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MitoCAMB Patient Newsletter

Issue 11
December
2024

UNIVERSITY OF CAMBRIDGE

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.

MitoCAMB TEAM UPDATES

In July, we welcomed Charlotte Addy as our Junior Clinical Research Coordinator. Charlotte graduated with a BSc in Neuroscience from the University of Warwick in 2024. She previously worked on a research project, within the Centre for Integrative Neuroscience and Neurodynamics at the University of Reading, to improve understanding of pain in breast cancer patients. She is looking forward to meeting patients at clinics and visits!

Dr Ignazio Arena is a visiting clinical research fellow from Italy. He graduated from the University of Messina and is now in his last year of his neurology resident program, also in Messina. Since joining the MitoCAMB team in April 2024, Ignazio has brought his clinical experience to our trials, and is also involved in developing novel clinical research projects with the team.

Dr Schon has been part of MitoCAMB for the last five years. She did her PhD in mitochondrial disorders, supervised by Patrick Chinnery and Rita Horvath. She has recently completed her specialty training in Clinical Genetics. She is starting a new job as Assistant Professor of Human Genetics and Honorary Consultant Clinical Geneticist in Cambridge. Dr Schon will continue to see patients in the Friday afternoon clinic as part of the MitoCAMB team. Her appointments will be booked through Clinical Genetics. If you would like to discuss any genetic aspects of your condition, please ask the team to refer you to see Dr Schon. Examples include discussing the implications for other family members or reproductive choices.

RAREfest ROUNDUP

RAREfest24, organised by the Cambridge Rare Disease Network, took place on 22nd-23rd November. There were a series of inspiring talks and stories, exciting exhibits (including one from the MRC-MBU lab team!), and some amazing research-inspired art! Check out their website for more information about the talks and exhibits that took place: https://www.camraredisease.org/rarefest24/

camRARE
Cambridge Rare Disease Network

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RESEARCH STUDY NEWSFLASH

LifeArc

DefINe

A trial to test the use of deferiprone in people with neuroferritinopathy is recruiting participants! We are currently contacting patients to invite them to take part, so if you haven't heard from us yet, please don't worry, we will be in touch.

The key facts are:

- The DefINe trial is comparing two groups: 1) a group who receive deferiprone, and 2) a group who will receive a placebo, or 'dummy' drug. Participants will be randomly allocated to one of the two groups and they will have a 50% chance of receiving deferiprone.
- Overall participation in the trial will last for 13 months, including 4 in-person visits at Addenbrooke's Hospital in Cambridge, 3 MRI scans, and monthly telephone calls.
- Travel expenses will be reimbursed.

So far during the trial, only people with symptomatic neuroferritinopathy could take part. An amendment to the trial has been approved, allowing people without any symptoms to take part. We will be contacting people about this in the new year.

Please spread the word about recruitment to this important trial. It is vital that we get enough participants to properly assess the effect of this drug. The research cannot happen without you! Encourage family and friends to get in touch using the contact details at the end of this newsletter if they would like more information.

LILY PRECISION DIAGNOSTICS PROJECT

A new project funded by the Lily Foundation has opened to try and get a diagnosis for patients thought to have mitochondrial disease. This is called the 'Lily Precision Diagnostics Project'.

Project aims:

- To use new scientific techniques that are not available on the NHS to identify changes in the DNA that cause Primary Mitochondrial Disorders (PMD).
- To find better ways to diagnose, prevent and treat these disorders so that families with PMDs can receive better counselling and management.

What would participating involve?

- Existing health information and samples and will be re-analysed by specialists in mitochondrial disease.
- If additional samples are needed, participants may be asked to attend a study visit in person.

If you would be interested to take part and would like to find out if you would be eligible, please get in touch with us.

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RESEARCH STUDY NEWSFLASH CONTINUED

TREAT-AT

We are running a study called Trial Readiness in Ataxia Telangiectasia (TREAT-AT) supported by the AT society. We are starting to invite further participants to take part in the study before the end of the year. Participants will be asked to attend up to 4 research visits over the 2 years. Some visits can be performed virtually or at home.

Why we are doing this study:

- Finding ways to track changes in the symptoms caused by a disease (e.g. using a brain scan or a blood test) is key in developing treatments
- This allows us to find the best ways to measure whether a patient is responding to a treatment.
- The information gathered from assessing the neurological progression of AT over 24 months will help to inform future clinical trials.

If you have been diagnosed with AT and would be interested to take part, we would be delighted to hear from you.

NuPower (SPIMD-301)

The NuPower Study (SPIMD-301) sponsored by Stealth BioTherapeutics Inc. has reached the start of the data analysis phase. The primary measure to determine the effect of the drug is the distance walked in the 6 minute walk test. We will be sure to update you when we hear any results from the study team!

KL1333 FALCON

The initial analysis from the first wave of recruitment to the KL1333 FALCON study has been positive, showing no concerns with regards to the safety of the medicine. Therefore, a second wave of recruitment will be starting soon! This trial is testing if the medicine KL1333 is safe and effective in improving symptoms of fatigue and myopathy (muscle weakness) in people with mitochondrial disease. For more information about the initial analysis, please see the press release from the Lily Foundation: Mitochondrial disease study shows positive interim results - News - The Lily Foundation

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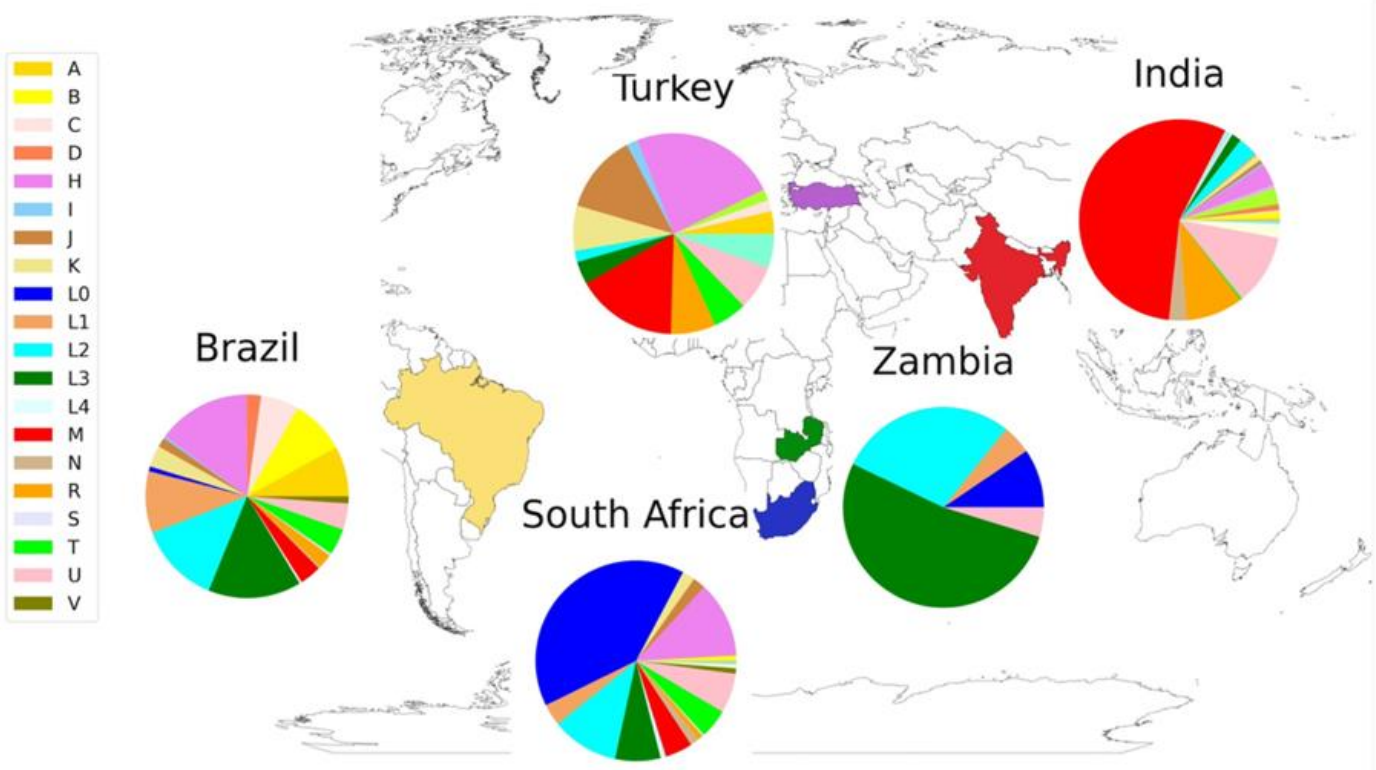
For those of you celebrating, we wish you a happy Christmas!

VIEW FROM THE LAB

In this issue, we would like to share some work which was led by Fei Gao, Katherine Schon and Rita Horvath as part of the International Centre for Genomic Medicine in Neuromuscular Disorders.

Mitochondrial DNA disorders in neuromuscular diseases in diverse populations

The team looked at the mitochondrial DNA in nearly 1000 people from South Africa, India, Brazil, Turkey and Zambia. The patients were recruited for research genetic testing because they have disorders of the muscles or nerves. The mitochondrial DNA analysis was done using the data from their main genetic test, called whole exome sequencing. The team found two new diagnoses of mitochondrial DNA disorders. One was a baby with Leigh syndrome (a mitochondrial disorder in children) and one in an adult with walking difficulties due to spasticity and peripheral neuropathy. Five people had a mitochondrial DNA change which can cause deafness if they are given specific antibiotics.



This study shows the value of analysing the mitochondrial DNA as well as the nuclear genes. They also looked at haplogroups in the different countries. Haplogroups are harmless mitochondrial DNA changes which reflect people's maternal ancestry (see map).

Ann Clin Transl Neurol, First published: 02 August 2024, DOI: (10.1002/acn3.52141)

https://onlinelibrary.wiley.com/doi/full/10.1002/acn3.52141

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: cuh.mitocambclinic@nhs.net or 01223 216751

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