

WELCOME!

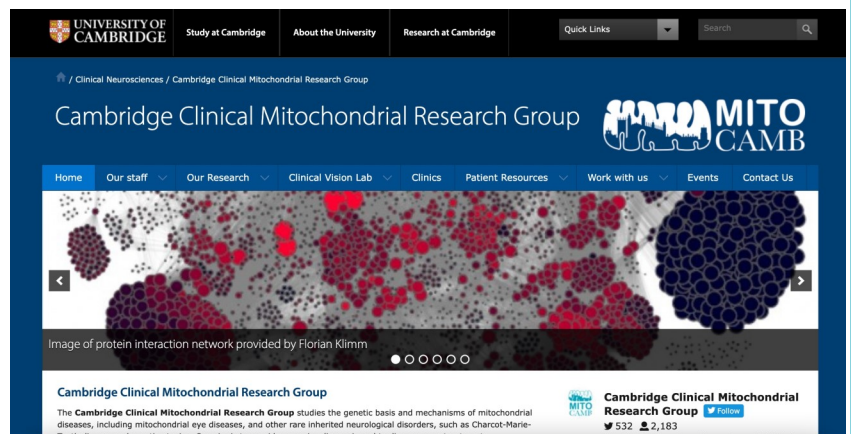
Welcome to another issue of the **Cambridge Clinical Mitochondrial Research Group** patient newsletter. You are receiving this because you have previously signed up to help with research into **mitochondrial disease or neurodegenerative disorders**. Our aim is to provide you with the latest news from 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.

In this issue, we will update you on some exciting developments from the team as well as highlighting some of our new research studies.

NEW MitoCAMB WEBSITE

We are pleased to announce that construction of our **new group website** is now complete, and the pages have gone **LIVE!** If you would like to find out more about our staff, our research and our clinics, then please visit:

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



All issues of this newsletter are also available as a pdf on the website.

Thank you to all our patients who contributed photos for the web pages—if you have any others you would like included, do email the team (contact details on the last page of this newsletter). In addition, if there is anything you would like to see featured on the website, let us know, as we are keen for our content to be up-to-date and relevant to you all.

SLEEP QUESTIONNAIRES

Back in the days of the first 2020 lockdown, we conducted a survey into **sleep disturbances** in patients with mitochondrial disease (via our **Genotype and Phenotype study**). We are continuing to analyse the responses from those original questionnaires. However, to make the data even more powerful, we have recently sent out a further set of questionnaires to some of our newer research recruits!



If you have recently received a **sleep questionnaire** in the post, your help is very much appreciated with this. Please do not hesitate to get in touch if you have lost your copy of the questionnaire, or if you need any help completing the questions.

Of course, as always, this research is **entirely voluntary**, so if you would rather not take part, do just let us know so we can update our records.



FOCUS ON...

In this issue of our newsletter, we would like to share with you information on two studies that have recently opened to recruitment, and which you might be contacted about to take part in...

NIHR BioResource - Rare Disease RNA project

NIHR | BioResource



WHAT IS THE NIHR BIORESOURCE?

The NIHR BioResource is a group of over **100,000 volunteers** who have already agreed to take part in **medical research**, with the aim of improving healthcare and the long-term **prevention and treatment of disease**. Volunteer participants who have already consented to the NIHR BioResource have agreed that they can be invited to participate in further research studies. They can choose whether or not they want to participate in each study they have been invited to.

WHAT IS THE NIHR 'RARE DISEASE' BIORESOURCE?

The 'NIHR BioResource – **Rare Diseases**' recruits **affected participants** and their **relatives** in more than 50 disease areas, including in immunity, **neuroscience**, haematology, rheumatology, and many more. The NIHR BioResource has used modern technologies to study the DNA of participants (whole-genome sequencing), and they have tied that genetic information to the clinical characteristics of the participants, which researchers call the 'phenotype'. It is hoped that this will help researchers to understand more about the **genetic causes of rare disease**.

WHAT IS THE RARE DISEASE RNA PHENOTYPING PROJECT?

This is a new study to look at the **expression of genes in cells** of Rare Disease patients. The aims of this study are:

- To **improve the diagnosis** of patients with Rare Diseases
- To gain an insight into what causes Rare Disease and the potential development of **new treatments**



Patients will attend an appointment to donate a sample of blood. Several **blood cell types** will be isolated in the laboratory from the whole blood collected. Then, something called '**RNA sequencing**' will be performed on each of the cell types isolated. RNA sequencing tells us **which genes are turned on in a cell** and at what times they are switched on or off. **The data will be available to research teams such as ours** to carry out further analysis in patients with rare disease. This may help us to diagnose or understand the mechanism or cause for a Rare Disease as a group.

WHAT WILL I BE ASKED TO DO?

If you are already signed up to the NIHR BioResource - Rare Diseases, **you may be contacted by the BioResource team** in the coming months to ask if you would be willing to **attend an appointment to donate a further blood sample** (50mls or 3 tablespoons) for this project. **Your travel costs will be covered** for attending this appointment. It is entirely **up to you** whether or not you choose to provide an additional sample.



WHAT IF I WANT TO TAKE PART BUT I AM NOT SIGNED UP TO THE NIHR BIORESOURCE?

If you would like to be considered for this RNA phenotyping project, **you need to be signed up to the NIHR BioResource - Rare Diseases first**. This involves signing a consent form, completing a short questionnaire about your health, and attending for a blood sample. Once you are signed up, you can then take part in the RNA recall study.

You can contact us at mitoteam@addenbrookes.nhs.uk if you require more information or would like to register as a member of the NIHR BioResource.



ION-PROM

ION-PROM: Development of a patient reported outcome measure (PROM) for inherited optic neuropathies (ION)

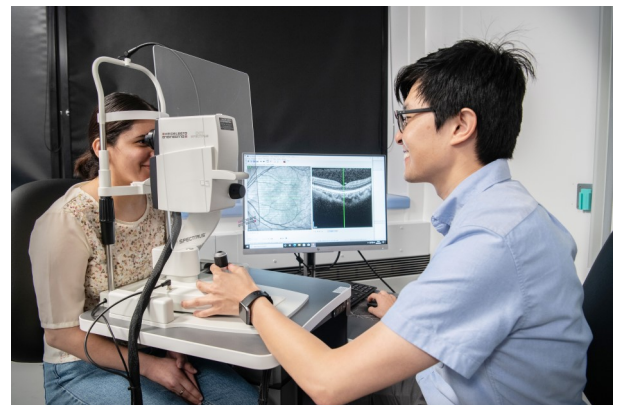
Dr Patrick Yu Wai Man from the University of Cambridge and Moorfields Eye Hospital (London) is leading a study to develop a new **questionnaire** for individuals affected by Dominant Optic Atrophy (**DOA**) and Leber's Hereditary Optic Neuropathy (**LHON**) to report their **quality of life** and **experiences** living with their condition. The questionnaire will be a useful tool in future clinical trials of both conditions and as part of regular clinical assessments to understand the impact of DOA and LHON.



We are looking for individuals to participate in **one-hour interviews** on Zoom to discuss their experiences of living with DOA or LHON and to **review the questions** we have developed. We are also looking for individuals to help **test the questionnaire** and make sure it is relevant to the LHON and DOA communities.

Individuals with a **genetic diagnosis** of DOA or LHON, **aged 16 years and older**, and with **symptoms** of their condition are invited to take part.

You may have already been sent information about this study via post or email. The research team is very happy to answer any queries you might have. Likewise, if you know of any individuals who might be eligible to take part, do share this information with them and encourage them to get in touch!



You can contact us at mitoteam@addenbrookes.nhs.uk if you require more information or would like to register your interest in the study.

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:



mitoteam@addenbrookes.nhs.uk



01223 335106



[@cam_mito](https://twitter.com/cam_mito)



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

