Feedback

Mito app **Would you like to get involved with developing an app for mitochondrial patients?** We are trying to develop an app for your smart phone to help patients and their families get direct access to useful information and tools. As a direct result of this year's Newcastle Patient Day, we have an idea of the information that would be useful to you as a patient (as shown in the picture to the right). Some of the most popular information requested was information on the heart, nutrition, fundraising, research and social networks. If you could add to this list, have any ideas or talents you could contribute, or would just like to get involved, we want to hear from you so please get in touch with Julie Murphy at Newcastle University at the following email address:

mrg.socialmedia@newcastle.ac.uk

Happy Christmas and best wishes for the New Year from the MitoNews team!

Thanks again for everyone in the Newcastle Mitochondrial Research Group who participated in Christmas jumper day which raised £70.36 for Mitochondrial research.

100% of patients rated the patient day **Good to Excellent**

It was another successful year for the Newcastle patient information day thanks to the contribution and hard work of you the patients and the Newcastle based organisers (from left to right above) Berni Caygill, Sister Catherine Feeney and Sue Callender. If you have any ideas or suggestions about the patient meeting please do not hesitate to get in touch with Mitonews at the address below.

Useful contacts

Newcastle upon Tyne
Specialist Nurse Tel: 0191 2821740
Address: The Medical School, Newcastle University, NE2 4HH

London
Specialist Nurse Tel: 0203 4488009
Address: 8-11 Queens Square, London, WC1N 3BG

Oxford
Julie Phipps, Clinical Genetics Research Nurse
Email: Julie.Phipps@ouh.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**

If you have a story you would like to share or have any feedback about this Christmas edition please email: mitonews@ncl.ac.uk

Oxford University Hospitals NHS Trust | MRC Centre for Neuromuscular Diseases | NHS Specialised Services | UCL | Newcastle University

MITO NEWS

Inspirational people -Christmas special

At seven weeks old you expect your little one to be reaching out to hold your hand not to be rushing with them into intensive care because they are having seizures. This is exactly what happened to Liz and Dave and their beautiful daughter Lily. It was only after spending a week in intensive care that they were given the news every parent dreads; their daughter was going to die from a disease for which there is no cure or treatment. This was mitochondrial disease and her prognosis was days.

As a result of Lily's determination and strength and the loving environment her parents and two sisters created, Lily survived six months. The knowledge that they would lose their precious baby and the uncertainty of when it was going to happen was harrowing. During this time they wanted to reach out to other families who were going through the same thing or had done previously, to ask for advice. Like many people who suffer from mitochondrial disease or have affected family members, they came up against a brick wall. There was very little information, support and awareness available to patients with this often debilitating disease. It was then that the Lily foundation began, to help families like them, to raise awareness and offer support to patients with mitochondrial disease and their families.

This issue's cover story:
At this years patient information day in Newcastle, **Alison Maguire** gave an eloquent and moving talk on her own experience of mitochondrial disease and the work the Lily foundation is doing to help patients and their families.

As a direct result of some of the 'Lily literature' circulated by Lily's mum, **Alison Maguire** got involved with the charity. Only days after losing her daughter she was at a soft play centre and found a leaflet about the Lily foundation. Alison's daughter **Niamh** had been in and out of hospital since she was 10 weeks old with seizures and movement problems, but it was not until she was three years old that she was diagnosed with mitochondrial disease. Unfortunately the brick wall was also present in the medical community before Niamh was diagnosed. Tragically, a year later, at the age of 4, Niamh died. For Alison, like Liz, there was no family history of mitochondrial disease and for any parent to be told that their child is unlikely to have any long term future, as there is no treatment or cure, it is devastating and unbelievably lonely. Alison got in touch with Liz and they talked about their daughters and their experiences. Alison and Liz became friends and worked together at the Lily foundation, making a difference, raising awareness, and helping people of all ages with mitochondrial disease and their families. They don't have big offices but work from home juggling busy family lives whilst trying to raise money through events they hold throughout the year. **They may be a tiny charity but they have already changed the way mitochondrial disease is diagnosed and provided cutting edge equipment that has advanced the way that mitochondrial research is currently being performed.** If you would like to get in touch for help, advice or support or would like to get involved with the charity please see their website:

www.thelilyfoundation.org.uk
Facebook: www.fb.com/lilyfoundation, Twitter:@4lilyfoundation

Welcome to our Christmas special edition of **MitoNews**, our newsletter for patients with mitochondrial disease.

Laura Jakubowski has multiple mitochondrial DNA deletions and is profoundly deaf with a cochlear implant. She is 28 years old and lives in West Yorkshire. We had the privilege of interviewing her for this special Christmas edition of mito-news.

How old were you when you were diagnosed?

I was 18 when I was diagnosed, at first I was scared as I, like many others, had never heard of mitochondrial disease but as the years have gone on I have got used to it. My family were relieved at finally getting a diagnosis as I had been having problems since the age of 8. After a period of seeing doctor after doctor and being misdiagnosed I had a muscle biopsy and was diagnosed with mitochondrial disease. As a result I could get the help I needed.

How do you like to spend your free time?

I like to sew cross stich, paint and go to the gym. I also like to chat to my mito friend via Facebook.

How has your life changed since your diagnosis ?

Most people I come into contact with haven't heard of mitochondrial disease and it's quite hard to explain what it is, and why I'm tired and achy. Sometimes I have one good day and three bad, but I take one day at a time and try not to worry about the future. ***I would tell anyone with mitochondrial disease not to be scared about it as there are lots of people out there that can help. Even though there is no cure at the moment, there is help out there.*** My friend Susan and I have set up a Mito adult support group on facebook, where people can get help and advice and we try to answer each others questions. It is a place where patients and their families can discuss what they are going through and the help they are receiving.

To get in touch with Laura search for Mito adult support on Facebook

doubting how far you can go, just remember how far you have come. Remember everything you have faced, all the battles you have won, and all fears you have overcome….You can do it!'

When we asked Susan about her quality of life she said she can no longer do the things she used to enjoy like shop for hours, play sports or dance but she remains inspirationally positive. She insists she does have a good quality of life, although it is different to what it was before. She has had to accept more help from family and friends which can be hard for an independent young woman. She has experienced many emotional ups and downs, but describes these as '***helping her to become a stronger person'.*** As a result of mitochondrial disease, she has felt isolated at times and admits that before she was diagnosed she had never heard of it herself. Unfortunately, in her experience, there is a lack of awareness in both the public and medical profession and she is passionate about changing this.

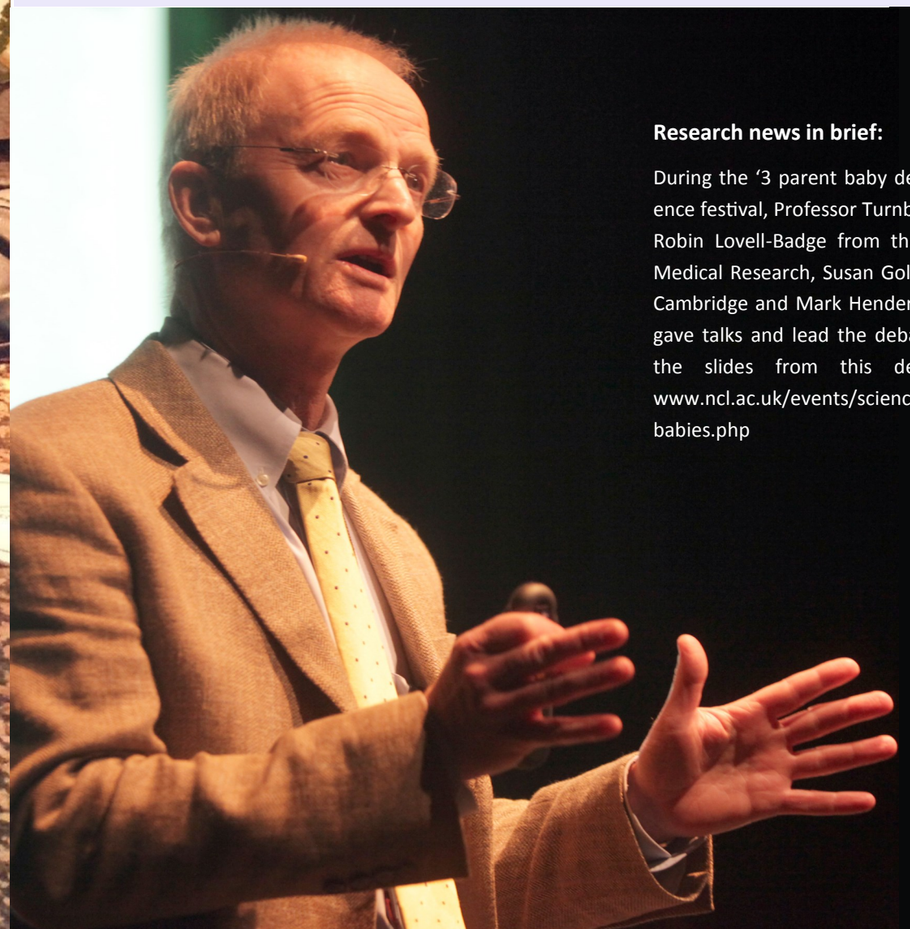
About a year ago, after realising there wasn't enough online support for adults with mitochondrial disease, she and her mito friend Laura (opposite) set up an online Mito adult support group on Facebook which has over 425 members across the globe. Susan and Laura act as administrators for the group and review requests for people to join. They are proactive in asking how the members are doing and try to facilitate conversations about topics relevant to mitochondrial patients. They have created an environment in which people can ask questions, offer advice, and support others going through similar experiences.

If you would like to get in touch or involved please search for Mito adult support on Facebook.

'Be positive, make the most of what you can do and don't ever struggle on your own. We are a mito family and we are all there to help and support each other.'

These are the words of advice from Susan Warnock. Susan is like any other 30 year old in that she loves to spend time with friends and family and her 6 month old dog Rolo. She comes from a very close family that includes 4 beautiful nieces. At 26 years old she was diagnosed with mitochondrial disease and like many others with this disease, she was having symptoms from an early age and was even misdiagnosed for 10 years. When she was diagnosed with mitochondrial disease she described it as a shock and overwhelming, but also a relief as previously some doctors had suggested it was 'in her head' She finds it hard but is determined not to let mitochondrial disease ruin her life, although the uncertainty of what will happen in the future has affected her family. When she is feeling down, her loved ones have been pivotal in helping her through those difficult times. One of her favourite quotes from a loved one is:

'Whenever you find yourself



Research news in brief:

During the '3 parent baby debate' at this years British science festival, Professor Turnbull from Newcastle University, Robin Lovell-Badge from the MRC National Institute for Medical Research, Susan Golombok from the University of Cambridge and Mark Henderson from the Wellcome Trust gave talks and lead the debate. If you would like to view the slides from this debate please visit: http://www.ncl.ac.uk/events/sciencefest/video/three-parent-babies.php