Issue 1

MitoNews UK

Specialised Services

Spring 2011

Welcome.....

Welcome to the first issue of MitoNews UK, a twice yearly newsletter for patients with mitochondrial disease. This issue gives you information on the Mitochondrial Disease Patient Cohort, and how to get involved, if you haven't already. Look out for recruitment progress in forthcoming issues!

Taking part in research helps us learn more about mitochondrial disease, so we can provide you with better care, and in each newsletter we will give you feedback on a research study.

Also look out for 'dates for your diary', like the forthcoming Patient Information Day in London on the 7th May, useful websites, and fundraising news! If you have any feedback or suggestions for the newsletter, or an article to be included in future issues please email the team at: mitonews@ncl.ac.uk.

Dates for your Diary...

- **Patient Information Day - Saturday 7th May 2011, London**
- **Mitochondrial Open day at Newcastle University - Spring 2012**

Useful Contact Information

Newcastle upon Tyne
Research Nurse Tel: 0191 282 9351
Address: The Medical School, Newcastle University, NE2 4HH

London
Helpline Tel: 0845 155 5000 (ext 3028)
Address: 8-11 Queens Square, London, WC1N 3BG

Oxford
Research Nurse Tel: 01865 226015
Address: Churchill Hospital, Headington, Oxford, OX3 7LJ

Websites

The Rare Mitochondrial Disease Service for Adults and Children:
www.mitochondrialncg.nhs.uk

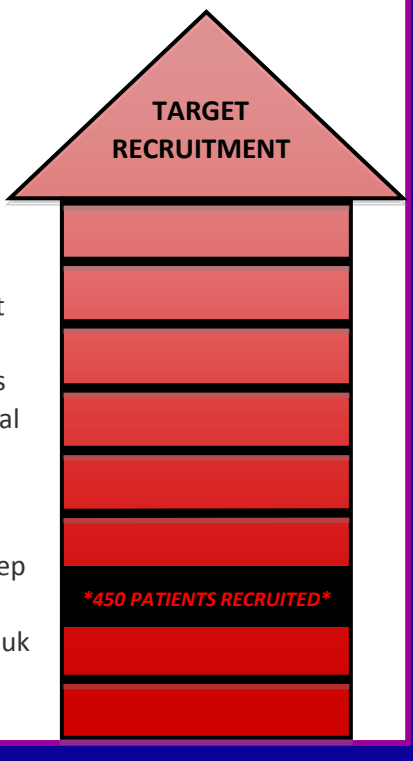
Muscular Dystrophy Campaign
www.muscular-dystrophy.org

Treat NMD	www.treatnmd.eu
CLIMB	www.climb.org.uk
We Move	www.wemove.org
The Lily Foundation	www.thelilyfoundation.org.uk
The Ryan Stanford Appeal	www.ryanstanfordappeal.org.uk

MRC Centre for Translational Research in Neuromuscular Disease Mitochondrial Disease Patient Cohort (UK)

What is it?

A database of patients (adults and children) in whom mitochondrial disease has been identified clinically or through lab tests. We plan to define a cohort of 1500 patients from across the UK.



Why do it?

Our aim in developing this cohort is to translate improvements in our understanding of the science of mitochondrial disease into direct health benefits for patients. We also want to further our understanding of mitochondrial disease and to evaluate the treatment of complications such as stroke, seizures, diabetes, poor growth and heart disease in patients with mitochondrial disease. The cohort will also act as a resource for assessing novel clinical interventions such as drugs targeted to mitochondria and exercise therapy.

How do I become part of it?

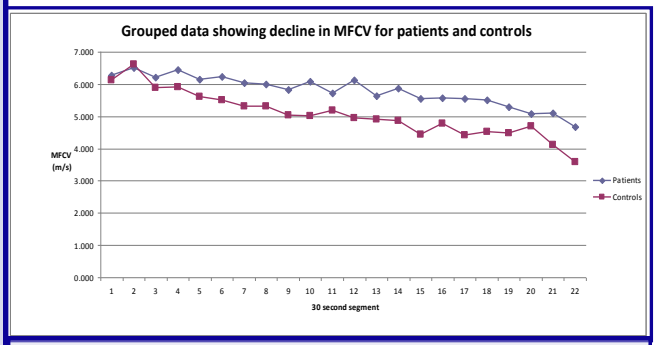
Many of you have already consented to be part of the cohort (thank you!) and we will keep you informed of our progress. If you have not yet consented, but would like to, please contact your local mitochondrial team for further information or email: mitonews@ncl.ac.uk and we can arrange for information and consent forms to be sent to you.

Research Feedback

Why do patients with mitochondrial disease get tired?

Firstly, a big thank you to all the patients who took part in our research study to look into this question. Most of the work was done by a medical student called Matthew Bolland who worked with me on this project; you'll remember his cheery smile and scruffy hairdo! Matt is now back on the wards, but partly as a result of this project was awarded an academic training post to continue a career in research, so hopefully he will continue to contribute to the field.

Matt demonstrating the experimental set up (a mug of tea was optional!)



Graph showing the decline in contraction speed with time. The patients (top line) show less of a reduction in contraction speed than the controls (bottom line).

Our project was to look into why patients with mitochondrial disease feel such extreme fatigue in their every day lives.

Fatigue means different things to different people, but to us in neurophysiology it means more and more weakness developing in the muscles. We tested this by measuring the speed at which the muscle contracted during exercise. In normal controls (people without mitochondrial disease), the speed of contraction goes down over time if you hold a steady contraction, say for example bending the elbow against a weight. This happens because lactic acid builds up in the muscle (which is what produces the burning feeling that you get after a bout of exercise).

We thought that patients with mitochondrial disease would show the same effect, but that it would happen faster. The really interesting thing that we found is that the exact opposite occurs; ***patients with mitochondrial disease can maintain the same speed of contraction for longer than controls***. This is a really intriguing result, and tells us a lot about how the muscles work in mitochondrial disease. Firstly, it tells us that in patients, the muscles are used to working well even in the presence of lactic acid. This means either that the muscle is adapted to high levels of the acid, or that it is able to pump it away into the bloodstream. Secondly, it tells us that the fatigue is originating somewhere else in the body; perhaps in the brain or in the connection between the nerve and the muscle. We are carrying on the research to try and find out the answers to these questions, so watch this space and please keep volunteering for our experiments!

Roger Whittaker, Consultant Neurophysiologist, Newcastle

Know your Team
Professor Doug Turnbull

Professor Doug Turnbull
Professor of Neurology &
Director of Newcastle University
Centre for Brain Ageing and Vitality
Mitochondrial Research Group, Newcastle Upon Tyne, UK.

I am a Consultant Neurologist who has been looking after adult patients with mitochondrial disease for a number of years. Based at the Royal Victoria Infirmary in Newcastle, patients are seen either in the outpatient department, as a day-case or occasionally admitted to the Neurology ward. I divide my time between looking after patients and the research laboratory in Newcastle Medical School. Laboratory based research is essential if we are to understand more about mitochondrial disease and develop new treatments. Outside of work I am a keen cyclist and a Newcastle United season ticket holder.

Team Talk
Mr Geoff Bell

Geoff Bell
Clinical Trials Co-ordinator
Newcastle Upon Tyne, UK.

I have worked with the MRC Neuromuscular Research Centre in Newcastle for 4 years and am based between the sites at the International Centre for Life and Newcastle University Medical School. My post can be extremely varied and I cover a wide range of duties from preparing new research studies/clinical trials for review at ethics committees, to meeting patients and families who are interested in participating in clinical trials, and discussing protocols with members of the medical team. The main challenge of the role is to try and keep on top of the ever-changing legislation that governs clinical research and pass that information on to members of the team, whilst remaining upbeat…the latter of which is not helped by me being a long term avid Newcastle United supporter!

Fundraising

The Mitochondrial Research Team at Newcastle University are extremely grateful for the generous donations that they receive from your fundraising efforts. Julie Corby, Nicola Bardett and Luke Bardett (pictured above) participated in a sponsored walk to raise funds towards MELAS research and kindly donated a cheque for £650 in memory of Gillian Smart, who was Nicola's mother, Julie's sister and Luke's grandma. As you know we are very active in trying to improve the diagnosis, obtain a greater understanding and eventually develop treatments for patients with mitochondrial DNA disease.

We want to hear your news! If you have a story you would like to share or if you have any feedback about our first issue please email: mitonews@ncl.ac.uk