BSUK NEWSLETTER

OCTOBER 2024

Photo by Ollie Baxter-Smith, Cotswold-Creative

Barth Syndrome UK

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UPDATE FROM THE CHAIR

Our annual board meeting highlighted the pressing need to continue finding ways to improve engagement and fundraising within our Barth community.

We are joined in our quest by two new members of the board: Paul Ford, grandad to Mackenzie Doran (and his twin brothers), who has thrown himself into fundraising and grant applications. Also Joanna Kozminska, who has a legal background and whose expertise will no doubt prove invaluable.

We've set up an Advisory Committee made up of adults with Barth syndrome. To families with younger children, we also need your insights on how to foster deeper connections going forward.

In addition to our active pursuit of fundraising from grantmaking organisations, we rely on continued fundraising support from the community itself.

We ask that where possible, you:

Let's unite as a community to overcome our challenges and create a brighter future for individuals and families affected by this condition.

Together, we can make a real difference and ensure that those living with Barth syndrome have access to the support and resources they need.

Thank you.

Ollie Baxter-Smith, Cotswold-Creative

Donate generously: Establish monthly direct debits or increase existing contributions if you can. Every donation, no matter the size, makes a significant impact.

Spread awareness: Reach out to family, friends, and colleagues, sharing information about Barth syndrome and our mission.

Get involved: Participate in Giving Tuesday (December 3rd) and community events.

Engage your school and workplace: Explore fundraising opportunities and corporate partnerships.

Leverage local networks: Connect with Rotary Clubs and Round Tables. These organisations can provide valuable support and resources. Our Trustees can guide you through this process.

Share online: Promote our social media posts to raise awareness and foster community engagement. Together we can reach a wider audience and spread the word about Barth syndrome.

FAMILY DAY UPDATES

Our recent family day was a resounding success! We gathered to celebrate achievements, learn about the latest research, and discuss future plans for our community. Families enjoyed connecting with each other, receiving medical updates, and participating in fun activities. Thank you to everyone who attended and made this event a memorable experience!

Ollie Baxter-Smith, Cotswold-Creative

Flappy the new Barth Service mascot has been enjoying spending his time with the Craigie family.

Which family will he choose for his next adventures? He looks forward to seeing who will take him home from the next Bristol clinic upon his return.

MINDFULNESS

Mindfulness Top 3 Things:

1. Mindfulness has been shown to support and improve mental wellbeing.
2. Mindfulness involves paying attention to what is going on inside and outside ourselves, moment by moment.
3. Lots of different techniques - find something that works for you which you can incorporate into your life.

Parental Resilience Top 3 Things:

1. Resilience is the process of managing stress and functioning well even when faced with challenges, adversity, and trauma.
2. Our session looked at how and what we can do to build our resilience (our resilience tree).
3. What to do if we need support - reach out to others, remember both the importance of self care as well as having a good support network.

HAEMATOLOGY

Filgrastim- alternative form of G-CSF

Benefits:

- more concentrated =less volume to inject
- comes in prefilled syringes (no need to make up syringe)
- is cheaper

Considerations:

- must be stored in fridge (can be out of fridge for up to 14 days)
- may need more frequent delivery
- may be difficult with smaller doses

If you have questions or are interested in trying this medication please discuss this at your annual appointment.

Research in Neutropenia

Presentation by Katy Fleming (working with Dr Amulic, University of Bristol)

We talked about the normal role of neutrophils in the body.

We discussed that neutrophils in people in Barth syndrome work properly but some problem occurs during the early stages of neutrophils which therefore ends up with decreased numbers.

Copper Levels - We looked at copper levels in the blood of Barth individuals and noted that people with neutropenia have lower copper levels. We will need to investigate further to find out if a low level of copper plays a role in low neutrophil levels.

DIETETICS

We're excited to announce that the Bristol Barth Syndrome Service now has body composition scales available for use at clinic by individuals over 12 years old.

These scales provide valuable information about your:

- Muscle, fat and bone mass
- Visceral fat
- Body water
- Basal metabolic rate

By understanding your body composition, you can make informed decisions about your health and fitness goals.

Thank you all for your purchases and donations at the Family Day. Together we raised an amazing total of...

£248.54

FAMILY DAY UPDATES

AFFECTED INDIVIDUALS’ FEEDBACK

Things that we really appreciate:

- **The charity always attempts to include us affected guys when making important decisions**
- **The Barth Service always provides us all with amazing, personalised care, regardless of what we may need.**
- **Gillian Alexander has been particularly amazing, consistently being the one person that we can all go to with whatever issue we may have.**

Things to work on in the future:

- **Even more access to mental health services for both affected guys and their loved ones.**
- **More opportunities for us to meet each other, ideally in a more social context, without as much of a focus on all the medical stuff**.

Ollie Baxter-Smith, Cotswold-Creative

CARDIOLOGY

- Cardiology provision for Barth individuals is now under Dr Catherine Armstrong (0-17 yrs) and Dr Eva Sammut (17 yrs+).
- Cardiac provision for 0-5 yrs is well managed by the acute hospital teams and intensivists and then the Barth Team.
- Adolescence can be a difficult time for our Barth affected young individuals as deteriorations are difficult to predict.
- In adults, there is stronger evidence for medications that will significantly improve outcomes. These are referred to as the **4 Pillars** of therapy. These reduce the work your heart has to do or slow the heart rate reducing the oxygen demand of the heart.

The Four Pillars:

1. Betablocker - e.g. Bisoprolol.
2. Dapagliflozin - sodium glucose cotransporter-2 inhibitor (SGLT2i). This can theoretically cause low blood sugar, so we monitor this when we introduce it.
3. Entresto - a combination heart failure drug, with slightly stricter criteria to start. We need to be careful to allow the patients on ACE inhibitors (captopril, enalapril, ramipril etc) to washout for around 48hrs before starting Entresto.
4. Finerenone -a diuretic like spironolactone but with less undesirable side effects like increased breast tissue.

Shared care is important in the management of Barth individuals. The challenge is that local cardiology services can vary and do not consistently lend themselves to shared care. This is an area we are looking at but there are no easy answers.

POWERBREATHE

Natasha Taylor (Physio) - Strength training of inspiratory muscles by breathing in against resistance has been found to be useful for adults with cardiomyopathy (not caused by Barth syndrome). It has helped to improve their breathlessness on exertion and decreased fatigue.

I am currently in the data collection stages, but I am looking into trialling this with people with Barth syndrome to see if it helps symptoms of fatigue. Theoretically this will be a 6 week course of using the Powerbreathe for 30 breaths, twice per day, with assessments before and after the course.

Minimum age is 7 years old and a heart transplant will not necessarily exclude you.

ZIOPATCH

Ziopatch is a new method being trialled by the Barth Syndrome Service intended to replace standard 24-hour Holter monitoring .
To maximise adherence, it is important to read and follow all the instructions carefully.
If it causes irritation, please let us know and it is okay to remove it. Some data is better than no data.

PLEASE remember to return it even if not used. There is a significant cost to the Barth Syndrome Service if you do not.

CONFERENCE UPDATE

Barth Syndrome Foundation Conference 2024: Impact on Research, Connection, and Progress

The 2024 Barth Syndrome Foundation (BSF) International Conference was a resounding success, bringing together 350 attendees, including 56 affected individuals, to advance research, medical care, and connections within the community.
Thanks to your fundraising support, BSUK was proud to sponsor two key medical professionals from the Bristol Barth Syndrome Service to attend, to learn and share best practice.

Research and Poster Presentations

Eight groundbreaking research projects were conducted onsite, covering a wide range of topics including cardiac data collection, strength testing, arrhythmia, cardiolipin testing, quality of life studies, pill swallowing techniques, and the Barth Syndrome Registry.

There were **38 different poster presentations**, covering a wide range of topics. UK dietician Victoria Wilkins won the Favourite Poster Award with her depiction of weaning – a project featuring none other than the famous Mackenzie!

Along with a packed agenda of **Scientific & Medical Talks**, BSF launched a significant new initiative this year—**Online Medical Training Modules**—designed to train the next generation of doctors about Barth syndrome. The Bristol team played a key role in developing these four hours of recorded sessions, which will be invaluable for future medical training and awareness.

A Community United

One of the most poignant moments was the Luminary ceremony below, with 331 glowing bags symbolising each living person with Barth syndrome, and 240 dragonfly cutout bags representing those we've lost. This moving tribute reminded everyone of our shared mission.

Workshops were held on quality of life, feeding, occupational therapy, physiotherapy, and sleep issues. These small group sessions, tailored to different age groups, provided valuable insights for both medical professionals and families.

Building Connections and Collaboration

A major takeaway from the conference was the **power of connection.** In a single week, face-to-face interactions between researchers, doctors, and families led to collaborations that would take months or years to achieve remotely. These connections are critical in pushing forward research and medical care for Barth syndrome.

Looking Forward

This conference was all about connection—between families, medical professionals, researchers, and affected individuals. From restorative sessions like yoga and meditation to emotional discussions around grief and resilience, there was something for everyone. The laughter, tears, and hugs shared throughout the week underscored the power of this community to support one another and drive progress.

As we look to the future, the bonds formed at the conference will continue to fuel our shared mission: to improve the lives of those affected by Barth syndrome through research, care, and above all, connection.

ELAMIPRETIDE

Elamipretide has the potential to be the first Barth specific approved medication.

Elamipretide binds to cardiolipin. It stabilises the folding of the inner mitochondrial membrane, improving mitochondrial structure and function, and reducing the proportion of immature cardiolipin which is increased in Barth syndrome.

Though the TAZPOWER crossover trial did not show statistical significant improvement, the TAZPOWER Open Label Extension study showed that those patients who continued elamipretide had statistically significant improvement in strength, functional exercise performance, balance, cardiac function, and also had a reduction in immature cardiolipin species.

In the US, Stealth Biotherapeutics along with the support of BSF and the Barth community, are making the case to get elamipretide approved by the FDA. On 10th October, the FDA Advisory Committee voted 10/6 in favour of elamipretide being approved for use in the US. Although the FDA still have the final say, this is a positive step for now. The outcome of their decision is expected by the end of January 2025. In Europe, Pharmanovia holds the license to market elamipretide if it is approved.

HOW CAN YOU SUPPORT FUNDRAISING?

Every donation, big or small, makes a difference. Whether it's hosting a cake sale, setting up a regular contribution, or setting up corporate partnerships, your fundraising efforts can help Barth Syndrome UK provide vital services to our families.

Contact us to find out how we can support you in your fundraising efforts.

Let's come together to create a brighter future for our community!

COACHING & COGNITIVE HYPNOTHERAPY

Penny Ellison *- I am able to offer cognitive hypnotherapy and coaching to individuals affected by Barth syndrome, as well as their parents and siblings.*

Cognitive Hypnotherapy *feels more like a conversation where clients are relaxed enough to find solutions to their problems. Despite the challenges we face, our minds are able to help us find new ways through when we feel stuck; using positive psychology and neuro-linguistic programming, clients can playfully challenge their situations.*

However, some people benefit from having ***coaching*** *sessions to work on their goals and focus on improving their future.*

Whichever is the best route for clients, I am able to offer daytime and evening appointments via Zoom on Mondays and Tuesdays. Please contact Michaela Damin if you would like to have a 30 minute chat to find out more.

https://longcroft-therapy.co.uk/

Psychology Barth Syndrome Service

Dr Martha Kenyon, Specialist Clinical Psychologist, has been in post since March 2024. She offers psychology support to the Barth community over a short, medium or longer term as needed. Reviews usually occur after the first 6 sessions to decide with you how many more sessions you need.

GRANTS AND OTHER FINANCIAL SUPPORT

Thank you to **Chelmsford Round Table** who have made a generous donation of £500 towards updating our website.

Chelmsford Round Table is the local Round Table for the Doran family. The family reached out to ask for support towards the development of BSUK's new website.

Trustees Paul Ford and Kelsey Doran were invited to introduce Barth syndrome to **Billericay Rotary Club** at its lunch on 22nd July. The lunch was very well attended and our representatives were made welcome.

Paul explained the background to the condition, the key symptoms and the support that BSUK provides and Kelsey candidly presented a mother’s perspective of Barth syndrome.

This event generated £350 towards BSUK’s core funds. This generous donation is much needed and appreciated.

Barth Syndrome UK is delighted and grateful to receive a grant of £7,500 from **Foyle Foundation** towards its core costs for the period October 2024 – September 2025. This type of grant is essential to maintaining BSUK’s existence and providing a base on which we can deliver our planned outcomes for our community.

FOYLE FOUNDATION

Rotary Club of Brentwood hosted a quiz night on 24th September to raise funds for BSUK. Participants from our community, including friends from BSF in America, enthusiastically competed in the challenging trivia contest. Paul Ford gave a brief introduction to BSUK and explained our fundraising goals and Kelsey Doran shared her moving personal story about raising a child with Barth syndrome.

The quiz night was a resounding success, raising £750. The Quiz Chair expressed his gratitude for the additional support from our community, which contributed to the significant fundraising total.

BSUK encourages our community to reach out to local Rotary Clubs and Round Tables to introduce Barth syndrome and explore potential financial support. The BSUK trustees are eager to assist in these efforts.

Rotary
Club of Brentwood

Shea Doran, father of Mackenzie Doran, affected individual, arranged a corporate partnership through his employer, **Barratt and David Wilson Homes Eastern Counties.**

The developer selected Barth Syndrome UK as its Charity of the Year in 2022, with BSUK receiving funds raised from various events held throughout the year. Events included a corporate golf day, a clay pigeon shoot, developer football tournament, raffles, bake sales, sweepstakes and an office BBQ. Barratts raised an impressive £41,946.20 for BSUK, with a subsequent donation of £3,000 in Jan 2024.

Saving lives through education, advances in treatments and finding a cure for Barth syndrome

BSUK INTRODUCES NEW ADVISORY COMMITTEE

A new Advisory Committee is being formed by BSUK to ensure that the voices of those directly affected by our services are heard at the highest levels of decision-making. This group will be comprised of individuals who have personal experience with the condition, providing invaluable insights into the challenges they face and the solutions they believe would be most beneficial. By working closely with the charity trustees, this advisory group will help to shape the future direction of BSUK and ensure that our programs and initiatives are truly responsive to the needs of the community we serve. If you are an affected individual aged 18+ who is interested in being involved, please contact Kelsey Doran for more information.

BOARD UPDATE

As we look ahead to January 2026, we will be saying a fond farewell to two very valued board members. Their contributions have been invaluable, and we are incredibly grateful for their dedication and service. To ensure the continued success of our organisation, we are seeking new volunteers to join our board and help guide us into the future. We are particularly interested in individuals with experience in finance, as we will be in need of a new Treasurer. If you are passionate about our mission and believe you have the skills and commitment to make a positive impact, we encourage you to consider joining our board.

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