Case for Support

supporting our vision:
a world in which people with Barth syndrome LIVE, and live WELL

Barth Syndrome UK

Barth syndrome is a rare genetic life-limiting condition. We know of 300 survivors out of a total of 500 cases in the world.

It can affect you in many different ways...

Your heart: causing heart failure, cardiac arrhythmias and sudden cardiac arrest

Your immune system: causing repeated and serious bacterial infections and sepsis

Your muscles: causing extreme weakness and often debilitating fatigue

There are currently no treatments and no cure for Barth syndrome

BARTH SYNDROME UK

Our Mission: Saving lives through education, advances in treatment and finding a cure for Barth syndrome

We work to support families, fund research and raise awareness of Barth syndrome. Because of its rarity, we work very closely with similar organisations. We share resources and information to accelerate progress and achieve results comparable with much larger organisations. But our targeted approach allows us to focus specifically on improving the lives of our affected families here in the UK.

History and Background

Founded in 2003, by parents of affected children, we are the only charity in the UK wholly dedicated to supporting families with Barth syndrome. We are run by a team of affected families and specialist talent.

Registered as a charity in 2003 under the name Barth Syndrome Trust, in 2019, we changed status to a Charitable Incorporated Organisation (CIO). We also changed our name to Barth Syndrome UK with a new charity number 1181830.

Our goal is ambitious - a cure for Barth syndrome. Until that day comes, we will work to help affected children and adults make the most of their lives.

How do we help?

Finding out that your child has Barth syndrome is devastating

Sarah and Dave's story

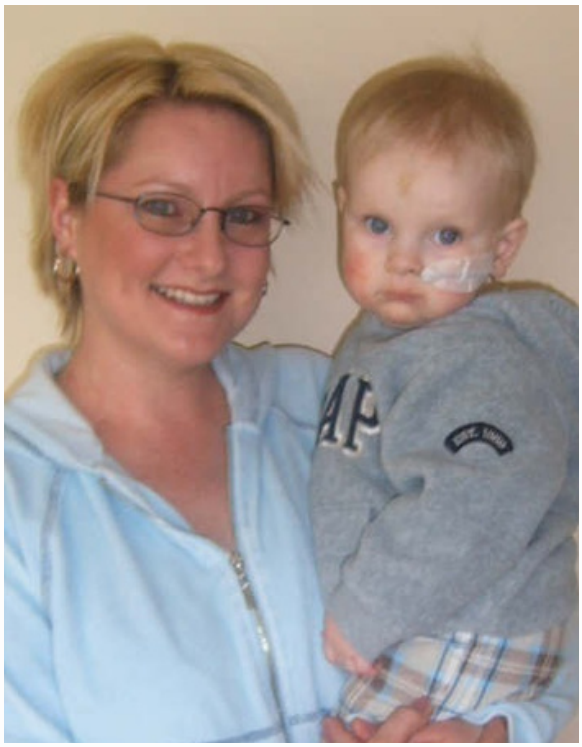
Written by Sarah

Our story began on the day Ashley was born. His cry was very quiet and we thought it was great not to have a screaming baby. I was concerned about his feeding. During the first few weeks he would not feed for more than 2-3 minutes... At the clinic I was told not to worry. I burst into tears and we were then seen by a doctor.

At 3 months, he was admitted to Bristol Children's Hospital and began nasogastric feeding. After a week of investigations, we were told he had dilated cardiomyopathy...We were in shock.

It wasn't long before we had a visit from Dr Colin Steward. He told us of a rare genetic condition that he thought Ashley could have - Barth syndrome. As he talked, Dave and I looked at each other in horror. We were still trying to accept that Ashley had a serious heart condition, and now we were being told he could suffer with more!!

We were put in contact with Michaela Damin (Chief Executive of Barth Syndrome UK). I'll never forget our first conversation. I had an instant feeling of support. She told us about the patient support group. We were introduced to the private email forum which gives us access to other families with Barth syndrome, doctors and other experts. Now we couldn't imagine our lives without it. Through it we have had so much knowledge and support from so many people, and we'd like to think that we have helped others too. We now feel we have an extended family, from all over the world.



Sarah & Ashley

Services we offer

We work on many levels at the same time...

PROJECTS	DETAILS
Diagnosis and access to medical care	We help families get the critically important diagnosis for their child. We connect them with specialist doctors and services to give them immediate support and help.
Barth Family Community	We help end the isolation of an ultra-rare disease by connecting them to other Barth families here in the UK and worldwide. We provide access to a private WhatsApp group and one-on-one support in early days and during periods of crisis. We offer families a chance to meet and offer mutual support. And we provide a chance for their children to play with others who are like them. We act as a knowledge hub for current best treatments and management for Barth syndrome. Our website, praised for its high quality, is regularly updated with expert guidance and information.
Research & Advocacy	We help guide and steer new developments that affect Barth families. We fund research into new ways to treat Barth syndrome. We fund research into long term projects in our search for a cure.

Some of our work

2003: Founded Barth Syndrome Trust

2004: First volunteer-led specialist medical clinic in the world

2006: First reliable and fast diagnostic test for Barth syndrome

2008: Family Route Map to help families navigate through the complex web of care (with Jeans for Genes)

2010: First sustainable national NHS Specialised Service for Barth Syndrome (first of its kind in the world)

2012: Launched new website www.barthsyndrome.org.uk

2019: rebranded as a CIO and changed name to Barth Syndrome UK. Hired first employee.

2019: Facilitated the first ever clinical trial for a treatment (CARDIOMAN trial) in the UK

2020: Provided specialised support to all families throughout COVID pandemic

2022: First project assessing prevalence of neurocognitive conditions in Barth syndrome, a potentially new element of the syndrome

First Volunteer Clinic led by Prof Colin Steward, 2004

Research we've funded

Since 2006, Barth Syndrome UK has funded

14 grants to the total value of over £300 000 for quality research into Barth syndrome

These research projects have all been carefully chosen to further our understanding of the causes and effects of Barth syndrome

It is our hope that they will lead to better survival rates and better quality of life for all our affected families

OTHER THINGS WE DO

AWARENESS

Run a long running targeted awareness campaign through the compilation of compelling family stories, regular newsletters, information brochures for families and doctors, hospital posters, liaison with specialist centres, presence at conferences

FINDING MORE FAMILIES

Identify new affected individuals/families in the UK: from 4 in 2003 to 37 in 2022

NATIONAL INFO/CONFERENCE DAYS

Provide regular outreach events throughout the UK with an emphasis on family support, education, skills and independence building and fun

INTERNATIONAL CONFERENCE

Help to create a quality international Family, Scientific and Medical Conference that takes place every two years

WHO DO WE HELP?

Families and individuals with Barth syndrome

Medical professionals

Teachers, Learning Support Assistants and Special Educational Needs Coordinators (SENCOs)

HOW DO WE USE YOUR MONEY?

We are an independent not-for-profit organisation. We use all funding only to support our stated programs

OUR PLANS FOR THE FUTURE

We've identified a number of projects that are needed by our community that we'd like to prioritise in the near future subject to funding availability:

- **Young Adult weekend workshops for help with transition into adult life.**
- **Specialist workshops for advice and support around neurocognitive conditions such as ADHD, ASD, dyslexia etc.**
- **Video project to create information resources for our affected individuals and their families.**
- **Help with getting affected individuals properly diagnosed with neurocognitive conditions.**
- **Family workshops around access to benefits and help with rising costs.**
- **More support for adults with Barth syndrome.**
- **Preparation for future clinical trials.**

Moving to the next level - why we feel we deserve support

As a small charity with limited resources, we've come a really long way but we believe we can do much more!

We believe that, with the right support, our children with Barth syndrome can have a happy and fulfilling life. They can grow into adults and live independently and with meaning and purpose.

They can't do this without our support. And we can't provide it without your support.

What our families and partners are saying about Barth Syndrome UK

'The Barth Syndrome UK charity is a small but dynamic charity for families affected with this ultra rare disorder. They were pivotal in the setting up of a multidisciplinary service and in subsequently sourcing funding for a specialised service.

The Barth Syndrome UK charity has changed the course for many families who now feel supported. As a healthcare organisation close working with the charity has ensured the continued delivery of patient centered care and led to identification of gaps in service provisions. Close working has helped with the development of a service that actually meets patient need. We have directly seen how much their work has benefited the care of these patients and their families and look forward to continued close working with them.'

NHS National Barth Syndrome Service - March 2022

'We've been volunteers since 2005 and have seen the difference the charity has made to the lives of affected individuals and their families. Before the charity there was nothing; diagnosis was difficult and slow; awareness in the medical community was low; families were struggling alone, suffering the loss of their sons, generation after generation and not knowing why. '

Nigel and Lorna - March 2022

How do we help?

Our Family Services team are here every step of the way

A mum's experience...

When my son was first diagnosed with Barth syndrome, my mental, emotional - and for me at the time in particular - financial circumstance was a big one.

Luckily I was offered a slot at the specialist clinic in Bristol. However, everything was incredibly overwhelming, even thinking about travelling there. The Family Services team at Barth Syndrome UK supported me through out the whole booking, talked me through every detail that I needed to know. BS UK supported me financially, if the organisation didn't help me out at the time, I would not have been able to go to my first clinic with my son. Their involvement at the time was of extreme significance for me and my family.

The organisation supported me with my stay at the hotel as well. Furthermore, any queries I had were answered in the most kindest of manner. The organisation was understanding of my difficult circumstance, from train tickets, to getting on the shuttle bus, to even the hotel stay, I was supported incredibly throughout it all.

Moreover, I had tremendous assistance with other things alongside the financial side, such as helping me with my DLA form for my son. I had great difficulty in getting my head around such a lengthy form, and didn't know where to start. Throughout the whole time I had support in ensuring I filled out my form to the best of my ability for my son, thus making sure I hadn't missed any details

.

I must also highlight the emotional and mental support I received after finding out my son was born with Barth syndrome. Two other mums both got in touch with me and reminded me that I was not alone, their reassurance and guidance pulled me out of a dark place. Knowing I'm not alone in this journey and that there's a whole community that can resonate with me had given me great comfort.