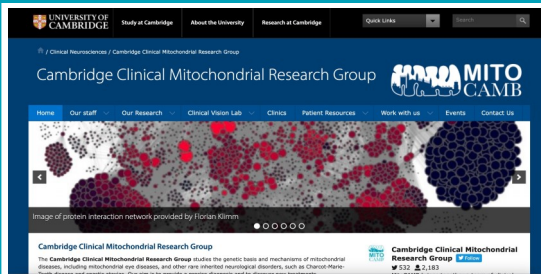


WELCOME!

Welcome to the latest issue of the **Cambridge Clinical Mitochondrial Research Group** patient newsletter. On behalf of the MitoCAMB team, we would like to wish you all a Happy New Year!

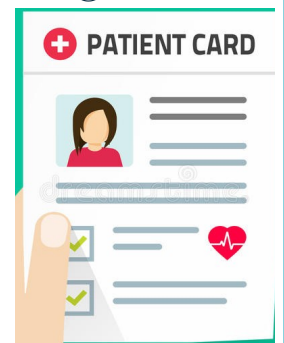
You are receiving this because you have previously signed up to help with research into **mitochondrial disease or neurodegenerative disorders**. Our aim is to provide you with the latest news from the research group, giving you more information on who we are, what we do, the science behind our work, and how we can support you, our patients. This issue aims to give you a peek into the exciting plans lined up for 2023...

PATIENT RESOURCES



Did you know that we have our very own website? We aim to 'refresh' this soon, so any thoughts or feedback you have are most welcome. You can visit by following the link below:
<https://www-neurosciences.medschl.cam.ac.uk/mitocamb/>
We also have a new email address: add-tr.mitoteam@nhs.net

Our clinical team have taken your feedback on-board and we plan to develop **patient cards** to improve your care. These cards will provide a summary of your diagnosis, with an aim to increase awareness and improve communication with other medical health professionals. Any suggestions on what you would find useful or informative are very welcome. Do just get in touch with us.



The Lily UK Mito Registry

The **Lily UK Mito Patient Registry** is a powerful online tool that allows people with a diagnosis of mitochondrial disease to log details about their condition and how it affects their daily life. That information can then be used to help direct research, improve understanding about mitochondrial disease and facilitate patient access to clinical trials.



Patients have full control over how their information is used. They can update it over time and give valuable insight into how a specific disease affects them. Users can also opt in to be contacted about any clinical trials or studies relevant to them...and it only takes 20 minutes to register!



To sign up, or to find out more information, please follow the link below:
<https://www.thelilyfoundation.org.uk/find-support/UK-mito-patient-registry/>



FOCUS ON...

In this issue of our newsletter, we would like to share with you information on some new **clinical trials** that are about to open to recruitment in Cambridge, and which you might be contacted about...

Neuroferritinopathy (DefINE) trial

WHAT IS IT ABOUT?

Neuroferritinopathy is a disorder in which **iron** gradually builds up in the brain. Certain brain regions that help control movement are particularly affected. The aim of the trial will be to find out whether **deferiprone** (a drug that removes excess iron in the body) is an effective and safe treatment for neuroferritinopathy.



WHAT IS INVOLVED?

Our trial is looking to recruit 40 patients with neuroferritinopathy, who will be randomised to receive either deferiprone or placebo. The trial will involve 4 visits to Addenbrooke's Hospital in Cambridge over a 12-month period. Study visits will involve MRI scans, clinical assessments and completing a medication diary. Fortnightly safety blood samples will be collected (at home/a local hub) and a member of staff will be in contact by telephone on a monthly basis.



WHEN WILL THE TRIAL OPEN?

The trial will hopefully be opening by the middle of 2023, but we will be in touch in advance to discuss recruitment!

KL1333 FALCON trial



WHAT IS IT 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.

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?

The sponsor for this trial is **Abliva AB**. Up to 50 sites in 12 countries worldwide are taking part. 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 eligible patients in the next few months.





View from the lab...

As well as our clinical work, we also have a strong **laboratory programme**, based in the MRC Mitochondrial Biology Unit (MBU), and the John van Geest Centre for Brain Repair. We thought we would share with you what our scientists got up to at **RAREfest 2022...**



Image: MBU scientists at RAREfest 2022

On Saturday, 26 November 2022 MBU scientists participated in **RAREfest** - an award-winning festival that is as **UNIQUE** as the people it champions. Featuring the brightest minds in research, innovators of life-changing technologies, pioneers in treatments, and those living with rare conditions.

The Guildhall was an excellent venue and we took hands-on, interactive activities showcasing the MBU’s research on mitochondria, specifically the **mtZFN ‘therapy’, through our “Destroying Mutant Mitochondrial DNA” game** - aimed at destroying mutant (bad) mitochondrial DNA by fishing it out of the mitochondrion and placing it in the mtDNA dustbin.

The venue was very busy - there were 25 scientific, technological, medical and creative hands-on exhibitions and, as well as explaining the MBU's research, we enjoyed taking the time to talk to familiar faces and make new connections. This was our first appearance at this event and we highly recommend it to all adults and children, both for education and entertainment – we very much look forward to future RAREfests!

You can watch highlights on YouTube here:

<https://www.youtube.com/watch?v=49D6CW649Xo>

https://youtube.com/playlist?list=PL1_lk1u_yLUZaudJrCifkaW_311_FKXOQ

Destroying mutant mitochondrial DNA!

Can we destroy the bad DNA, leaving only the good DNA?
We have made a chemical, called mtZFN, that can destroy only the mutated (bad) DNA, leaving the normal (good) DNA behind. The good DNA can then copy itself, and the mitochondria can function as normal!

○ Mutant mtDNA ■ Compromised OxPhos
○ Normal mtDNA ■ Normal OxPhos

The challenge

- How fast can you remove the bad DNA (textured balls) and save the mitochondrion?
- Put the bad DNA in the DNA dustbin!
- Remember to leave the good DNA (smooth balls); this is needed to allow the mitochondrion to work properly!



Leigh Syndrome Global Registry



CURE MITO
FOUNDATION

Do you, or someone you know, have a diagnosis of Leigh Syndrome? The Cure Mito Foundation has partnered with Sanford CoRDS to create the **Leigh Syndrome Global Registry**. The registry aims to bring patients, families and researchers together to gain a better understanding of Leigh Syndrome.

Once enrolled, participants complete questionnaires about their experience of living with Leigh Syndrome. Participants have control over their data and can update this at any time to provide insight into the course of Leigh Syndrome.

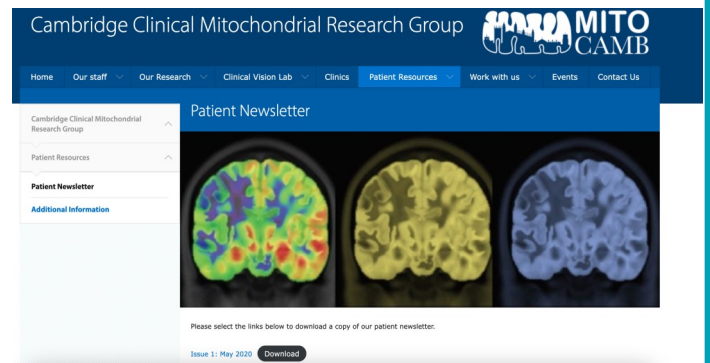
To sign up, or to find out more information, please visit the website:
<https://www.curemito.org/leighsyndromeregistry>



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**, 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



01223 335106



@cam_mito



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

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

