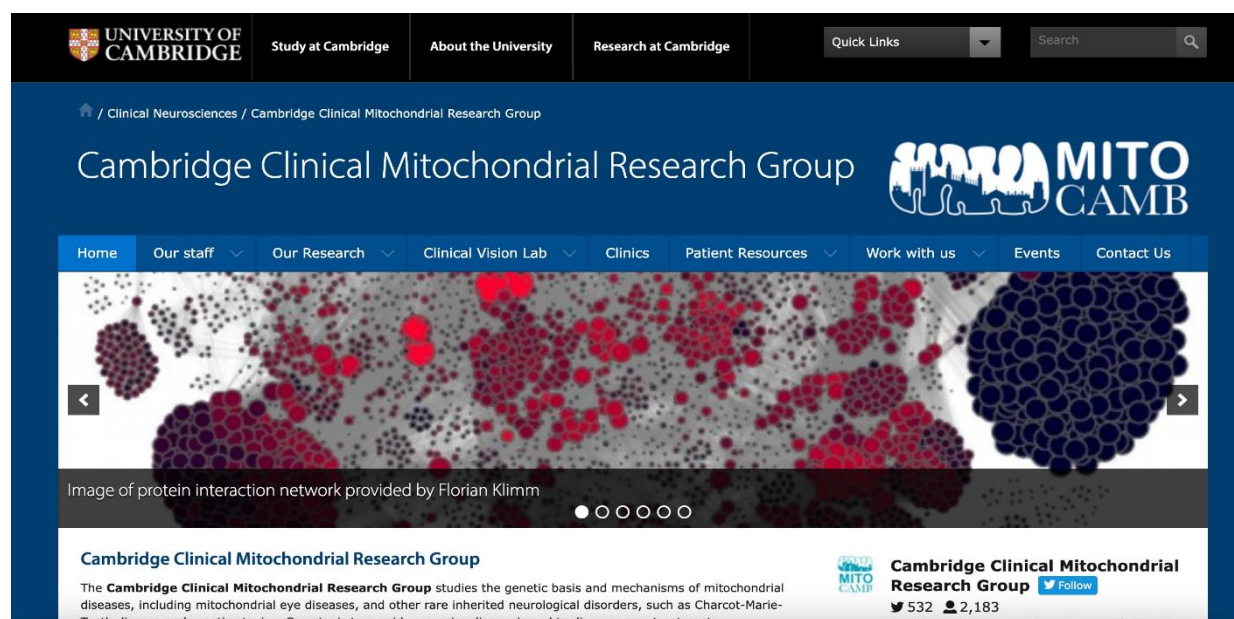


The Lily Foundation provides funding for Emergency contact/Medical Alert cards for mito patients. The MitoCAMB team now has a stock of these for all our mito patients under the care of Cambridge University Hospitals NHS Foundation Trust (CUH NHS FT).

We will be giving these out to relevant patients being seen in our clinics. If you think you should receive one, do ask your doctor at your clinic appointment, or get in touch with us using the contact details below.

## NEVER MISS AN ISSUE

Inbox heaving under the weight of so many emails? Remember reading something interesting in a previous issue of the newsletter, but you cannot remember where you saved it? Never fear! All of our patient newsletters get added to our [website](https://www-neurosciences.medschl.cam.ac.uk/mitocamb/information-for-our-patients/patient-newsletter/), so you can always go back to them. Just head to the following page to download them: <https://www-neurosciences.medschl.cam.ac.uk/mitocamb/information-for-our-patients/patient-newsletter/>



## 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](mailto:add-tr.mitoteam@nhs.net)  01223 335106  @cam\_mito

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

For queries regarding [routine NHS clinic appointments](#), please contact Katrina Dedman: [cuh.mitocambclinic@nhs.net](mailto:cuh.mitocambclinic@nhs.net) or 01223 216751

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