

# MitoCAMB Patient Newsletter

Issue 12  
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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



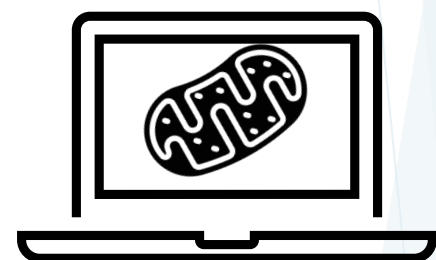
In April we welcomed [Denise Hatherly](#) as Clinical Research Centre Coordinator. Denise joined from Cambridge Cardiovascular on the Cambridge Biomedical Campus where she worked with academics from across the University and beyond to support the wider interdisciplinary research community working in cardiovascular medicine. Denise has a variety of skills from grant finance monitoring and management to website optimisation; general Comms & Events organising. Prior to joining the University two years ago Denise ran her own limited company which she sold in 2022.

## THE LILY FOUNDATION

[The Lily Foundation](#) have lots of exciting events, information and opportunities available!

### Zoom Rooms:

The Lily Foundation's Zoom Rooms are designed to educate and empower the mitochondrial disease community on a variety of topics. The first of these, '[Coping with Mitochondrial Disease](#)' took place on 18<sup>th</sup> March. You can watch the recording on this on [YouTube](#). These will be running quarterly, so keep an eye out on the Lily Foundation website for the next one!



### IMPACT:

If you are interested in guiding mitochondrial research and care, [IMPACT](#) is the Lily Foundation's [Mitochondrial Patient Advisory Committee](#). It is a virtual committee of people affected by mitochondrial disease. Sharing your experiences can help to shape research, clinical care and treatments to reflect the needs of the mito community. If you would like more information about IMPACT, please see the Lily Foundation website: [IMPACT](#).

### Research Zone:

Check out the latest news and updates in mitochondrial disease research on the newly launched [research zone](#). This includes articles about everything from summaries of studies to PPIE round-ups.



### Research Survey Opportunity:

The Lily Foundation are conducting a study to understand more about the experience of diagnosis in mitochondrial disease.

Click here to complete the survey!



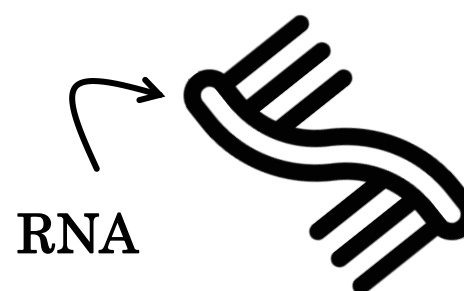
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## RESEARCH STUDY NEWSFLASH

### BioResource RNA Phenotyping Project

**STATUS: Open to Recruitment**



The BioResource RNA Phenotyping Project is **closing soon!** This is a study to look at the expression of genes in cells of rare disease patients. The project uses 'RNA sequencing', which is a technique that can be performed to give more information about which genes are turned on or off in a cell

#### Project aims:

- Improve diagnosis for patients with rare diseases
- Gain an insight into the causes of rare diseases
- Potential development of new treatments

#### What would taking part involve?

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

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!

## BioResource Long Read Sequencing Project

**STATUS: Open to Recruitment**

The BioResource have launched a project, which Professor Patrick Chinnery, Professor Patrick Yu Wai Man and the MitoCAMB research team are supporting, 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 in order 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. If the clinician believes you are eligible, you may be contacted to ask for your permission to pass your contact details to the NIHR BioResource team.



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## RESEARCH STUDY NEWSFLASH 2

### DefINe

#### STATUS: Open to Recruitment



A trial to test the use of deferiprone in people with neuroferritinopathy is [recruiting participants!](#)

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.

**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.

So far, 10 people are taking part in this trial. Take a look at this [BBC news article](#) about the trial!

### C12orf65

#### STATUS: Closed to Recruitment (Final Interview and Data Analysis)

Recruitment is now closed for this study looking into the [natural history](#) and [progression](#) of [Charcot-Marie-Tooth Disease Type 6](#) (CMT6) caused by mutations in MTRFR/ C12orf65.

The last interview for the study is due to take place in the coming weeks. The team will begin collating the data collected over the coming months and start analysis of this, with a view to produce a report or publication that will be shared with the community.

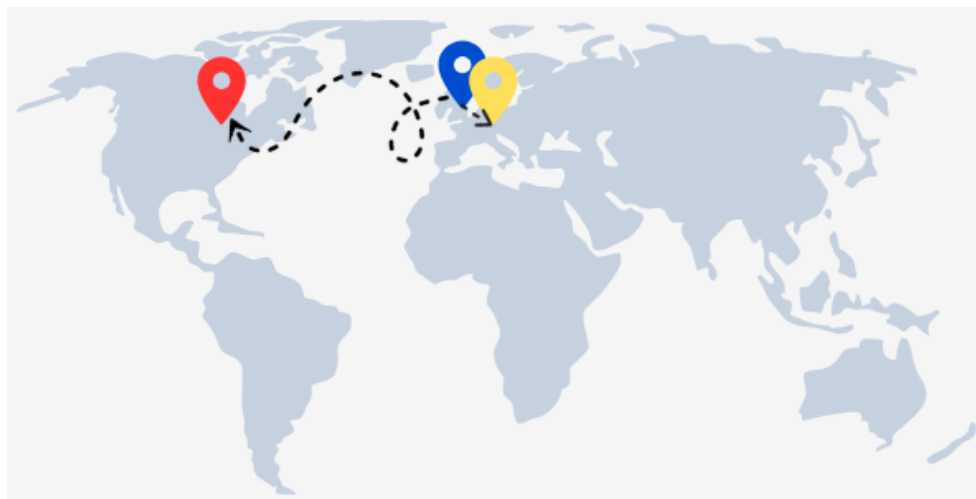
Details of the study can be found on our [website](#).

[Thank you](#) to everyone who participated in this critical study. Participation in studies like this help to progress research into CMT. You can find details of currently open opportunities on the CMT association website on the [Patients as Partners in Research page](#).

### NR

#### STATUS: Closed to Recruitment (Data Analysis)

The team are busy putting all the data together and [analysing the results](#).



The muscle biopsy samples before and after the NR treatment were split into parts. Some tests were analysed in Cambridge and others were sent to laboratories around the world. We have metabolomic data from Canada, proteomic data from Germany, complex activity levels from London and many other tests have been done in our laboratories in Cambridge.

We hope to be able to share the results soon! Many thanks for your patience in waiting for the final outcomes of the study.



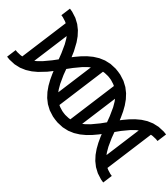


## LIFEARC CENTRE FOR RARE MITOCHONDRIAL DISEASES

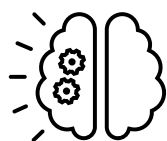
Professor Patrick Chinnery and the University of Cambridge are leading the LifeArc Centre for Rare Mitochondrial Diseases!

The centre aims to progress research into rare mitochondrial diseases through 3 main areas of research focus:

- Diagnosis



- Biomarkers



- New Treatments.






Rare  
Mitochondrial  
Diseases

The centre is funded by LifeArc and Muscular Dystrophy UK (MDUK) and it is a collaboration between the University of Cambridge, University College London, Newcastle University, in full partnership with The Lily Foundation. The Lily Foundation is leading patient and public involvement and engagement (PPIE) activity. For more information about the centre, please see the [LifeArc website](https://www.mitocamb.org.uk).

## 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  @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](mailto:cuh.mitocambclinic@nhs.net) or 01223 216751