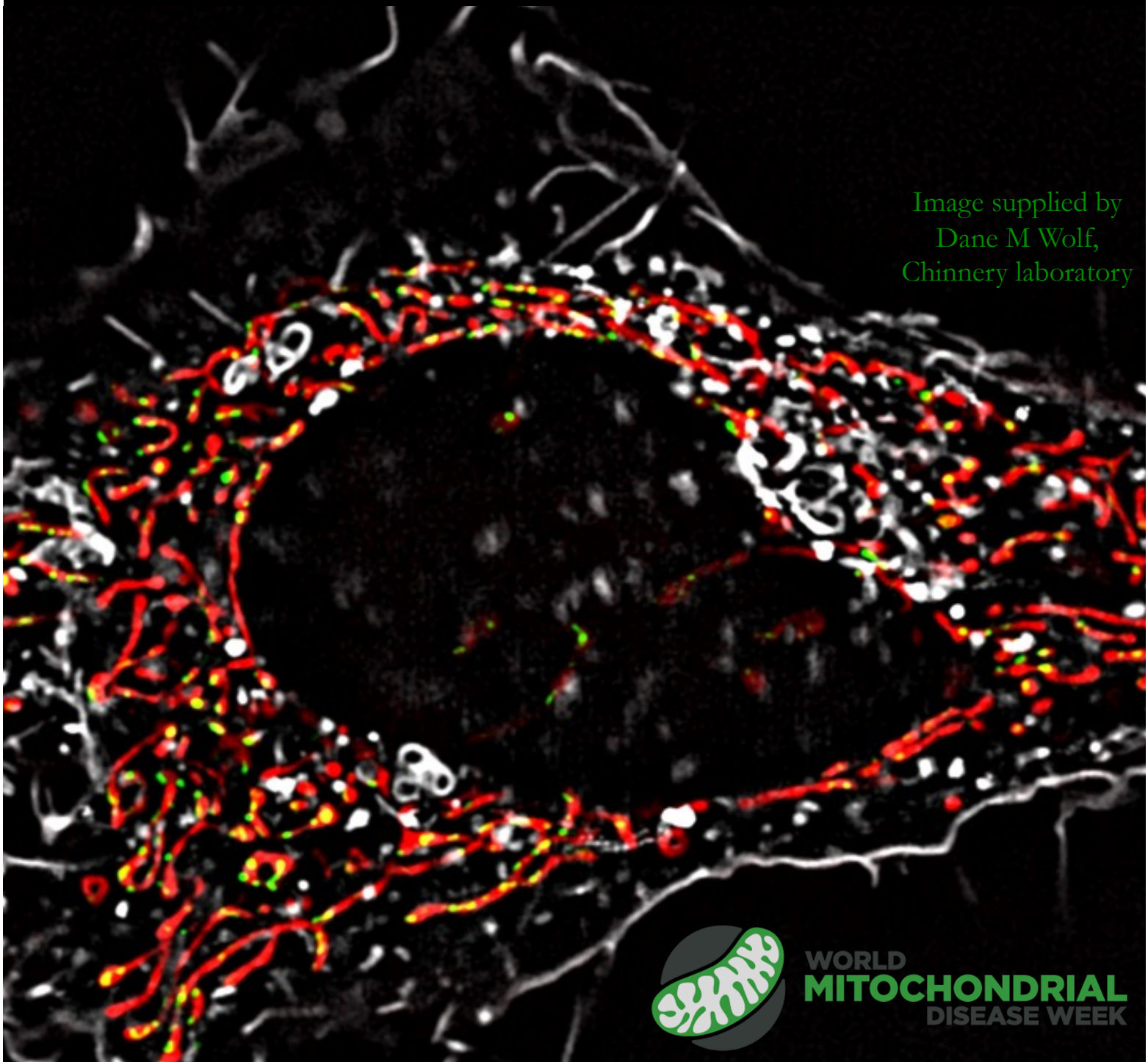
MITO CAMB

MitoCAMB Patient Newsletter

Issue 3, September 2021



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Dane M Wolf,
Chinnery laboratory

WORLD MITOCHONDRIAL DISEASE WEEK

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WELCOME!

Welcome to a special issue of the **Cambridge Clinical Mitochondrial Research Group** patient newsletter, marking **World Mitochondrial Disease Week**! 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 and from the wider mitochondrial disease research community in Cambridge, as well as highlighting some of our new and ongoing research studies that are focused specifically on mitochondrial disease.

WORLD MITOCHONDRIAL DISEASE WEEK

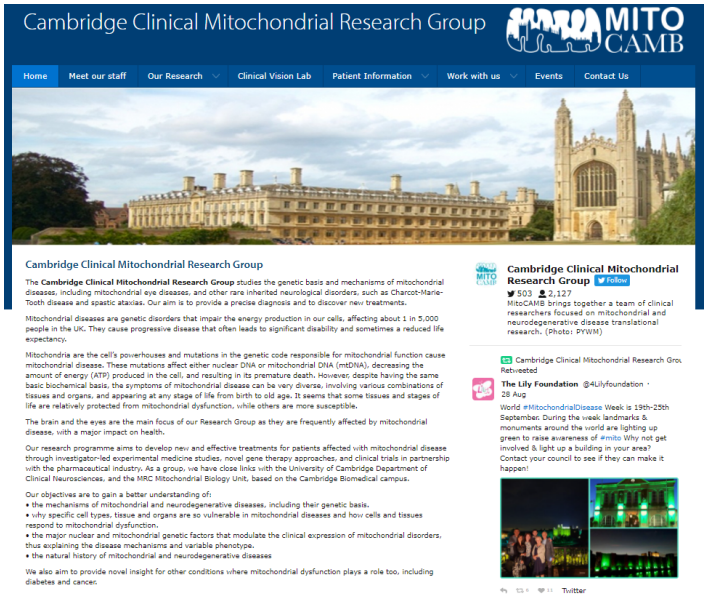
19th-25th September 2021 is World Mitochondrial Disease Week. Every year, monuments around the world are lit up green to raise awareness for mitochondrial disease (**#MitoAware**). 2021's **Light Up for Mito** will take place in participating countries on Saturday 25 September 2021.

Last year, the John van Geest Centre for Brain Repair on the Cambridge Biomedical Campus went green all week, thanks to technical support from Cambridge-based CFM Events Group. If you know of any buildings near you that will be lighting up green this year, then tag us on Twitter **@cam_mito** and we will highlight them on our page!

NEW MitoCAMB WEBSITE

As reported in our last issue, with a growing team of clinical researchers in Cambridge, focused on mitochondrial and neurodegenerative disease translational research, we wanted to encompass this group with a unified web presence.

Construction of our **group website** is almost complete. We hope the website will provide useful information for patients, research participants and other investigators, regarding our research, vacancies and opportunities.



Before formally launching the website, we would like to ask you, our patients, for your input, to check that the design and content is clear and easy to navigate. We may be in touch with a few of our patients to ask for your **feedback**. This is entirely voluntary, so if we contact you and you would rather not review the website, then do just let us know!

COMING SOON!

FOCUS ON...

COVID-19 has presented us all with many challenges over the past year. We are very pleased to have been able to re-start some of our studies, and even to open new ones! In this issue, we would like to focus on two **experimental medicine studies** that are open and actively recruiting patients with mitochondrial disease…

The role of Nicotinamide Riboside in mitochondrial biogenesis

What is the study about?

Some scientists think that **increasing the number of mitochondria** in your body (mitochondrial biogenesis) might be a good way to treat the symptoms of mitochondrial disease. Studies in mice have shown that a type of B-vitamin called **Nicotinamide Riboside (NR)** is able to increase the number of mitochondria, leading to increased energy and a reduction in the symptoms of mitochondrial disease. The aim of this study is to investigate if the same B vitamin, Nicotinamide Riboside, can increase **energy production and reduce symptoms** in humans with mitochondrial disease.

What is involved?

This study requires 6 visits to Addenbrooke's Hospital. We carry out a series of standard tests including a **muscle biopsy** and an **MRI scan** before asking participants to take a course of NR twice daily for 4 weeks. After 4 weeks of treatment we will carry out the same tests again to see if there have been any changes in response to the treatment.

Who can take part?

You may be eligible to take part if you have a diagnosis of:

1) Progressive external ophthalmoplegia **(PEO)** caused by a single deletion of mitochondrial DNA
2) Mitochondrial disease caused by the *m.3243A>G* or *m.3243A>T* mutation in mitochondrial DNA **(MELAS)**

Progress update:

10 participants have completed this study so far. **We would like to recruit 5 more participants to the study.**

The effects of oxygen in the context of mitochondrial dysfunction

What is the study about?

When people with mitochondrial disease develop acute illnesses, or have routine operations, they are given very **high oxygen** levels to breathe. Doctors do this because they think it will be helpful. However, recent studies in animals with faulty mitochondria suggest that this might make mitochondrial function worse in some situations. We want to know whether this happens in humans, so that we only use high oxygen treatments when it is **safe and helpful** to do so.

What is involved?

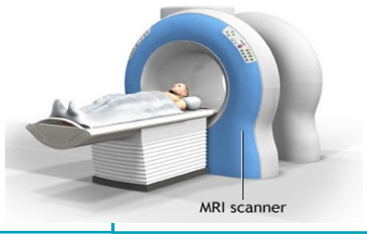
We will compare the effects of inhaling high level oxygen and regular room air oxygen over 2 consecutive days. Each day, participants will have an **MRI scan**, donate **blood samples** and undergo non-invasive blood oxygen measurement and **cognitive assessment**. We will study the effects of oxygen in patients with mitochondrial disease and compare this with people who do not have mitochondrial disease.

Who can take part?

You may be eligible to take part if you have a diagnosis of mitochondrial disease caused by the *m.3243A>G* or *m.3243A>T* mutation in mitochondrial DNA **(MELAS)**

Progress update:

This study opened earlier this year and 2 participants have already taken part. **We would like to recruit 8 more participants to the study.**



Can YOU help with our research?

If you or someone else you know are interested in taking part in either of these studies, please contact the research team using the details on the following page. We would be delighted to discuss our research with you further.

NEW CLINICAL VISION LAB

Patrick Yu Wai Man is an academic neuro-ophthalmologist based in Cambridge with a specialist research interest in mitochondrial eye diseases. In exciting news announced this month, he has just received an NIHR Moorfields Eye Charity Advanced Fellowship award in partnership with Moorfields Eye Charity, to undertake research into inherited optic neuropathies, which are genetic diseases that affect the optic nerve, causing progressive and irreversible blindness. A major focus of this ambitious research programme will be on the mitochondrial disease **Leber Hereditary Optic Neuropathy (LHON).**

This research programme will also make use of a new research facility: the **Cambridge Clinical Vision Lab (CCVL),** supported by the NIHR Cambridge Clinical Research Facility, the NIHR Cambridge Biomedical Research Centre and the Cambridge Eye Trust. The CCVL will use state of the art technologies to drive forward research into ocular and neurodegenerative diseases, with a particular focus on gene therapy and cell-based therapies.

These are very exciting developments for the research community in Cambridge and for our patients, and we hope to bring you more information regarding the CCVL (website currently under construction) and the wider research programme, in future issues of this newsletter.

To read more about Dr Yu Wai Man's research and the CCVL please see the following **press release: https://cambridgebrc.nihr.ac.uk/researchers-eye-up-new-gene-therapy-trial-that-could-reverse-hereditary-blindness/**

CAMBRIDGE CLINICAL VISION LABORATORY

LIKE A CHALLENGE?

The Lily Foundation are running their virtual **"Around the World for Mito"** event which kicks off on **Sunday 19th September**. Their aim is to complete 24,901 miles – the circumference of the Earth! You can contribute as many miles as you like to help them circle the globe. Ask others to sponsor your challenge, and you'll be making a difference with every step. Sign up via their website. Our Clinical Fellow, **Dr May Yung Tiet** is a keen runner, and will be taking part!

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

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