Welcome.....

Welcome to our second issue of MitoNews UK, which provides news and information for patients with mitochondrial disease. Thank you very much for your feedback and suggestions following our successful first issue and if you have a story that you would like to share please let us know.

In this issue find out further information about a possible new IVF technique which could prevent the transmission of mitochondrial diseases from mother to child. There is a progress update on the Mitochondrial Disease Patient Cohort and how you can get involved if you are not already! Find out more about a patient focus group that took place in Newcastle and read about two families and their incredible fundraising efforts.

Don’t forget to visit our website: www.mitochondrialncg.nhs.uk which provides information on all aspects of mitochondrial disease. If you would prefer to receive future copies of MitoNews UK by email instead of by post then please email the team at: mitonews@ncl.ac.uk

**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**
Julie Phipps, Clinical Genetics Research Nurse
Email: Julie.Phipps@orh.nhs.uk
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

**Dates for your Diary...**

- **Patient Information Day at John Radcliffe Hospital, Oxford on Friday 16th September 2011 at 5pm.**
- **Mitochondrial Open day at Newcastle University - Spring 2012**

**Hope for Families with Mitochondrial Disease**

There has been a lot of publicity recently about a possible new IVF technique which could prevent the transmission of mitochondrial diseases from mother to child:
http://news.bbc.co.uk/1/hi/health/8619533.stm and http://www.youtube.com/user/wellcometrust#p/u/27/0wFn9Oj4u2E

The research being led by Professors Mary Herbert, Alison Murdoch and Doug Turnbull is collaboration between Newcastle Fertility Centre and Newcastle University supported by the Muscular Dystrophy Campaign, Wellcome Trust and MRC. The technique involves transferring nuclear DNA (which, in humans is packaged into 46 chromosomes) from a fertilized egg (with affected mitochondria) to another donor egg (with healthy mitochondria). This new IVF technique would only be suitable for families who have abnormalities (mutations) in their mitochondrial DNA. Although encouraging results have been obtained in the pre-clinical stages, the current law does not allow this approach to be used as treatment. An expert panel commissioned by the regulatory body known as the Human Fertility and Embryology Authority (HEFA) has called for additional studies to be done to ensure that the techniques are as safe as possible before being allowed in the clinic. These new experiments are being performed by the Newcastle group and involve working with normal human eggs which are given for research by women interested in helping others. If you are interested in donating eggs please contact the Newcastle team (maria.nesbitt@ncl.ac.uk).

The Newcastle team is working closely with opinion leaders to encourage the government to change the current legislation. Currently pre-implantation genetic diagnosis (PGD) can be offered to some women with mitochondrial disease as a potential way of limiting the transmission of affected mitochondria from mother to child. PGD involves undergoing IVF treatment to produce multiple embryos, which are tested for mutations in mitochondrial DNA. Only the embryos with the lowest level of mutation are replaced in the womb. This is also a very new technique but both Newcastle and Oxford mitochondrial centres are able to offer this treatment. Prenatal diagnosis is another option offered to women known to have mitochondrial mutations who would have the option to terminate the pregnancy if the mutation was detected. Cells taken from the placenta (taken between 11 & 14 weeks gestation) can be analysed to estimate the level of mutated mitochondria in the developing fetus. This is not straight-forward for all mitochondrial diseases however as the levels of mutated mitochondria causing disease varies for each mutation. The Mitochondrial Research Group at Newcastle University continue to lead further studies in this field with the hope of providing future options to preventing the transmission of mitochondrial disease in those affected. Further information can be found on our website: www.mitochondrialncg.nhs.uk/pa-adviceforwomen.html or an appointment in our out-patient clinic can be made to discuss things further.
Ryan Stanford (pictured above, centre) was born on the 7th August 2008 with Alper’s Syndrome, a genetic condition which affects fewer than 1 in 200,000 children. The Ryan Stanford Appeal was set up on the 16th October 2009 by Ryan’s parents David and Anne (pictured above), in order to raise money to fund medical research into Alper’s Syndrome.

Helen Bogle, a PhD student at Newcastle University, (pictured above, right) is currently completing a research project titled ‘Fight Alpers’, which is funded by the appeal. To find out more about the Ryan Stanford Appeal please visit: www.ryanstanfordappeal.org.uk.

**The Focus is You!**

March and April were busy months for us to get you more involved and tell us what you think. After a national press release about technological advances of IVF techniques that could affect mothers with Mitochondrial Disease, we invited several patients to attend an informal meeting to express their views. The group were able to inform us of where patients think our future research developments should be focused and Professor Turnbull was able to take this information direct to parliamentary debates.

Research within our department continues and new projects are continually being planned. A further focus group was asked to look at the challenges and barriers patients face when asked to take part in studies and also to help us develop user friendly patient information. Both meetings were successful.

As well as being informative for both patients and professionals, those involved found meeting others with similar symptoms very supportive. There were lots of names and phone numbers swapped. A big thank you goes to all those who attended! The mitochondrial team are keen to know what you think about your care and our service. If you would like to take part in a future focus group or be part of a patient led support group please contact: Catherine.Feeney@nuth.nhs.uk

**Know your Team**

Dr Robert McFarland

I am a Clinical Senior Lecturer and Consultant Paediatric Neurologist and I’ve just realized that my job title probably doesn’t help you understand who I am or what I do! I work as a senior doctor at the Great North Children’s Hospital in Newcastle, where I treat children with a variety of problems affecting their brain, nerves or muscle and some of these children have mitochondrial disease. In fact every Monday, Prof Turnbull, Dr Schaefer, Dr Gorman and I run a specialist Mitochondrial Clinic, open to anyone with mitochondrial disease from throughout the UK. I see the children who attend the clinic but, just occasionally, I also review the ‘grown-ups’, although I have to say, I usually put the toys away! I also work in the Mitochondrial Research Group trying to tackle the problems caused by mitochondrial disease and look at ways to bring advances in the laboratory into the Mitochondrial Clinic. Sometimes this works the other way round and believe me, we still learn a lot about mitochondrial disease from the patients we see! Outside of work, I’m fast becoming a professional taxi service for my 3 boys, who are engage in a variety of sporting activities including rugby, which, with variable success, I also try to coach!