

BBS UK Conference Updates

Advice Service and Clinics Updates

The Blind Warrior: Shodan to Nidan

Adam Crème:
Personal Perspective



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Foreword

Welcome to the 2023 Spring Newsletter

Over recent months, BBS UK has been busy, and the team has been changing. In December we said farewell to Christine, who retired from her role of Fundraising Co-ordinator; we are incredibly grateful to Christine for her contribution to the Charity and staff team over the past three years and wish her well with her next adventures – keep in touch Christine!



We also have two new trustees to welcome, Chetan Meshram and Gareth Owens, and thank them for offering their time and skills. I'm sure you'll join me in warmly welcoming them all to the team.

BBS UK is in the process of setting up a Service User Advisory Group, and when finalised, we will reach out to our members to see who would like to volunteer their time. The Service User Advisory Group is a subgroup of the Board and will provide a forum where a broad and diverse group of BBS service users, their parents and carers across the UK and ROI can share their perspectives and use their knowledge to help improve BBS UK.

I'm pleased to say that it has also been a busy time for fundraising; you can read more about this in our fundraising round up and I would personally like to thank you all for your fundraising efforts. As you know, our charity relies on donations and funding to provide the many services that we offer for members and their families.

Although the conference in September 2022 feels like a long time ago now, it's good to reflect on what a fabulous event it was and how great it felt to be able to get together in person after a 3 year 'gap'. Being back at the Hilton Northampton felt to me like coming home, and it was fabulous to meet many old friends and new ones alike. The speakers and sessions were all excellent and there was such a buzz in the air.

The team have already started planning for the next conference to be held on 22nd-24th September at the Hilton Northampton; booking information can be found enclosed with this newsletter and on our website. This will come around quickly I'm sure and will be another great opportunity to meet up, learn about research, and hear from expert speakers on a wide range of topics.

I hope that by the time this newsletter reaches you, the weather is starting to get a bit warmer, and we have sunnier days ahead. I know that I'm definitely looking forward to getting out and about more and my daily walks on the beach. As always, please keep in touch with us, and with each other.

Happy reading!

Laura Dowswell

Chair of the BBS UK Board of Trustees

Conference September 2022

For those who couldn't attend, and for those who loved it so much that they want to relive it, we've created a summary of the presentations for you to enjoy. The full videos from each presentation can be found on the BBS UK Website and YouTube channel.

Update on Research and Study of BBS

Professor Phil Beales

Professor Beales began the sessions by welcoming the attendees and commenting on the large number of new faces as well as familiar faces from previous conferences. The last in-person conference was in April 2019 and Professor Beales gave an update on the many developments since then, as well as drawbacks and delays caused by the COVID-19 pandemic.



- The BBS clinics teams rose to the challenge of not being able to meet face to face with the introduction of telemedicine clinics, as well as putting together a series of online webinars aimed at relaying the government information surrounding COVID-19 to provide the best possible guidance to our members amidst the uncertainty and confusion.
- The BBS clinical teams have continued to provide exceptional patient care and actively use feedback from patients to improve services, research and potential therapies going forward.
- The number of new referrals to the clinics has increased significantly from the estimated figure given to the NHS in 2010, putting a strain on the service.
- Clinical and registry data continues to show that BBS is a worldwide condition affecting all ethnicities and age groups.
- Two babies were diagnosed with BBS while still inside their mother's womb, marking incredible progress in raising awareness of the condition amongst medical staff.

- Dr Elizabeth Forsythe has been appointed as a consultant and will be returning to the adult and children's clinics in London.
- Everyone who attends a BBS clinic undergoes genetic testing, with the expectation that when genetic therapies become available, this data will be vital in determining who may benefit from treatment.
- Dr Victor Hernandez and his team have shown that gene therapy can prevent vision loss in mice with BBS1. Axovia Therapeutics was set up by Professor Phil Beales and Dr Victor Hernandez to take the therapy to market and they hope to commence human trials in late 2024.

Using Stem Cells to Study Kidney Disease in BBS

James Williams, PhD Student

James Williams, PhD student of Professor Phil Beales and Professor Fiona Watt, gave an overview of his research using BBS stem cell lines to investigate kidney disease in BBS. James explained how they turned stem cells into kidney cells, with which to investigate the causes of kidney disease, specifically in BBS10. James reported that they were able to grow kidney cells with BBS10 mutations in the lab and then give them a healthy phenotype, by inserting a circle of DNA containing a healthy copy of the BBS10 genes.



BBS Registry

Dr Elizabeth Forsythe

The BBS Registry is an electronic database that collects patients' medical information from all four of the BBS clinics in Birmingham and London.



This medical information will include: profiles of the eyes, kidneys, hormones, blood test results, medication, clinic measurements (height, weight etc.), diagnostic criteria and genetics.

The database is only accessible to those professionals who are already involved in the patient's care, and is hosted by University College, London, who comply with strict data protection measures; patient consent is required for information to be collected.

Going forward, the database will aid collaboration and research into new treatments.

Patients and those supporting them are invited to ask any questions about the BBS Registry when visiting the clinic.

LOOK UK

Elin Williams,
Mentor Development
Coordinator



LOOK UK is a charity supporting young visually impaired (VI) people and their families to thrive through mentoring, transformational events, youth forums and parent support groups.

The team are visually impaired and use their own lived experience to provide support.

Mentoring

LOOK UK now has over 100 active mentoring partnerships within three categories, Peer Mentoring, Student Mentoring (For Post 16 Education) and Career Mentoring. The mentoring programme is free of charge and offers 12-month partnerships, matched on shared interests, studying similar subjects, same eye condition, career aspirations etc.

Maria, a mentor with BBS, said the platform was safe, easy to use and the mentor role could be fitted around her life. Benefits of being a mentor include, increased employability, improved confidence, improved mental health and sense of purpose and identity.

A LOOK UK Mentee said, "My Look mentor is so helpful because he just gets it. He has been there and done that and come through the other side. Just knowing that he has made a great life for himself and is successful in his job is a great boost for me and it is reassuring for my Mum as well."

For more information, visit www.look-uk.org

Update on Setmelanotide

Dr Elizabeth Forsythe

Dr Forsythe gave an overview of the complexities of hunger within the body, and explained how different organs such as the brain and stomach, combined with different hormones maintain a balance between increasing and decreasing appetite. In BBS, the process of keeping this balance doesn't work properly, and the drug Setmelanotide has been shown in trials to correct the deficiency by controlling hunger.

Results from the clinical trial

- Decreased weight (an average of 8% body fat)
- Decreased hunger
- Increased reported quality of life

Next steps

- More trials pending in 2023
 - * Small number of participants due to very particular criteria and capacity
 - * Participants from all four BBS clinics (must be over the age of six)
 - * Weekly injections for participants instead of daily injections
 - * The clinics will approach prospective participants directly

Research into Neurodevelopmental Profiles of Children with BBS

Dr Emma McGibbon (Clinical Psychologist)
Gabby Leen (Trainee Clinical Psychologist)

Neurodevelopment is a term referring to the brain's formation of pathways that affect the development of thinking skills, social abilities, sensory skills, attention and much more. When there are differences in this development, these are called neurodevelopment differences, and these usually occur during the early stages of childhood and continue to develop into adulthood.



BBS and neurodevelopmental differences

As evidenced in recent studies such as Kerr et al, 2016, and Barnett et al., 2002, people with BBS are more likely to have neurodevelopmental differences compared to their similarly aged peers, and these can vary widely in type and severity.

- **Intellectual or Learning Disability (ID/ LD)** affects the way a person learns new things throughout their life. People with ID/ LD may find it difficult to process complicated information, to learn skills, or to look after themselves and live independently.
- **Social Communication Difficulties** affects the way someone behaves, communicates and interacts with others. People with Autism Spectrum Disorder (ASD), often have difficulties with understanding the unwritten rules of social communication and have restricted or repetitive behaviours or interests.

Why do we need more research?

There is very limited literature available with few families given the opportunity to be part of research. With a better understanding of the strengths and difficulties a child has, their support needs can be better managed and more accurate information can be given to families, clinicians, teachers and other professionals involved in the child's care.

Education, Health and Care Plans (EHCP) tend to focus on a child's medical needs, rather than any neurodevelopmental difficulties. Additional research and evidence will potentially influence EHCPs so that they support a child's neurodevelopmental difficulties more effectively.

Current research: Neurodevelopmental Profiles of Children and Young People with Bardet-Biedl Syndrome (BBS)

Study 1: Questionnaire data analysis

The findings of this study have yet to be fully analysed, however preliminary findings show that 80% of the children had difficulties with social communication as reported by a parent/carer, with over 50% reported as 'severe'.

Proposed Study 2: Neurodevelopmental assessments at GOSH

If approved, those attending clinics will be asked if they wish to take part in the study. The results would then be added to an anonymised database.

Direct assessment: Pilot Neurodevelopmental Assessment Clinic for Children with BBS

The pilot has been running for one year and has already shown that the majority of children assessed so far have learning needs that meet

criteria for an ID diagnosis, and that motor skills (fine and gross) need to be assessed for all children.

The Global BBS Community

Tim Ogden, President, BBS Family Association (USA)

Tim Ogden has a 16-year-old son with BBS and began by talking about the importance of family connection, and how he is thrilled to be able to meet in person, the wider BBS family at the conference.

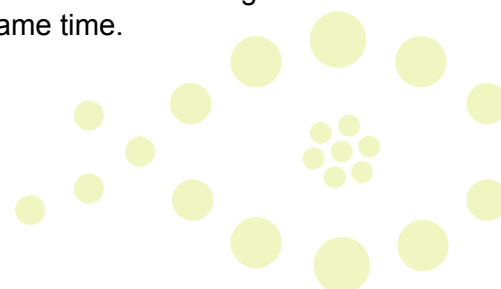
BBS groups from Europe and the US have been connecting and working together over the past couple of years which is important; even if care is given locally, research and collaboration must be global so that it can benefit everyone.

While the BBS community are incredibly grateful to researchers in the field, it is the people who are directly affected by BBS who should be driving the direction of research; for research to more quickly and thoroughly benefit BBS families, we need to coordinate globally to set the research agenda.

Every BBS organisation has things in common with one another. For example, BBS US and BBS UK both have a high number of South Asian patients. Together, they can create and share resources to make them accessible to patients who speak Urdu, Hindi etc. Similarly with research, in France they have been focusing on retinal organoids which can benefit people regardless of what type of BBS they have; in the UK, gene therapy research has been focusing on BBS1, the most common gene mutation, whereas in the US they are working on BBS10. The progress made in one part of the world will ultimately benefit those living somewhere else, and together we can continue to raise standards of care globally.

How can you collaborate globally?

BBS International's BBS Global Day is a cooperative effort, coordinated by BBS International, and with support from BBS family organisations in France, Italy, the Netherlands, the UK and North America. You can get involved on social media, fundraise, share posts, support the organisations that are leading research into treatments, and feel connected to the global community at the same time.



Guide Dogs

Kerry Kernan, National Operations Manager of Children's and Young People's Services

Guide Dogs are a registered charity offering support and information pathways to both adults and children.

Help for Adults: Information and advice, My Life Skills, My Sighted Guide, Guide Dogs

Help for Children and Families: Information and advice, My Life Skills, My Time to Play, 'CustomEyes' Books, Tech for All, family events, Guide Dogs and Buddy Dogs

In addition to the Guide Dogs service, there are many other services on offer, all of which are tailored around the needs of the individual.

Education support service: run by qualified teachers who specialise in teaching students with visual impairments, offering information, advice and guidance about school placements and

support, transition and Education, Health and Care Plans (EHCP).

Family support service: offering information and support with benefits applications, housing, social care and offering emotional support.

Technology: offering information, advice and guidance about accessing and making the best use of technology.

CustomEyes Books: offers a completely bespoke service so each child receives their books with the font size, spacing, colour and more, tailored to their needs.

Family Events: Guide Dogs Experience Days and a Family Weekend provide an opportunity to experience workshops, talk to professionals, play with technology, meet Buddy Dogs, and enjoy social activities.

www.guidedogs.org.uk
GuideLine 0800 781 1444

Katie Oates: Personal Perspective

Katie is 23 years old and was diagnosed with BBS10 when she was 5. She is registered as severely visually impaired, uses a cane, and underwent a successful kidney transplant.

Katie shared her fitness journey with the attendees at our 2022 conference. You can watch her full Personal Perspective on our YouTube channel, along with all of the other presentations from the conference.

Katie said, "during the first lockdown in 2020, I joined my Mum and brother in doing fitness classes online. It was really hard work, but I very quickly felt fitter and happier. After lockdown, I decided to employ a Personal Trainer, called Stu, to come to the house once a week for a one-to-one session. Stu sets me goals around strength, fitness, weight and body composition, which I work towards and, when I hit them, we set new ones. This keeps me really focused.

I also started an online dance class for adults with learning difficulties, which is great fun and gave me a whole new circle of friends.



And on top of that, I take our dog Rosie for long walks around the village, which I can manage fine with my eyesight.

Currently, my mum is helping me work through the NHS Couch to 5k programme which is going really well. My goal is to take part in our local Park Run and to be able to run all of it.

My weight has now dropped to 79.8 kgs, so in total I've lost 25.2kgs in about 3 years and I am in a mentally and physically better place than I ever thought I could be.

It's hard work with BBS but it's a great feeling when you start to see and feel the benefits of being fitter and stronger.

I'm loving my fitness journey and all I can say is that if I can do it, you can do it too."



Your thoughts about Conference 2022

"I thought the father telling his story about his BBS journey with his son was brilliant."

"So good to know that there is someone with real commitment there to help when one is at a loss and doesn't know where to turn."

"Always a great mixture of updates on research and personal perspectives."

"Interesting to hear about potential trials and treatments running and upcoming."

"The BBS UK team made us feel so welcome, as did the other delegates."

"It was lovely to spend informal time chatting and getting to know one another in a relaxed environment."

"Our little girl is 3 and the staff in the crèche took great care of her. She sometimes finds new surroundings difficult, but they were so kind to her and had lots of beautiful sensory toys and books. They created a lovely play environment."

"By Sunday I felt like I had really bonded with people who share similar stories and just knowing you weren't alone was so reassuring."

New BBS UK Team Member

Natalie Braunton: Operations Assistant

As everyone at BBS UK knows, Tonia Hymers has been juggling many responsibilities, and Natalie has been employed to help relieve Tonia of some of these tasks. Natalie has been working for small charities since 2016 and brings with her a wide range of administrative skills in governance, finance, membership, events and more. She has a passion for improving and simplifying processes, is a team player who gladly helps wherever she can and uses her creativity to solve problems.

Natalie lives in Wales and enjoys listening to music, sewing, playing video games, and walking her dog, Theo. Natalie says: "I've received such a warm welcome from everyone and feel really privileged to be working for BBS UK. Going forward, my aim is to relieve Tonia of some of her workload, support the Trustees, and work with the team to run another successful conference. I'm really looking forward to meeting some of you later this year!"



Fundraising Update

Nancy Carter joined the team in January and has been developing the fundraising and communications systems and processes of the Charity. Nancy will be with us until mid-May, when she will sadly be moving on to take up a position elsewhere. Nancy has been an absolute pleasure to work with and has made a significant difference in her short time here; we wish her well in her next position.

Photo shows Nancy with some of the wider team.



Trust and Foundations

We have ambitious plans for trust and foundations fundraising this year and have made one bid already, with another in the pipeline, with a number of potentially aligned trusts in our sights for the future. We have made a strong case for support with a small charitable trust, requesting a contribution to the running costs for our Advice Service and are in the process of preparing additional budget and reserve information to support that. It's not a done deal but we remain cautiously optimistic.

Small donations

Unfortunately, Amazon Smile has closed. Although that generated only a small annual amount, everything helps, and diversity in our income stream is of utmost importance, so we are disappointed to lose it. We have, however, joined 'easyfundraising' which we hope will bring in many more free donations, via a wider range of shops and organisations, from insurance companies to travel agents to supermarkets.



Easyfundraising is a user-friendly platform producing free donations from online shopping, supported by lots of well-known shops and brands. The supporter simply joins, selects BBS UK as their chosen cause and whenever they shop online at any of the providers that are signed up, a small donation is generated. There is no cost to the donor with retailers paying to be featured.

Please sign up to support us at:
www.easyfundraising.org.uk/causes/bardet-biedl-syndrome-uk/
or use the QR code. And do share whenever you feel you can.



Events



Cilia Cyclists

In September, a team of cyclists from the UK and Germany undertook an epic cycle ride to raise funds for BBS UK, and to raise awareness of the syndrome.

This incredible team were: Professor Helen May-Simera and her team of BBS researchers, Alina, Peter and Rike; Dr Beth Hoskins from ICH, London, and Steve Oates, father of two young adults with BBS. They were expertly supported by BBS UK Chair, Laura Dowswell and husband Nick. The trip took five days in total and covered approximately 420 miles and 3 countries; Steve led the group all the way from Mainz University to the Conference in Northampton, arriving on the Friday evening. Tim Ogden, president of the US BBS group flew in from the States and joined the team for the UK leg of the trip. Our heartfelt thanks to all the Cilia Cyclists and their support crew; together the team raised a fantastic £5,000!

Well Done Evie!



Young Evie Swales did a wonderful job of manning her stall at the Titus Salt Resourced Provision Christmas Fair. All the staff and pupils worked tirelessly to make this event a success and lots of fun was had on the day. This year the team voted to support BBS UK out of the proceeds and a fabulous £250 was donated!

Tony Francis smashes the Richmond Half Marathon

Tony Francis ran the Richmond Half Marathon on 9th October, he said, "I've been looking for ways to support my sister, Laura, for a while and have just been too busy/lazy to get a fundraiser going, so this was a great way to get things moving!"

Tony achieved his aim to get around the course in under 2 hours and raised an incredible sum of money for BBS UK in the process.

Through JustGiving donations from family, friends and business contacts, Tony raised a whopping £14,500 for BBS UK!



Richmond 1/2	
Tony Francis	
7974	
Richmond 1/2	
Gender:	Male
Category:	Age 30 - 34
Status:	Finished
Club:	
Add To Favorites	
Finish Time	01:57:55
Overall	421/887
Category	46/78
Gender	306/495



We Carry the Gene

WE CARRY THE GENE

Jog, walk, ride, swim or hop 20 miles to raise £10,000. Anything you can do will help to keep BBS UK running!

£10k

bbsuk To register or donate: www.justgiving.com/campaign/wecarrythegene
If you have any questions, please contact fundraising@bbsuk.org.uk

From John O'Groats to Lands' End, via Northern Ireland, Ireland and Wales, we are all coming together to walk, run, cycle, swim and ride to 'Carry the Gene' and raise funds and awareness for BBS UK. This inclusive event can be supported by anyone irrespective of ability. You could walk, run, cycle or even swim your distance; this is a virtual challenge, so you can do it on a standing bike, in your garden, or in a local park. How about hiking the length of a local river or a strolling circle of a football pitch? Whatever you can do, is just the ticket for this one!

To sign up, visit our Just Giving 'We Carry the Gene' page (or scan the QR code) and select the orange 'Start fundraising' button.

You'll be prompted to create an account where you can detail exactly how you will 'virtually' Carry the Gene. Then it's as simple as sharing that with your friends, family, work colleagues, and, well, anyone who might chuck in a little bit towards our £10,000 target. This event has launched and is running through to the end of August; if you can't take part yourself, please do support someone that is. It all counts, and it's all very much needed this year.



We Carry the Gene: Team Scotland

Team Scotland, led by **Liam and Ash Wilkie** have made an awesome start to the campaign and will physically 'Carry the Gene' baton from John O'Groats, starting in April and hand it over to Team Ireland in May. **Graeme Roache** has gotten the campaign off to a flying start, with 30+ miles completed on a tandem cycle, raising a fantastic £367. Team Scotland's total currently stands at just over **£9,000**, such great work and truly inspirational. You can check on their progress here: www.justgiving.com/crowdfunding/carrythegeneteamsotland or scan the QR code:



We Carry the Gene: Team Ireland

The BBS UK Regional Group in Ireland are delighted to be getting involved in this wonderful fundraising campaign and are currently very busy finalising preparations for 'Team Ireland'. Team Leader, **Ciaran Moy** reports, 'our plan is to physically receive the BBS UK Baton from our friends on Team Scotland in Belfast on Sunday 14th May (the morning after our 1st BBS Irish Information Day) and continue to 'Carry the Gene' Baton through Northern Ireland, crossing the border into Ireland and onward towards Dublin Port where we will then pass the Baton to our friends across the Irish Sea to continue the journey towards Lands' End.

A JustGiving page has been set up to accept donations on behalf of Team Ireland. To date, we have many volunteers and supporters that have willingly committed to individual legs of walking, running & cycling in their own time – either physically or virtually if they wish – to cover the miles, raise awareness and generate funds for this super campaign. Every single step of this journey counts - so if you are not yet involved, please do so and simply get in touch with us via the Regional Group Ireland Facebook Page if interested.

It's all about you!

We couldn't do any of this without you – the real BBS UK team. The people who fundraise day in, day out. Who throw their pounds in the pot when we shake our buckets at them and put on a BBS UK tee to run their 5Ks - thank you to each and every one of you. Every person who has donated to BBS UK, who's filled in a form for us, who's supported us at events and occasions or shared something to your network for us. You are wonderful and we genuinely could not do the work we do, without you.

Inclusive Rugby

Liam Wilkie: Trustee, BBS UK

In the summer of 2021, our family was made aware of a local rugby club running classes for all different age groups. We decided to make contact, knowing how much Caitlin would enjoy it, not to mention the benefits the exercise would bring.

We reached out to Strathmore Rugby Club and discussed Caitlin's needs. Immediately, we could tell that the club was welcoming to everyone who joined, regardless of age or ability. They were eager for Caitlin to join, and so she quickly attended her first training session for the 'Strathie Sharks'.

For the next 90 minutes of the training session, Caitlin had a huge smile on her face. The coaches were brilliant, and the other children and adults alike couldn't stop smiling and laughing throughout. From this moment we knew this was going to form a huge part of not only Caitlin's life, but the whole family as well.

The club's Business Development Consultant was kind enough to give our family a tour of the facilities, as well as tell us about the history of the club and what they hope to do in the future.

Strathmore Rugby Club run numerous groups for under 18s, as well as two men's teams and more recently a women's team. They work closely with Strathmore Community Rugby Trust, a charitable organisation that aim to operate sustainable programmes centred on our values of rugby, health and community. The Trust encourages those it works with to develop and enjoy the physical and mental health benefits of the sport, play an active part in their local community and have a lifelong involvement in rugby.



Inspired by the incredible activities of the Club and Trust, and eager to support in any way I could, I approached my employer, Kilmac Ltd, a civil engineering and groundworks contractor based in Central Scotland, and they agreed to become a sponsor of Strathie Sharks! The sponsorship money funded new strips, not only for Caitlin's group, but for every under 18s group.

With the rugby club fully aware of BBS UK and Caitlin's syndrome, they decided to surprise us by getting the kits made with the BBS UK logo on the sleeve which will help raise further awareness of the Syndrome.

Caitlin and now our other daughter Niamh are both thriving at Strathmore Rugby Club, confidently taking on any challenges they face. You never know, in a few years you may see them running onto the pitch representing Scotland!



Friends of Ferguson Heritage Raise £900

In what can only be described as a great case of community spirit, vintage tractor enthusiasts travelled 22 miles across the Scottish Borders to raise money for BBS UK.

The annual event which was started 17 years ago in the Borders by the late Mr Robert Gourlay as a way of getting enthusiasts and their prized tractors out and about on a Sunday, has gathered momentum over the years resulting in a major community event with followers who use their hobby to do good.

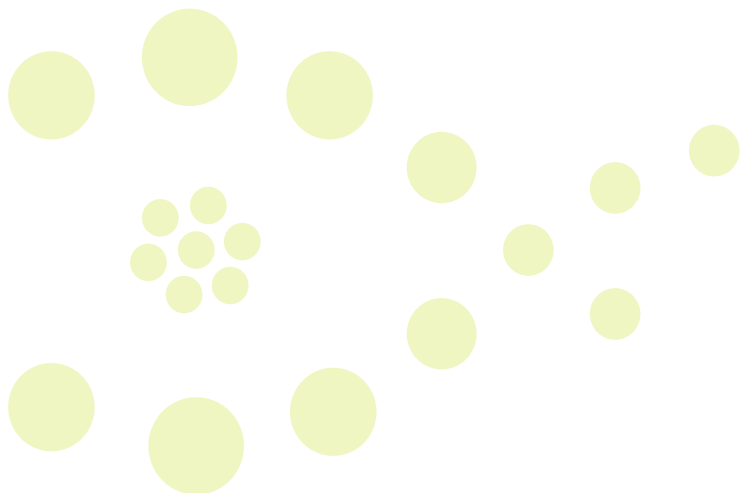
Every year the group, led by Chairman Rob Snowden, aims to raise money for a different charity. The charity is usually chosen due to a connection with someone in the local community



and the decision to raise money for BBS UK was inspired by local teen Jemima, who was diagnosed with BBS in 2018 and who is now registered severely visually impaired. It is her 'strong, brave and lovable character' and 'courageous and witty take on life' that makes her a 'pioneer in raising awareness of BBS in Scotland and the rest of the UK', says her proud mum, Jayne.

When the Friends of Ferguson Heritage heard about Jemima's diagnosis, they decided to dedicate their 17th Annual Run to BBS UK and raised an astounding £900 from the event, as well as raising awareness.

Jemima, her brothers and her team of supporters now wish to further raise awareness of BBS with other events, and are proud to be supporting Team Scotland and the Carry the Gene Challenge in May 2023.



BBS Clinics Update

Birmingham Children's Hospital

Birmingham Children's Hospital are pleased to announce the funding of additional telemedicine clinics (clinics held via video link) for children attending the BBS service. These will run in addition to the telemedicine clinics they are already offering and are an opportunity for clinical teams to assess patients and for them to answer questions on symptoms and treatments in between face-to-face clinic appointments.

In recent years the time between face-to-face clinic appointments at Birmingham Childrens Hospital has increased from 12 months to 18 months or two years due to the ever-increasing demands on the service.

By adding more telemedicine clinics the team can offer children a review appointment every year which will be virtual or face-to-face. If you have attended a telemedicine clinic in one year, then your next clinic will be face to face.

The telemedicine clinic will be with the Consultant, Clinical Nurse specialist and Dietitian and the team will only offer this type of clinic appointment if it is clinically safe to do so. Your local healthcare team will be asked to arrange yearly blood and urine samples locally prior to these appointments taking place so the team have all the information they need to hand.

Over the last year the children's clinic in Birmingham has had:

- 15+ new referrals.
- 8 face to face clinics
- An increase in virtual appointments to 8 per year
- 12–16 month follow-up appointments

Clare Stevens returned to her role as a Rare Diabetes Care Coordinator alongside Lindsay Wilson. Clare and Lindsay are the first point of contact for families to liaise with the hospital and medical team. Clare and Lindsay can be contacted on **0121 333 9999** or via email: bwc.rarediabetes@nhs.net

Talia Treen has joined the team as Eye Clinic Liaison Officer and is available to speak to our families during the clinic day.

Queen Elizabeth Hospital

With Covid-19 restrictions eased, we are once again spending our lunchtimes together in the resource room, allowing patients, their families

and/or carers a relaxed environment in which to chat and spend time together.

Over the last year the adult's clinic in Birmingham:

- has increased the number of virtual clinics to 10 per week
- is currently offering 12–18 month follow-up appointments

Maureen Higgins has joined the QEH admin team as an Administrator and BBS Co-ordinator and is the first point of contact for patients contacting the hospital. Maureen can be contacted on **0121 371 8705** or via email: renalBBSreferral@uhb.nhs.uk

Great Ormond Street Hospital Guys & St Thomas' Trust

We are all very pleased to welcome back Dr Elizabeth Forsythe to the BBS services at St Thomas' and Great Ormond Street Hospitals. Elizabeth has been away for a few years completing her PhD, and training to become a consultant. Elizabeth has always remained very involved with the BBS service, however its fantastic to have her back in clinic again.

Clinics have been very busy and we continue to run both our 'in-person' and telemedicine clinics at full capacity; we have welcomed 8 new patients into our adults' service and 10 into our children's service. We hope they feel better supported and informed about their diagnosis after visiting us in clinic, and learning about the services and support available at BBS UK. We know there are some new patients and families waiting to have their referrals processed and we look forward to meeting them in clinic soon.

Our sincere thanks go to everyone involved in the BBS clinic day. We are so lucky to have an amazing team of clinicians that are always so generous with their time and knowledge. A big shoutout must go to our clinical nurse specialists and clinic administrators; they are all so supportive and patient and the service just would not run without them. Thank You All - you are our BBS heroes!

Amy Clapp, Patient Liaison Officer, Birmingham
07591 206787 or amy.clapp@bbsuk.org.uk

Angela Scudder, Patient Liaison Officer, London
07591 206788 or angela.scudder@bbsuk.org.uk

BBS UK Advice Service Update

By Rebecca Perfect, Advice Service Manager

Last year, BBS UK partnered with London Met University and Sam, our first student social worker joined our team at the end of October 2023. Sam has settled into the team well and has enjoyed getting to know our service users, by supporting the BBS clinics and working on cases for the Advice Service. Sam will be with us until the beginning of April and has been a real asset. We hope that Sam will be the first of many students to come and work with us. It's great to get fresh perspectives on the way that we work, and to contribute in a small way towards ensuring the next generation of social workers have some knowledge of BBS.

The work of the Advice Service is varied; following, is a snapshot of how we have been supporting our service users.

A common enquiry is from people looking for support into employment. Whilst we can't offer employment support ourselves, we are aware of and can signpost to agencies both nationally and locally who do support adults with disabilities into employment. We are continuing to monitor how our service users are getting along with these employment support projects and hope to hear of some successful placements soon. In the meantime, we are gaining valuable insight into which projects best meet the needs of those with BBS, which in turn will enable us to signpost others to more appropriate support.

In recent months our work in Advice has included supporting parents to ensure their children's Education Health and Care Plans are fit for purpose and to ensure the child receives an early review if they are having difficulties within their current provision.

We have supported adult service users to move across from the old style DLA benefits to Personal Independence Payment (PIP), completing forms and helping to gather evidence as needed. Our knowledge and experience of BBS and PIP has ensured that benefits levels have been maintained.

We are excited that some of our adult service users have recently moved into their own homes and we continue to support others to explore



independent and supported living options. Our remit includes working with other professionals to ensure our service users receive the right support following their move.

Unfortunately, we are finding that more and more people are struggling to secure an assessment of need from their local authority and to access the support that they are entitled to. An ongoing and significant aspect of our work will be to hold local authorities to account and ensure they meet their legal obligation to complete assessments of need for our service users.

Both the Advice Service and clinics team have noticed an increase in requests from schools for staff training about BBS and this is something we will look into over the coming year. In the meantime we have information leaflets and other resources which we circulate, and we are always happy to offer virtual, and where possible, face to face support for schools to ensure children receive the best support possible.

The need for our Advice Service is clear and this need is going to increase. In recent months we have sadly had to introduce a waiting list, because so many people have needed our help, however we try and manage this so that we can respond to both urgent and non-urgent requests for support. In the coming weeks and months we will be seeking funding and encouraging fundraising so we can continue to support our service users when they need us most.

As always, thank you to all our members for your support and if you need our help, please do not hesitate to contact us.

Friends of BBS UK

Your contribution, however small, makes a huge difference to the individuals and families we help and enables us to continue to sustain a high level of support. Examples of what we have achieved over the last 12 months includes:

- Provision of an Advice Service and Clinics Support Service offering support for families with accessing local authority support, benefits and housing applications and Education, Health and Care Plans
- Provision of newsletters and information booklets
- Support for BBS research programmes including those looking at mental health and wellbeing, and kidney function in BBS
- Organised subsidised events including the annual conference, activity and social events
- Support for newly diagnosed families with information and regular contact

How to become a Friend of BBS UK?

Complete the 'Become a Friend of BBS UK' online form which can be found on our website: www.bbsuk.org.uk/friends-of-bbs-uk/

If you want to make a **regular donation**, a standing order can be set up via your banking app, your online banking website, or you can visit your branch in person.

Our bank details are:

Bardet-Biedl Syndrome UK (BBS UK)
Acc. No: 20368253
Sort code: 20-19-97

- ✓ Please remember to use your name as the reference so we can allocate your donation and know who to thank!
- ✓ There is no minimum, or maximum amount that you can donate each month.
- ✓ If at any time you find the regular donation challenging or your circumstances change, you can reduce or increase the amount of your donation. **You are in control.**



If you have any questions, please contact Natalie Braunton, Operations Assistant at admin@bbsuk.org.uk or 07784 922654



Eat Healthy, Save Money: Five Top Tips!

Sarah Flack

Principal Dietitian, Great Ormond Street Hospital for Children

Food prices have risen dramatically over the last few months and still seem to be increasing every week. Understandably, eating healthily may feel even more challenging now so read on for some tips that should help to cut your shopping bill a little.

Plan your main meals and shopping

This may seem like a lot of effort but from personal experience it is well worth it! My weekly meal plan keeps us on track throughout the week and has reduced our food waste. If you need some inspiration to help you get started, try looking at the cheap and healthy recipe ideas on the BBC Good Food website or check out the NHS Healthier Families recipe section:

www.bbcgoodfood.com/recipes/collection/cheap-and-healthy-recipes

www.nhs.uk/healthier-families/recipes/

Save on your '5-a-day'

Choose seasonal fruits and vegetables as they will be most widely available and so best priced, for example, broccoli, carrots, new potatoes, spring greens and spring onions are in season in May.

In shops, loose fruit or vegetables are often cheaper than pre-packaged; choosing these will only add a few minutes to your shopping trip. Local markets can be a great way to buy local produce at a good price. Look out for special deals in your local supermarket of fruit and vegetable



boxes that contain either unusually sized fruits and vegetables or extra stock they want to sell to reduce their food wastage.

Tinned fruit and vegetables can be an easy, convenient, and usually a cheaper way to add a portion to your meal. If possible, choose fruit in juice, and vegetables tinned in water without salt. Frozen fruits and vegetables are normally cheaper. They are a nutritious choice and ready prepared so easy to add into your meals!

If you have a garden, you could try growing your own vegetables. The National Trust website has an informative section on seasonal food to help you plan your gardening or help you shop seasonally:

www.nationaltrust.org.uk/discover/gardening-tips/guide-to-seasonal-food

Get cooking!

Cooking meals from scratch can be an effective way to save money. A tomato-based pasta sauce can cost 2 to 3 times the price of a tin of chopped tomatoes. The tinned tomatoes also contain significantly less calories, so this swap is a win! Use less meat and add extra vegetables or pulses (peas, beans and lentils) to produce a satisfying meal with less cost. Chicken pieces with bones and skin-on is cheaper than skinless, boneless chicken so look out for the best value option. Try to remember to take the skin off before cooking! Tinned fish or frozen fish is typically cheaper than fresh fish.

Try making your favourite take-away – there are lots of fake-away recipes to help you save money and often produce a healthier meal. Need some inspiration to help you get started – have a look at the following websites:

lets get cooking.org' with <https://www.slimmingworld.co.uk/blog/a-taste-of-slimming-world-fakeaways/>

Healthy Snack Choices

Sometimes offers on snack foods appear amazing value but often there are good value, healthier alternatives.

Bananas, seedless oranges and apples are great fruit choices to take out with you for a snack. At home you could try keeping a box of carrot, celery, and pepper sticks in the fridge to eat at snack time. Children can easily help themselves to this snack – try serving with a tablespoon of low-fat dip for a more exciting snack!

Making your own popcorn in a saucepan, microwave or a popcorn maker is a lower cost snack choice – try plain or add a sprinkle of salt or icing sugar.



Energy-efficient cooking

Think about how you will cook your meals when you are menu planning so that you're using each gadget most efficiently. Try having a side salad rather than garlic bread with your pasta dish. This simple change means you only need to use your hob, save a few calories, and get closer to your '5-a-day'!

Check what kitchen gadgets you have hidden in your cupboards as slow cookers, air fryers, and electric griddles can all be energy-saving gadgets. Remember a microwave oven is an efficient way to heat and cook food too. Cook as much as possible in your oven at the same time so you use all the available space to maximise the use of the heat. Batch-cooking meals is a good option if you have the time – freezing extra for another time and then reheating in the microwave. Defrost food in your fridge overnight so you can save on the energy costs of using the microwave to defrost and also shorten the main cooking time.

Remember to talk to your clinic dietitian at your next appointment for further advice.

Sources of Support:

If you are struggling in this cost-of-living crisis please ask for help and support from the BBS UK clinics team who are always amazing!

If you are pregnant or have young children you may be eligible for Healthy Start Vouchers to buy cow's milk, fruit, vegetables, pulses or infant formula.

<https://www.healthystart.nhs.uk/how-to-apply/>

Food banks can also provide invaluable support if you are struggling to buy food, but most need a referral from a school, health care professional or a social worker – the following websites can help you find your local food bank:

<https://www.trusselltrust.org/get-help/find-a-foodbank/>

<https://www.salvationarmy.org.uk/foodbanks>



Weight Loss with Slimming World

This article has been reprinted with permission from Slimming World. It was originally posted on their website on 15th February 2023.

Helen Rourke, from Cradley Heath, felt like she couldn't lose weight due to her diagnosis of Bardet- Biedl Syndrome (BBS). She has now lost more than 5 stone with Slimming World and feels happier and healthier.

A recent service evaluation, undertaken by Slimming World, shows those with BBS who joined a Slimming World group through a referral programme and attended nine or more of the sessions, on average, lost 4% of their body weight over 12 weeks and reduced their BMI.

BBS UK offers its members 12-weeks free membership to Slimming World to help those who wish to access support in losing weight. Helen, 50 says:

"In August 2021 I joined Slimming World weighing 19st 2.5lbs. I spotted a post on the BBS UK Facebook page for free membership to Slimming World for 12 weeks. I've tried several methods to lose weight in the past, and they've never really



worked. My mum encouraged me to give Slimming World a go and so I walked into my first group at the end of the summer, and I've not looked back! My Consultant Maddie was lovely and the other members really welcoming. My mum came along to support me, and we both really enjoyed the session."

"Since losing weight, as well as needing a new wardrobe because my clothes no longer fit, I've found moving is a lot easier. Previously, when I've gone for a walk my mum has had to guide me, but now I'm the one with the quicker pace, walking ahead of her. I'm part of a singing group and love being on the stage performing – now I've lost weight I'm so much more confident and happy in myself. Because of BBS, I didn't think I could lose weight, but joining Slimming World has been the best thing I could have done to improve my health!"

Registered dietitian Jennifer Kent, from Slimming World's Nutrition, Research and Health Team, says:

"Helen's weight loss success backs up our findings which show that people with BBS, who were referred to Slimming World, successfully lost weight during their 12-week referral, with more than a quarter losing a clinically significant amount of weight during this time."

"Slimming World's healthy eating plan is flexible, takes into account individual needs and preferences and encourages members to fill up on those foods that are naturally lower in energy density (calories per gram) and also highly satisfying, while limiting foods that are highest in fat and sugar and are less filling. This means they can lose weight without feeling hungry and reduce their calorie intake without needing to count them." There are a limited number of free places remaining; to access the scheme simply call the Slimming World on Referral team via the following number which is specifically for BBS members:
01773 302475

Asian Food on the Slimming World Plan

We have been asked whether the Slimming World programme is suitable for a South Asian diet and the answer is a resounding YES!. The Slimming World magazine frequently features recipes and articles from various cultures. Azra Bashir lost 4st over 15 months and wrote, 'I didn't think Slimming World would fit in with my life and I was worried I wouldn't be able to eat Asian food but it fits perfectly. The basic concept of the plan is making

healthy meals from scratch and that's the Asian way – finding fresh ingredients and spices and putting them all together to make beautiful food'.

The Slimming World website includes really useful information and recipes about how to tailor the plan to suit individual needs and any religious or cultural considerations for example during the special month of Ramadan.

The Blind Warrior: Shodan to Nidan

Richard Parker; Roger Thyer-Jones; Drina & Michael Parker

Richard Parker began his Seido Karate journey back in 2008, at the age of 27. Roger Thyer-Jones, 6th Dan from the World Seido Karate Organisation in the UK had experience of teaching visually impaired students and so began a partnership that was to take Richard all the way to Black Belt Nidan. Richard, Roger, and Drina and Michael (Richard's parents) told us about Richard's journey to Nidan:

Roger: I was hard on Richard. I expected the best that he could achieve in the sessions and made no excuses for him. We had to build a firm foundation if he was to progress. However, I knew that whatever I required him to do, he had suffered worse throughout his life. He had had to deal with school bullies, manage his own self-worth as he grew older knowing that he could never fully engage in his family's world except on his own terms. I knew that he was tough-minded and even with the support of his family, he had had to be locked in his own mind coping the best way he could. The karate training that he was experiencing was nothing compared to dealing with endless hospital visits and the slow progression of sight loss at a young age. Richard already had taken the path of the Way of the Warrior, he just didn't know it. I was simply a guide on the new path that he had chosen."

One of my favourite films is based on an old Japanese movie series depicting the life of Zatoichi-Ichi the blind monk. Ichi san carried a sword hidden in his walking stick and roamed feudal Japan righting wrongs and defending the weak from bullies. The Japanese loved his



character and did not view his lack of sight as a hindrance to what he was capable of. He was a martial artist. The idea of him being able not only to defend himself, but to defend others who were sighted, gave the series an unexpected twist. Difference definitely did not mean deficit.

I had great respect for Richard's loss of sight but at no time during our relationship did I ever pity him. I treated him like any other student and it was my responsibility to find the key that unlocked his abilities. It was his responsibility to keep an open mind and practice the drills that I gave him so that we could build a firm foundation for advancement.

A true partnership.

Has it been tough for Richard? Yes. Besides laughter we had tears of frustration, melt downs when he just couldn't reproduce a technique to the standard required, but not once in our relationship did I patronise him. My favourite saying is: *even monkeys fall out of trees*. Expect to make mistakes but learn from them.

Drina: When Richard achieved his Black Belt Shodan, First Dan, we were very proud but did not for a moment imagine that he could go beyond this amazing achievement. However, at the end of last year that is exactly what he did when he went through the rigorous grading to be awarded his Black Belt Nidan (Second Dan).

Nidan is a difficult enough preparation process for the able-bodied candidates to go through, those without Richard's challenges. The year of building up to his grading was particularly tough for Richard. He had written an essay as part of his preparation for his Shodan when he had written about his life's experiences which he didn't find too daunting. For Nidan, he had to write on the subject of 'Balance' with particular reference to how Seido Karate impacted on balancing his life. Richard struggled with the concept until a light bulb moment when he realised that balance can be applied to every aspect of life – even his beloved football team!"

The physical training and the early morning sessions with other Black Belts were tough. Dojos were just opening up after the lockdown during Covid so it was a novel experience to be back training with real live people rather than on Zoom. It was more exacting too. His teacher and mentor, Sei Shihan Roger Thyer-Jones, gave Richard confidence in his ability and patiently guided him throughout the year.

Richard: The first part of my Seido Karate Nidan Black Belt promotion was to do a series of kata. These are set patterns of movements, variations of which are used in all martial arts. The order and exact form of movement have to be memorised. Since I am blind, unlike others in my dojo, I have to try to picture the shapes and can't copy from my teacher or other trainees. I was instructed to perform many kata, chosen by the examiners and then I could choose my favourite kata: I chose to do the breathing one, called Sanshin, which translates into three battles signifying the unification of the mind, body and spirit."



I then had to talk, uninterrupted, for four minutes about my essay which was on the subject of balance and afterwards answer questions about it from all members of the dojo. I found it very difficult to write the essay and even more difficult to talk about it, but I had practised with my parents and, in the end, I went with the flow and just thought of it as if I was talking normally down the pub or at football. It went very well and I wasn't given questions that were too difficult, so I was able to answer them without getting too nervous.

The second formal part of the grading was a series of different fighting moves using weapons called a bo and a jo. These were new to me until I started training for my Nidan promotion. Learning to use these weapons was very difficult to start with, but I had lots of practice and eventually got the hang of it.

The final part of the grading was a long series of karate fighting bouts called kumite, during which I wore protective fighting gear and boxing gloves. This seemed to go on for hours but it was great fun to let off steam and go for it. I felt very proud when Sei Shihan Roger presented me with my Nidan Black Belt after which we all went to the nearby pub to celebrate.



Drina: From a parent's perspective it

was emotional during kumite, the fighting parts of the challenge, to see Richard stretching every sinew with fully-focused concentration. He had to locate his opponent (with the help of a tambourine being activated by another member of the dojo) and strike with his bo or jo, the long sticks used as weapons and introduced at this stage. His concentration and memory were further called upon when performing a series of defensive Kata involving complex moves and patterns.

To add to the mix of stress, Richard had Covid between his first and second grading sessions but he refused to be daunted by this and remained determined to succeed.

Both Michael and I were willing every fighting bout to be the last one but even though Richard was physically exhausted and pouring sweat at the end of the fighting component of his grading, he did not falter in his ambition to succeed. Throughout both grading sessions, the other members of the dojo shouted encouragement which lifted Richard's spirit and helped him to keep going.

His forays with his bo gave rise to some anxiety in his opponents who were fortunately well-protected by padded head gear! One of the most advanced Black Belts complimented Richard's perfect hand positions and finely tuned grip

adjustments. He also commented that Richard's punches were among the most powerful in the dojo!"

Roger: Richard has now achieved what he once thought was impossible. His first degree black belt, Shodan, which he gained pre-Covid lockdown, involved not only demanding technical ability but true grit in having to complete 40 fights against able-bodied opponents. His second degree black belt, Nidan, which he achieved in November 2022, involved the complex use of weapons as well as demanding self-defences, techniques that involved 360 degree turns and patterns of over 100 techniques to remember and reproduce. He was confident enough to stand in front of a class of his peers and instructors and lead the exercises giving clear commands. In his essay, which is part of the black belt process, he wrote clearly on the difficult subject of balance, which he applied to every aspect of his life.

He is not now the same Richard that set out so many years ago, stamping his feet on my studio floor, out of balance and out of breath. We are proud that he is an important part of the UK World Seido Karate Organisation.

He is an inspiration to students of every ability.

Our blind warrior.

BBS UK Regional Group Ireland Update

By Lindsay McKnight

In Ireland there has been very little knowledge or understanding of BBS both in the medical profession and support frameworks for families who receive a diagnosis. With the work of BBS UK and the families within the group, we aim to change this by joining together and making our voices heard.

On 3rd November 2021, two members from the BBS UK team and four parents of children with BBS, held their first Zoom meeting to discuss the possibility of formally setting up the group. It was unanimously agreed that the need was there as the lack of local knowledge and understanding of BBS was having a detrimental

impact on our daily lives.

In June 2022, the core leadership group was set up to run the Regional Group. This group consists of 11 individuals with lived experience of BBS, either as parents of affected children or adults living with the syndrome; members are from a wide geographical spread from Northern and Southern Ireland. The group has been instrumental in getting the forthcoming 'Information Day' off the ground and has been a pillar of support for all our patients and families in Ireland.

From early 2022, monthly Zoom meetings were

opened up and soon we were working with over 30 patients and their families giving the opportunity to support each other and share vital information. Facebook and WhatsApp groups have been set up to facilitate these connections and support.

Two 'Meet Up' days were organised; the first being in Portlaoise and the second in Belfast. Both were well attended and highlighted the growing need to have a day where we could all come together.

Ireland Information Day

On Saturday 13th May 2023, patients, families and professionals from North and South and the UK will come together at the Dunsilly Hotel in Antrim, Northern Ireland to learn, connect and raise awareness of BBS. There will be

representatives from the RNIB, NCBI, Sense NI, Autism NI and professionals from the BBS Clinics medical team.

Connections have also been built with some Scottish families through the fundraising initiative 'We Carry The Gene'. Plans are set in motion for a 'handover' of the baton from Scotland to Ireland which we hope will take place on Sunday 14th May, the morning after the Information Day. We hope to have some tandem cyclists who will kick-off the Ireland leg of the journey. The baton will then travel the length of Ireland before being passed over to Wales.

None of this would be possible without the continuous dedication and support from the BBS UK team and we look forward to welcoming them to Ireland in May!

Adam Crème: Personal Perspective

Adam Crème is a husband to Danielle and father to Zach and Tyler. He shared his story at the BBS UK Conference in September 2022.

Hearing or thinking about BBS filled me with fear and anxiety up until very recently. Even though it's been a long time since Zach's diagnosis, today is the first conference we've attended. I wasn't sure what effect sharing my personal perspective would have, but my fears were superseded by my overwhelming desire to try and help others avoid some of the pain my wife and I have felt over the years.

We were filled with excitement when Danielle became pregnant and never contemplated there being any health issues with our baby. Even after a Professor was called in to an appointment and asked us whether polydactyly was common in our families, which it wasn't. We enjoyed preparing for Zach to arrive with the usual antenatal classes and copious amounts of baby shopping.

When Zach was born, he was given a clean bill of health and referred to GOSH to have his extra fingers monitored and removed. Zach didn't learn quickly, and his speech and movement development was slow. He loved food and



put weight on easily. We thought nothing of it because all babies develop at different stages, don't they?

After a routine optical appointment, I was given the devastating news that Zach had Retinitis Pigmentosa and that Zach would probably lose his sight completely. That evening was a total blur. We cried, we showered Zach with more love and cuddles than ever before, cried some more and put him to bed. We then opened the alcohol cabinet to try to get some relief from the pain, but it had no effect.

Why us? Why Zach? It's not fair.

We decided to only tell our immediate family. For me it was not about shame, it was about allowing Zach to have the most normal childhood as possible, without people treating him differently. This decision had a huge impact on myself and Danielle's mental health.

It was in the early years that I suffered with my first bout of depression. I sought professional help through medication and counselling, and eventually after several severe relapses and different counsellors, I came to terms with Zach's condition, and my own issues.

But everyone is different. My wife has been unable to talk to professionals and deals with it her own way. We're both different people now. From fun loving and carefree to a couple with a weight on our shoulders and a dark shadow looming over us.

A gene specialist referred us to Professor Phil Beales at GOSH who first introduced us to Bardet-Biedl syndrome. He explained that every child with BBS was different, with varying symptoms and that there was no timescale that could be accurately applied to when or if these deteriorations would occur. When Danielle became pregnant with Tyler, there was no way for us to know if he would also be born with BBS as the mutated gene within Zach hadn't been identified yet. It took 7 years before we knew which gene was mutated.

We got excited over potential treatments and therapies we researched online, but this was soon replaced by dread after reading contradictory research. Danielle became obsessive about Zach's food, spending hours reading labels in the supermarket for the lowest fat treats. We were determined to teach him to eat healthily, but I felt terrible that he couldn't have the same treats as other kids.

Zach finished primary school without the need for an EHC plan although we spent a considerable amount of money on private speech therapy lessons. When Zach

started secondary school, he needed additional help, so Angela at BBS UK provided us with great support on the long and arduous journey of getting an EHC plan. Initially, Zach was incredibly reluctant to make use of the teachers assistant, however over time he has appreciated their help in ensuring he stays in mainstream education. He performed amazingly in his GCSEs and is currently in his final year studying for a BTEC in Business.

His vision remained OK, but he suffered from night blindness from an early age. It didn't seem to bother him but it gave me incredible anxiety to think that one day he might not see at all. The kids thought it was great that we skipped the queues at Disneyland with an accessible pass and went on every single ride in one day. However, I longed to be stuck in 2-hour queues like everyone else. We've since learnt to accept the benefits, such as free carer tickets and being fast tracked at airports.

Zach never asks any questions about his condition, even during our frequent trips to the BBS clinic or to his ophthalmology appointments. He never complains about all the tests, except for the eye drops. Zach has always been a content and happy child. He loves football, plays FIFA, and going to watch West Ham every week. But I'm always concerned that the things he loves so much will one day be taken from him.

Zach started using a cane two years ago, and I was amazed that my inbuilt repulsion of seeing a cane disappeared as soon as I could see how much it helped him. We could no longer hide the fact that Zach had a vision impairment and as a



result became more open to confiding in friends. People know Zach has a syndrome, but I've not told them the name or any of the other more personal symptoms. He's a young man now after all.

The fitness study Zach participated in was a huge benefit. We were so lucky to be paired with Steve Berman at GOSH, who bonded with Zach immediately. It was a total no brainer for us to continue on with his personal training. Zach has also been playing football for an inclusive team for over 5 years and I'm now one of the coaches. He has grown up with our favourite word "moderation", and leaves food on his plate when he's full, self-regulates, and has an incredibly low BMI. We're so proud of him for that.

When Zach was 12 and Tyler was 10, Tyler overtook him physically and mentally which caused some issues amongst the family. A few weeks ago, we finally told Tyler everything about Zach's condition. This was one of the reasons that we decided to finally come to conference so that he could talk to other siblings who would understand him.

If I'm honest, we approached all the BBS clinic appointments with dread, praying for no deterioration or added complications. Tonia and Angela at BBS UK, and Kath and the team of doctors at GOSH have always been so welcoming and supportive. I'm ashamed to say that we've avoided other families with kids with BBS. This isn't because we're unsocial or uncaring but due to fact that by talking to others we would have to accept the reality that Zach has BBS. I think we're past that stage now.

It's been a journey that none of the family wanted, but I'm finally at the stage of acceptance where I'm ready to move forward. Hopefully during the conference, I can gain information to help us plan the family's future, Zach's further education, a job, and the rest of his life.

In typical Zach style, he didn't ask many questions when we talked about BBS and the conference. I hope that attending the conference will help him on his own path to educating himself. Zach is an amazing young man that surprises me daily and I'm looking forward to him surprising me much more in the future.

Campaigning and Volunteering for the Visually Impaired

Sarah Leadbetter, National Campaigns Officer, National Federation of the Blind of the UK

Sarah Leadbetter lives in Narborough, Leicestershire and ran her own holistic therapies business for 18 years until she had to retire at the age of 42 due to difficulties with BBS. She trained with a long white cane in 2010 while being on the waiting list for Kiki, her first Guide Dog. With Nellie, her current Guide Dog, she likes to travel, go to the theatre, take part in lots of social activities with friends and colleagues, and being part of local community groups.

Volunteering

Sarah started to volunteer for Vista, her local blind society, and got involved in fundraising, speaking to groups for Leicestershire and Rutland branch of Guide dogs and for the RNIB in the East Midlands.



Campaigning

Sarah says she 'campaigns and speaks up as many people do not have a voice and their access needs go unheard, leaving them isolated and disconnected from their local communities.'

Sarah has been a member of NFBUK (National

Federation of the Blind of the UK), playing an active role in their campaigns, as well as many other national and local campaigns about issues. Sarah was awarded 'Campaigner of the Year' by RNIB after successfully taking legal action against the UK Government about inaccessible health information during COVID-19. She also took legal action with other disability campaigners to force the Department for Transport to undertake the national consultation on pavement parking.

Media appearances

Sarah has appeared on national and regional TV, such as 'Rip Off Britain' to talk about the effect of parking on pavements on pedestrian accessibility, BBC Breakfast, BBC East Midlands, ITV Regional News and Channel 5 News. She has also spoken on the radio and written several press pieces. Sarah is 3 years in remission for cancer and is now a campaigns champion and cancer voices volunteer for Macmillan Cancer Support.

A word from BBS UK: Here at BBS UK, we are inspired and motivated by the actions of Sarah, in raising awareness, campaigning for change,

and volunteering her time to help others. You too can get involved in local and national campaigns and volunteering opportunities; here are some resources to get you started:

- Engage with government: www.gov.uk/government/get-involved
- Transport For All: www.transportforall.org.uk/campaigns-and-research
- RNIB: www.rnib.org.uk/our-campaigns/
- Sight Loss Councils: www.sightlosscouncils.org.uk/campaigns/
- Volunteering Matters: www.volunteermatters.org.uk/
- LOOK UK: www.look-uk.org/volunteer/
- Possability People: www.possabilitypeople.org.uk/support-us/volunteer/what-we-offer-in-return/
- Thomas Pocklington Trust: www.pocklington-trust.org.uk/priority-national-campaigns/
- Volunteer

Sight Village 2023



Queen Alexandra College (QAC) are running their annual Sight Village events in Birmingham, Exeter, London and Cardiff.

What is Sight Village?

Sight Village is an exhibition for people of all ages living with varying degrees of sight loss, professionals supporting and advising VIP's and for all businesses and other organisations wishing to improve their services for the visually impaired. Exhibitors display their product/services at tables where attendees can freely talk to the exhibitors, view the information they are displaying and ask questions to learn more. Food and drinks are available to buy at the venues, so you can have a rest, talk to other

attendees and perhaps even spread the word about BBS UK over a sandwich!

How to book

The events are held in Cardiff, London, Exeter and Birmingham and are **free to attend**, so if you're interested, book your place by visiting www.qac.ac.uk/exhibitions.htm

Sight Village Central

Eastside Rooms, 2 Woodcock Street, Birmingham

17th July – Tuesday 18th July 2023

BBS UK will be exhibiting across the two day event in Birmingham in July and welcome any volunteers who would like to assist at our exhibition table. Please contact Natalie by sending an email to admin@bbsuk.org.uk or calling 07784 922654

BBS UK would love to know if any of our readers attended the events, so please tag us on social media, take photos, or even write a review that we could use in future newsletters to let others know what it was like to attend.

BBS Global Day

BBS Global Day is an annual global event, coordinated by BBS International, and with support from BBS family organisations in France, Italy, the Netherlands, the UK and North America. Supporters can raise awareness online or in person, fundraise, build connections, all while helping to accelerate research into potential therapies and treatments. We will be celebrating BBS Global Day during the weekend of our conference, across the 23rd-24th September 2023.



Anne McCloskey, a member of the Ireland Regional Group was actively supporting BBS Global Day on Saturday, the 24th of September.

“We decided to mark the occasion by raising money

for BBS UK. The Charity has been instrumental in setting up a Regional Group for individuals with BBS and their families living in Ireland. This

time last year, we knew of only one other person in Ireland with BBS. Now, thanks to BBS UK, we have met several families - both in person and online via regular Zoom calls. We were also lucky enough to attend Conference this year in Northampton for the first time and were able to meet the fantastic team and see how much they do to help the BBS community in the UK and internationally.”

“I set up a fundraising page on Facebook and was thrilled to receive donations from friends and family in Ireland, England and even Australia. My daughter Amy has BBS1 and cannot walk herself yet, so she joined me in her buggy. We walked around our local area in Cork. Fitness is not my strong point, so we walked the 10 miles at a leisurely pace over the weekend. Thankfully the sun was shining which made it even more enjoyable. It felt great to get involved and try to contribute to such a wonderful cause. Amy is my favourite little walking buddy, and we’ll definitely try to do something like this again soon.”

Thanks Anne, what a great idea!



Sense Professionals' Open Day

Amy Clapp, BBS UK Patient Liaison Officer

I was invited to attend the Sense Professionals Open Day at TouchBase Pears in Selly Oak, to get an update and overview of the services and facilities that Sense has to offer and the support they provide to children and adults with complex needs.

Sense has an incredible amount of services on offer, not just in Birmingham but nationwide which I would like to share with you:

- **Siblings & Young Carers (5-18yrs):** providing online and in-person meet-ups with activities for wellbeing support and developing friendships.

- **Virtual Buddying (all ages):** matching anyone with a disability to a volunteer who shares the same interests and hobbies, meeting an hour a week for a minimum of 3 months.
- **Support for Disabled Adults:** residential care and day services for disabled adults.
- **Sense Holidays (all age groups):** accessible holidays for children, young people, and adults both in the UK and abroad. Upcoming trips include a canal boats tour from Hemel Hempstead, exploring the outdoors at Rosliston Forestry centre, action packed trip to Avon Tyrell activity centre and a trip to Holland.

- **Arts and Sports:** available at Touch Base Pears, in the community and online, including 1:1 music studio sessions, rock climbing, seat pilates, boxing, tennis, street dance, arts and craft, body massage, gardening... the list is endless.

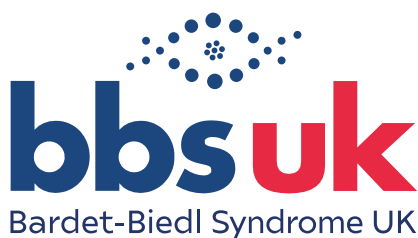
Sense can also provide support and guidance with benefits and money, care assessments and personal budgets, education, transition, planning for your future and adult life. Support for adults is available in the form of a day care service

which is available in all centres with a range of activities. Community support is available from support workers who can assist with weekly shops, accessing the local community and building friendships at social events.

BBS UK can make referrals to Sense on behalf of our service users or contact can be made directly.

www.sense.org.uk

0300 330 9256 (Mon-Fri 9am-5pm)



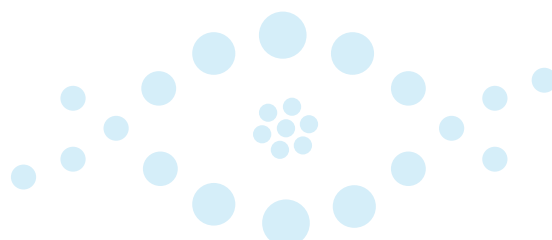
Your Views About BBS UK

We are a lifeline to so many people and it is vital we can continue to offer this kind of support. Our members told us recently what BBS UK means to them; their words are testament to the impact we have and would like to continue to have in the future:

- “Fantastic support in the early days of diagnosis!”
- “The charity welcomed us in with open arms and it was the first time ever that we didn’t feel alone or judged”.
- “The warm supportive family network made me feel confident and accepting of living with BBS. It has also helped me develop many life skills. My life would not be as joyous without this charity”.
- “We were warmly welcomed, and everyone involved with the charity could not have been more helpful. The conferences are brilliant, really informative but fun as well. BBS UK has shown us that we are not alone,

the charity and everyone involved and associated treat you like extended family and without all of you we would be lost”.

- “BBS UK helped me to not have to pay the extra bedroom tax and have my hours increased for my direct payments, I have always had great support from the BBS UK team”.
- “The best thing we look forward to is the BBS Conference and the BBS Activity weekend. My children love it and it makes them feel very special. Thank you all so, so much and please continue what you are doing so every family can stay happy”.



Contact Details

General Information

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General Contact/Enquiry:

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