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.

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 Niámmí 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 Niámmí was diagnosed. Tragically, a year later, at the age of 4, Niámmí 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
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 group on Facebook.

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