

BBS UK Conference Updates

Advice Service and Clinics Updates

Ireland Information Day



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Foreword

Welcome to the 2023 Winter Newsletter

It really doesn't seem that long ago since we held our annual conference in Northampton, and I've been reminiscing about it; how lovely it was to meet so many of you, to hear updates from the professionals, and take away useful information.

I don't know how the team manage it but conference weekend really does get better each year.

You can read about it in this newsletter, and there are many photos of the weekend showing just what a great event it was.

Over the weekend, our trustees enjoyed meeting and chatting to many of you. If you weren't at the AGM or didn't get the chance to chat to them, you can read all about our newest trustees, Chetan Meshram, Tazeem Abbas and Gareth Owens on our brand new website.

We said a sad, but very fond farewell to Abbie, as she retired from the Board of Trustees. Abbie has been instrumental in the development of the Charity in recent years, and we will all miss her dearly.

We are eager to hear from those interested in becoming a trustee. More information about being a trustee and how to get in contact with us can be found later in the newsletter.

BBS UK has been out and about with staff and volunteers representing the Charity at various events, including two Sight Villages and the Retina UK conference. You can read more about these events in this newsletter, and, if you wish to volunteer your time to help represent BBS UK at events please get in touch.

I'm pleased to say that it has also been a busy time for fundraising and I would like to personally 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.

Our 'We Carry the Gene' campaign has been a huge success and has reached all four nations of the UK. I myself was very proud to take part and carry the baton on my (almost) daily runs along the beach, following which I handed it over to a family in Wales. The campaign is still running for a few months, so if you wish to take part please let us know.

This really is a packed newsletter, so I hope you find time to sit and have a read – I always look forward to receiving my copy as it gives me a good excuse to sit down with a cuppa for an hour!

On behalf of everyone at BBS UK, we wish you a very Happy New Year.

Laura Dowswell
Chair of the BBS UK Board of Trustees



BBS UK Annual Conference 2023

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 some 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 and Professor Helen May-Simera

Professor Beales opened the conference by welcoming delegates, old and new, in person and via livestream, from around the world, including the Netherlands, Germany, and China. Professor Beales told the audience that people all around the world have BBS, however the number of people with BBS varies from country to country. New data from the UK Biobank database shows 1 in 135 people carry one mutated gene associated with BBS, which means we can now estimate that in the UK, 1 in 70,000 have BBS, as opposed to the previous estimate of 1 in 100,000.

Professor Beales updated delegates about developments in gene therapy, a method of restoring the function of a faulty gene within a cell. Professor Beales, Dr Victor Hernandez and their team are working on gene therapy for BBS1, as this is the gene that is most often faulty in BBS patients. Gene therapy involves injecting a gene, using a harmless virus as a transporter, into cells in the back of the eye. In the studies on mice, this method was shown to prevent further deterioration of sight loss and it is hoped that this positive effect will last for decades.

Currently, the gene therapy is at the preclinical testing stage, which is when the therapy is made



as safe as possible before clinical trials can start in human patients; this will hopefully start in late 2024 through to 2026. It is important to note that gene therapy cannot restore vision that has already been lost, however it brings a lot of hope for preventing further sight loss.

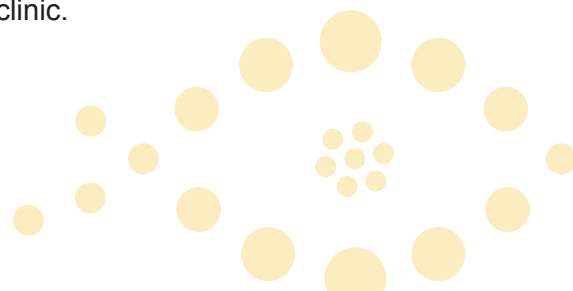
BBS Registry Update

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.



Personal Perspective

Richard Zimbler

Richard shared his experience of being an adult living with BBS, he said, "I'm hoping this will give inspiration to members living with BBS and show you can still live a good life".

Like many people with BBS, Richard was born with extra fingers and toes. Before his diagnosis, he was just considered a "clumsy baby", but in reality, this was due to problems with his eyesight. He was diagnosed with BBS when he was 6 years old.

Richard attended mainstream primary and secondary schools, and then the RNC (Royal National College for the Blind) specialist college for young people with visual impairment in Hereford where he studied performing arts.

In 2003, Richard recalled playing pool around the time he lost most of his colour vision. The yellow balls looked like the white ball, and he could barely see the black ball in low light areas of the table; by January 2004, he had only light perception.

Richard described a difficult period during one of the later lockdowns due to COVID-19, where he wasn't allowed to see anyone apart from his Mum who is his P.A. Due to frustration and loneliness, Richard began eating and drinking a lot, causing weight gain and the development of a wheeze as he spoke, however since 2021, he has lost over 5 stone in weight by managing his portion sizes and doing exercise.

Richard shared with the audience that some of the best experiences in his life occurred after losing his sight. With the support of staff at train stations, hotels, and taxi firms, he gained the confidence to lead an independent and fulfilling life.

Richard, formerly a trustee, vice-chair, and later chair of the board at BBS UK, continues to be an active volunteer and advocate. He frequently attends Sight Villages to raise awareness of BBS and utilises his people skills to host the music quiz



at the annual conference. Additionally, Richard volunteers for Retina UK, leading an online group for Wrexham and Wales to combat social isolation.

He also volunteered for RNIB in Wales as a 'Technology for Life' volunteer, using his love for technology to help blind and visually impaired people to become more confident with using technology.

Richard enjoys attending concerts, socialising with friends, experiencing audio-described theatre shows, and cheering for his hometown team, Shrewsbury Town, at their football stadium. Despite his visual impairment, he has travelled to various countries, including Spain, Australia, and most recently, Italy. Richard is engaged to Rachael Foley (BBS UK trustee), and they are planning their wedding for 2024. Richard ended his talk with an inspirational message to try the things you want to do, make the adjustments you need, and live a good life.

Urology Differences in BBS

Dr David Game

Dr. David Game spoke about the important role of kidneys in various bodily functions, including wee (urine) production, chemical balance control, waste removal, blood pressure regulation, and temperature control. The kidneys filter blood and create wee through the process. If we drink a lot, we will

wee a lot and it will be diluted, however if we don't drink enough we will wee less and it will be concentrated. It has long been thought that people with BBS do not concentrate their wee properly, but the BBS clinics team think it might be because they drink lots of fluids and therefore make lots of diluted wee, which is the correct body response.

While there is limited information available on bladder function in BBS, the clinics team have noticed that some patients hold onto their wee for a long time, while others may wee more often, possibly linked to a stretched bladder or weak pelvic floor muscles. Dr. Game presented



findings from a 2015 BBS UK survey which looked at common issues related to weeing, including infections and bladder control.

Dr Game encourages those with BBS to drink regularly, and wee regularly – it is important not to let the bladder become over-stretched. Pelvic floor exercises are recommended for those who wee a lot, or wet themselves when coughing; patients should discuss any problems with their BBS clinics team. The team plan to conduct a follow-up survey to further improve their understanding of wee related issues in BBS.

Personal Perspective

Ciaran Moy

Ciaran and his wife Marie travelled from Ireland to attend the BBS UK conference for the first time, to share their challenging yet hopeful experience of raising their 3-year-old son, Pauric, diagnosed with BBS in 2021.

Pauric had extra digits on each hand and foot at birth, but medical staff dismissed concerns and recommended surgical removal. Marie raised her worries about his delayed development and Strabismus, a vision disorder, but these concerns were also brushed aside. Although online research hinted at Bardet-Biedl syndrome, the family initially dismissed it, believing Pauric's eyesight was fine. They then opted for a private consultation with a genetic specialist who confirmed that "nothing was wrong."

In a June 2021 consultation however, Ciaran saw the words BBS on a piece of paper and diagnosis was confirmed. The prognosis included the possibility that Pauric might never walk or talk, could face complete blindness, and would likely have special needs for the rest of his life.

The weeks following the confirmed diagnosis were the most challenging for both Ciaran and Marie. They mourned the diagnosis, their son's future, contemplated the impact on their roles as parents, and grappled with the stark contrast between their envisioned family life and the reality they now faced.

Connecting with BBS UK helped ease the burden of the diagnosis, gave them the information they needed about BBS and proved to be vital in preventing potential irreversible damage to Pauric's kidneys, after an issue was identified by the nephrologist that required major bladder surgery. To access the BBS clinics service in

England, the family fought the Irish healthcare system and the Treatment Abroad Scheme (TAS), and, after three appeals, were finally able to attend. They saw first-hand the benefit of a joined-up, well-managed, multi-disciplinary approach for BBS patients.

Despite the forthcoming challenges and numerous appointments, Pauric is thriving. He's a mischievous boy, cherished by his family, and has two supportive older sisters. Instead of comparing him to his peers, the family celebrates Pauric's achievements, including walking, running, talking, and attending playschool.

"The milestones may take longer to happen, but when they do, they're so much more joyful."

Ciaran has played a key role in establishing the Ireland Regional Group. He firmly advocates for relieving Irish families of challenges relating to accessing services, pursuing appointments, and navigating difficult systems and administrative processes.

A full report on the Ireland Information Day is available later in this edition.

Obesity in BBS: mechanisms and treatments

Dr Elizabeth Forsythe, Dr Lukas Foggensteiner and Sarah Flack

Obesity in BBS

Dr Elizabeth Forsythe stated that obesity is increasing worldwide, partly due to the easy availability of high calorie food that has been designed to make us want to eat more of it, as well as genetic factors that contribute to our weight.

In the medical field, the terms overweight and obese are specific terms defined by the World Health Organisation (WHO), relating to BMI. 'Overweight' is a BMI (Body Mass Index) of between 25-30, and 'obese' is a BMI of over 30.

Data from the BBS clinics shows that, like the general population, individuals with BBS experience weight gain with age. At the BBS adult clinic in Birmingham, 10% had a healthy BMI, 20% were overweight, 27% were obese, and 50% were severely obese.

Individuals with BBS often experience heightened

hunger (hyperphagia) compared to others. Normally, signals in the body communicate with the brain, assessing whether the body has enough food. If more food is needed, the brain signals hunger. In BBS, these signals malfunction, leading to feelings of hunger even when the body doesn't require more food. Besides increased hunger, certain eating behaviours contribute to obesity, including stress eating, habitual eating, boredom-driven eating, and other complex behaviours associated with autism.

How can we treat obesity in BBS?

Obesity can be treated with diet, exercise, medication and psychological strategies.

Exercise is crucial not just for weight management but also for mood balance, brain sharpness, and reducing the risk of, or delaying dementia, heart attacks, strokes, and more. Unlike medication, exercise provides comprehensive benefits for the body. Dr Elizabeth Forsythe encouraged attendees to embrace a physical activity in the next few weeks, challenging them to share their experiences at the next conference.



In addition to the BBS clinics, there are obesity clinics available for both adults and children to address weight-related concerns. Patients can be referred to these clinics, for ongoing support and guidance between BBS clinic appointments.

Dr. Foggensteiner spoke about weight-management drugs and presented results from case studies of BBS patients prescribed Semaglutide (known as Wegovy), which showed significant weight-loss and improved wellbeing, however progress was disrupted because of supply issues.

An international trial studied the effects of another weight loss drug, Setmelanotide on hunger and weight loss in adults and children with BBS with positive results; participants over 12 years old experienced a weight loss of greater than 10%. At the time of going to print, it is unknown when Setmelanotide will be available for those with BBS in the UK.

Diet and Exercise

Sarah Flack shared the importance of individualised dietary advice for people with BBS, whilst recognising that common factors include, lower energy needs, increased hunger (hyperphagia), and food seeking behaviours.



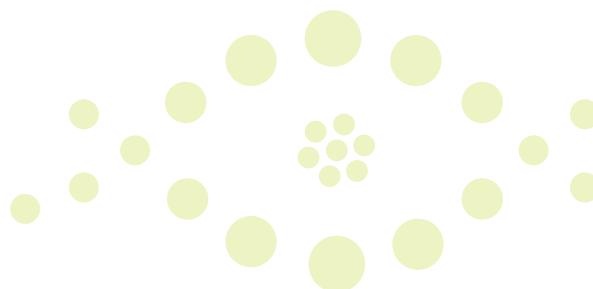
Sarah shared positive results from the children's clinic at Great Ormond Street Hospital (GOSH), with over half reducing their BMI between clinic appointments, showing that an increase in BMI in childhood can be avoided,

Sarah highlighted the challenges for children and adults with BBS in managing their weight due to excessive hunger and encouraged people to be kind to themselves and to keep trying their best.

Making lasting changes to our diet or activity level takes time and we should try to remember that new habits take a few weeks to stick. Don't expect to jump into a new routine without hiccups, try tackling one thing at a time and aim for those small victories to build momentum; other helpful tips include:

- ✓ **Sleep Well:** Lack of sleep can lead to craving higher energy foods like biscuits or crisps. Try cutting back on caffeine in the evening and going to bed a bit earlier.
- ✓ **Smart Drink Choices:** Swap high-calorie drinks like fruit juice or fizzy drinks for water.
- ✓ **Portion Control:** Use smaller plates and fill half your plate with vegetables.
- ✓ **Move More:** Find something active that you enjoy, sneak in bits of activity during your day, set realistic goals, increase gradually, and appreciate the benefits of being more physically active.

Sarah emphasised the importance of sharing your eating, drinking, exercise and sleeping habits with the BBS clinics team, who are there to help you.



From feeling alone to making an effort to change the BBS world

**Bendert De Graaf,
Bardet-Biedl Syndrome
Netherlands**

Bendert de Graaf and his wife Nienke travelled from the Netherlands to experience the BBS UK conference for the first time. Bendert shared their experiences as parents to Jayke, their 10-year-old son who has BBS, as well as the efforts and mission of BBS Netherlands and BBS International. Bendert has a PhD in Molecular and Medical Genetics and is educated in rare conditions, while Nienke trained as a nurse working with people with disabilities.



Back in October 2013, when Jayke was born with extra fingers, Bendert asked the surgeons if there was a syndrome involved. They were told that one in seven kids in the Netherlands have extra fingers or toes, and the fix was just to surgically remove them. As new parents, not wanting anything to be wrong with their child, they went along with it.

Nienke became pregnant again when Jayke was one year old. However, the ultrasound for his sister revealed severe issues: her kidneys were almost completely non-functional and she had extra fingers and toes. Doctors delivered the heartbreaking news that she would likely not survive long after birth. Subsequent genetic sequencing revealed that Jayke had Bardet-Biedl syndrome (BBS).

Bendert and Nienke had so many questions about long-term prognosis and how to navigate the situation. Bendert leaned into his education and became the “mad scientist”, researching online and contacting the main organisations and scientists associated with BBS. He reached out to Professor Phil Beales and BBS UK, and this support gave Bendert the confidence and tools to start a foundation dedicated to BBS in the Netherlands.

At this time, there was very limited information available about BBS in the Netherlands, especially for the general public. Experts in rare conditions were unfamiliar with BBS, most having never heard of it. Bendert and Nienke sought connections with other parents of children with

BBS and aimed to create a platform for shared experiences. They knew that to enhance their son’s future prospects, they needed to initiate international efforts, raise funds, and advocate for research, particularly in gene therapy and improved kidney failure management.

Bendert extended his contacts internationally, working not only with the UK, but with America, France, Denmark, Norway, Sweden, Poland, Italy and more. The group wants to work with any organisation or individuals with BBS, with the end goal being to set up a global registry.

Bendert concluded his presentation by reiterating that if we work together, we can collectively raise global awareness, enhance diagnostics, healthcare, and support throughout the BBS lifespan, as well as provide mutual support within the global BBS community.

Introduction to Microsoft and Accessibility

David Prince

David Prince works for Microsoft and has a daughter with BBS. Vision, hearing, neurodiversity, learning, mobility and mental health are all factors Microsoft consider when developing accessibility in their products/technology and David gave a very helpful presentation, sharing some useful tips on getting the most out of readily available technology.



David encouraged users to explore the many settings available on any Windows PC or laptop, to customise them according to individual needs. To access these features, simply press the ‘Windows Key’ and ‘U’ at the same time. This action will open a list of accessibility features that can be tailored to improve accessibility, including: text size and colour, mouse pointer size and colour, colour contrast, speech recognition, narration of text and voice control. Dictation features allow users to dictate emails and documents, as well as control their PC using their voice. Immersive Reader offers playback of written text.

David went on to talk about developments in Artificial Intelligence (A.I), which will allow features including speech, image and natural language

recognition to be integrated into technology more widely. The 'Seeing AI' app for iPhones can identify currency, describe surroundings, recognise friends and people, provide audio cues for locating barcodes, and scan and identify products, along with text recognition and reading capabilities.

Additional Microsoft accessibility resources on offer include:

- YouTube channel called 'MSFTEnable' with videos explaining how to use accessibility functions.
- Free Microsoft Disability Answer Desk dedicated to helping people with disabilities get the most out of their Windows PC, laptop or Xbox.
- Accessibility resources and training on the Microsoft website
- For businesses, there is a free 'Accessibility Fundamentals' training course available.

For those who wish to improve access to technology, David recommends:

- Contact RNIB, Microsoft, Apple, Google, and ask for advice and guidance
- Use a 15" touch screen PC with the ability to zoom in and out
- Ask your child's school to supply JAWS software
- Embrace technology! Using voice over and accessibility functions on your PC and phone, and download apps such as Seeing AI.

Thomas Pocklington Trust

Matt Harrison, Engagement Manager and Rachael Foley, Sight Loss Council Member



Thomas Pocklington Trust (TPT) is a national charity which supports blind and partially sighted people with a focus on education, employment and engagement. They provide support for adults and young people, and they fund Sight Loss Councils across England.

Matt explained that Sight Loss Councils are driven by blind and partially sighted adult volunteers who address local issues and collaborate with businesses and service providers to enhance accessibility. They run visual impairment awareness sessions, VI forums, and undertake 'mystery shopper' activities. Volunteers meet monthly to discuss projects and then form subgroups. Current areas of focus include transport, the built environment, retail, sports and leisure, and arts and culture.

Rachael Foley shared her experience of being a Sight Loss Council member in Greater Manchester, she said that joining the Sight Loss Council had allowed her to meet new people, make new friends and increase her confidence in navigating her local area. She is looking forward to future projects involving public transport, the built environment and ongoing work on retail accessibility.

To join your local Sight Loss Council, contact the Thomas Pocklington Trust through the contact details on their website www.pocklington-trust.org.uk.



Azrat Mirza

Azrat Mirza is a BBS UK trustee and sits on the Scientific Advisory Board. He works in the pharmaceutical industry, is from a minority ethnic background and is very keen on representing the views of others from similar backgrounds. Azrat speaks Urdu and Punjabi.



Azrat spoke about his 18-year-old daughter Haniyah's progress and achievements over her life so far, as well as some of the stumbling blocks they've encountered.

Doctors identified that Haniyah had polycystic kidney disease during the antenatal 20-week scan. Haniyah was born with a sixth digit on her right foot and experienced constant hunger, leading to weight gain. Additionally, she had a head tremor caused by eye issues, resulting in her eyes moving, and her head compensating with side-to-side movements. Her walking was significantly delayed, leading to challenges such as frequent falls and difficulty interacting with other children at school. Haniyah also exhibited signs of learning difficulties.

A kidney doctor treating Haniyah raised the possibility of Bardet-Biedl syndrome (BBS) after attending an educational talk. Noticing features consistent with BBS, such as being overweight, kidney problems, extra digits, and eyesight issues, the doctor arranged for genetic testing. Initial results were inconclusive, but upon further testing, the diagnosis of BBS was confirmed.

After receiving the diagnosis, Azrat's family experienced mixed emotions, relief for understanding Haniyah's challenges and sadness, especially concerning potential blindness. Despite the heart-wrenching thoughts of limitations, the family united and sought assistance from BBS UK.

Haniyah has recently left 6th form college and has worked tirelessly to achieve qualifications, despite her learning difficulties and has achieved her Grade 4 in Maths! She even presented to college about her positive journey as well as the difficulties she's had through college.

The support Haniyah receives has

been fundamental to her development, success, confidence, and happiness. She receives Direct Payments and has support from a PA (Personal Assistant) who helps with her educational needs, as well as supporting her to access the local community.

Haniyah aims to work in the NHS and recently secured a volunteer placement at a hospital tea bar in Sheffield. Initially nervous, Haniyah received positive feedback from her manager, expressing confidence in her ability to serve customers independently and handle end-of-day cashing. Azrat encourages others to engage with local voluntary organisations for potential placements.

Haniyah also secured an internship with the NHS through Project Choice, a program offering tailored educational support and a year-long supported internship for young adults (16-24) with learning difficulties, disabilities, and/or Autism.

Azrat concluded his talk with the following advice:

- Advocate for support—abundant resources are accessible, beginning with BBS UK
- Explore local schools with specialised provisions for visually impaired students
- Reach out to the BBS UK Advice Service for guidance on benefits, Education, Health and Care (EHC) Plans, and advice on Direct Payments

And finally, a piece of advice directly from Haniyah: always support and encourage people with BBS.



Your thoughts about Conference 2023



“We really had a great time, we felt so welcomed by you all.”

“Although it was only our second conference it felt like a reunion! Both my girls and I love our time we get to spend learning and having fun with you all. Thank you for all the energy and effort that goes into making this happen!”

“It was great to have the opportunity to meet other families, attend the workshops, and we also met some amazing staff.”

“A brilliant conference as always: keep up the good work!!”

“We found the conference informative, welcoming, organised, fun, kind, and a wonderful opportunity to connect with the BBS community and professionals.”

“Professional and perfect audio and video quality!”



“Everything about the day is really lovely. It’s a very special event that we really look forward to. Everything is so well organised and we can just relax and make the most of the wealth of information, support and advice that are on offer without having to worry about anything else.”

“Excellent activities and support from all staff.”



Welcome to Team BBS UK!

Samantha Serido: Fundraising and Communications Officer

We are very excited to welcome Samantha Serido to Team BBS UK, as our new Fundraising and Communications Officer. Samantha has devoted the



lion's share of her professional life to date, to non-profit and health systems management, fundraising, and content development. Before moving to Manchester, she earned a Bachelor of Arts in Writing in the United States, where she was raised. Samantha also holds a Master of Arts in Social Anthropology from the University of Manchester, with a specialisation in Wellbeing Anthropology. In her spare time, Samantha enjoys hiking, yoga, and spending quality time with friends and family.

Samantha says, "I'm so thrilled to be contributing to a charity that is so well-integrated with effective care and inclusivity. Moving forward, I'm excited to champion the incredible stories from our community, strengthen our communication and fundraising channels, and bolster the vital work undertaken by BBS UK overall. I am moved to express my gratitude toward the team I have met so far for offering me such a warm welcome, and I am looking forward to connecting with more of this wonderful community in the coming months!"

If you would like support with a fundraising idea, please email Samantha on fundraising@bbsuk.org.uk.

Trustees

At the BBS UK Annual General Meeting (AGM), held during conference weekend in September, members voted as follows:

- Laura Dowswell commences her final year as Chair.
- Chetan Meshram is Vice-chair after being co-opted as a trustee earlier in the year.
- Tazeem Abbas is Treasurer after being co-opted as a trustee earlier in the year.

- Dianne Hand will remain as Secretary.
- Gareth Owens is a trustee after being co-opted earlier in the year.
- Liam Wilkie, Rachael Foley and Stefan Crocker will continue as trustees.

You can find out more about our trustees by visiting our website.

Thank you and goodbye to Abbie Geeson



It is with a sad heart that we said goodbye to Abbie Geeson, who was our Treasurer and trustee for many years. During the AGM, Team BBS UK gifted Abbie with a beautiful bouquet of sunflowers, a gift voucher, and plenty of gratitude for the incredible amount of work and devotion she has given. Abbie gave a heartfelt farewell, however will still be actively engaged in the BBS UK community.

Truly, thank you so much Abbie.

Fundraising Round Up

First of all, a huge 'Thank You' to everyone who has raised funds, donated and helped in any way over the past year; it is only through your efforts and commitment that BBS UK can provide the services that it does. So, what have we done?

Our Advice Service has supported service users with a range of issues including, benefits applications, care package reviews, education issues, future planning and housing issues; the team's knowledge of the education, health and social care systems, together with their understanding of BBS make them invaluable advocates for our community.

We held a weekend conference in September, attended by approximately 230 people keen to learn more about BBS, research and the support available. 98% said their reasons for attending were met and 96% said their experience was Good or Very Good.

Earlier in the year we funded a programme to support adults living with BBS who were experiencing anxiety and had become socially isolated. Beneficiaries reported improved communication and self-advocacy skills, being able to see problems in a different way, better management of anxiety and feeling understood. With funding we aim to continue developing this much needed service.

We supported our Ireland Regional Group to hold a day conference in Belfast which was very well attended and enormously valued, easing the isolation and lack of information that many had experienced.

We have supported research into BBS and understanding of BBS in many ways, engaging with researchers and working with the international BBS and ciliopathy communities. This is just a snapshot of the work that we do to improve the lives and wellbeing of those living with BBS and your contribution over the past year has made this possible – thank you!

Key aims for this year were to diversify income, secure some long-term funding and develop our fundraising and communications



strategy, however this stalled with the loss of a key staff member in May. We are delighted to be able to report that we have now filled the post, with Samantha, our new Fundraising and Communications Officer joining the team at the end of November.

General donations and fundraising have been less than expected so far this year and we will be looking at ways of diversifying and generating more income in the next few months. Our We Carry the Gene campaign however, has been enormously successful and we are immensely grateful to everyone involved.

We have had donations from businesses that employ members of the BBS community and this is an easy way of raising funds and awareness for the Charity. Heartfelt thanks to the Engine Lease Finance Corporation, W M Thomson & Sons, and to ARAG Legal Protection for their generous support this year.

Other companies offer matched funding which provides a good incentive to do some local fundraising; maybe you could ask your employer if they would



consider BBS UK to be their charity beneficiary this or next year? We have also received donations from local Rotary Clubs and sports clubs, so again, if you or someone close to you is involved in a local group, please do consider putting BBS UK forward as a beneficiary.

Our loyal band of 'Friends' continue to give valuable support to the Charity through their fundraising events or monthly donations, ranging from £5 right up to £100; every single pound helps and we thank you all for your generous commitment. We have around 50 regular givers, which is less than 7% of those who use our information and support services, so imagine what we could do with more of you on board! If you or someone you know would like to become a Friend of BBS UK, please complete the form on our website: www.bbsuk.org.uk/friends-of-bbs-uk/, email admin@bbsuk.org.uk or telephone Natalie on 07784 922654.

In September we were delighted to receive a grant of £9,500 from the National Lottery Community Fund towards our conference costs,

which enabled us to heavily subsidise attendance and make the event more accessible to all. Our grateful thanks go to National Lottery Players for making this support possible.

We were also grateful to receive £10,000 conference sponsorship from Rhythm Pharmaceuticals further enhancing the support we could offer those wishing to attend.

Looking ahead, we welcome Samantha, our new Fundraising and Communications Officer to the team and are excited to see how our fundraising function develops in the coming months. If you would like support with a fundraising venture or would just like to introduce yourself and welcome Samantha to the community, you can email her at fundraising@bbsuk.org.uk

We feel incredibly blessed to have such a supportive and engaged community and thank you all from the bottom of our hearts for all that you do to enable us to continue providing and developing the support services that are so desperately needed. We wish you all a very happy New Year!

BBS Clinics Update

Amy Clapp, Patient Liaison Officer
Angela Scudder, Patient Liaison Officer

Birmingham Children's Hospital: Catrina Leal, BBS dietician from the Birmingham Children's Hospital (BCH) service, welcomed a beautiful baby girl back in August, and will be on maternity leave for the next 12 months. We are fortunate to have Safia Ravat supporting our clinics and Safia is looking forward to meeting you all in both face to face and virtual appointments.

We are looking forward to holding our first ever BBS UK transition clinic event, supporting our young people who are transitioning from the children's service at BCH over to the adult's service at Queen Elizabeth Hospital, Birmingham (QEH). We have organised an orientation visit at the hospital where our young people will have the opportunity to have a look around the department and speak with our Clinical Nurse Specialist, Sarah Borrows and Transition Co-ordinator, Marie McGee, in preparation for their clinic appointments the next day. In the evening they will enjoy a couple of games of bowling and dinner and get to know each other and hopefully make new friendships. BBS UK Patient Liaison Officers, Amy and Laura will be present throughout to support.

Queen Elizabeth Hospital: QEH continue to offer alternate face to face and telemedicine appointments with no changes to staff. It has been a long and ongoing problem at QEH that clinic reports have not been sent out in an accessible format. We are delighted to report that there has now been a change of policy which means that those who wish to receive their reports via email, are now able to do so. During the clinic appointment, patients will be able to request their report in this way and sign a disclaimer, acknowledging the risk. BBS UK has long campaigned for the availability of emailed reports, which will undoubtedly improve privacy, independence and safeguarding for patients.

We are aware that some patients have not received their clinic appointment letters from the hospital. This is due to the settings and preferences in the Dr Doctor system. Please ensure when using Dr Doctor, you have selected your correct method of contact. If you have any queries, please contact your Birmingham Patient Liaison Officer.

Great Ormond Street Hospital (GOSH) & St Thomas' Hospital (GSTT)

Since April 2023 we have welcomed six new patients and their families to our BBS clinic at GOSH and four new patients to GSTT. Through a combination of in-person and telemedicine clinics we have seen 42 patients across 7 clinics in Gosh and 80 patients across 9 clinics in GSTT.

Attending appointments and communicating with the clinics about cancellations is vital, to support us with ensuring the clinics run smoothly, and to make sure we are using every valuable clinic appointment to help someone with BBS. Unfortunately at GSTT we are experiencing many late cancellations. The trust policy only allows for 1 DNA (Did Not Attend) or 2 cancellations before they discharge patients back to their GP. We understand that there will always be times that patients will need to cancel due to ill health, and to give us the best chance of filling the appointment, patients should contact Angela directly either by email: angela.scudder@bbsuk.org.uk or phone: 07591 206788 as soon as possible.

Well Done team GOSH you had 100% attendance! We are still not able to provide lunch to our

GOSH patients due to the trust's policy, so please bring food with you as it's a rather long day. We still provide tea and coffee at GOSH on Friday morning, and if you have your own travel cups please bring them for us to fill as we like to be as green as possible! At GSTT we continue to provide breakfast, lunch, tea, coffee, and water for our patients.

In October, a new computer system called Epic was launched at GSTT, allowing patients to access their MyChart patient portal. Patients will receive their set-up instructions and codes at their next appointment.

Gt Ormond Street Hospital uses the MyGosh patient portal, If you haven't already accessed MyGosh please contact the MyGosh team at MyGOSH.Helpdesk@gosh.nhs.uk. It only takes a couple of minutes to set up your account which will give you access to your scheduled appointments, blood results and your BBS clinic report.

Finally, we would like to thank the amazing teams at GOSH and GSTT Rare Disease Centre that support the clinics and do so much hard work to make them run smoothly, with special thanks to Tanjina and Maria (GSTT) and Chelsey (GOSH).

BBS UK Advice Service Update

Rebecca Perfect, Advice Service Manager

The Advice Service continues to be busy, but it was lovely to catch up with so many of our service users, past and present, at the conference and to run another successful Advice Service drop in. We continue to support people in a wide variety of areas. To date since our creation in 2020, we have received over 110 referrals on a wide variety of issues around social care and education. Finding and securing appropriate housing also remains a big issue, as does getting the right care packages and Education Health and Care Plans (EHCPs).

Interestingly we are seeing an increase in reviews of care packages as county councils seek to ensure that they are not overpaying for care. We are happy to support you if you are worried about your support being cut. Although there are concerns about packages being cut, what we more often find is that many people with the complex needs of BBS are underprovided for. We regularly work to support people to ensure they are getting the right level of support to live how they want.

If you think you or a family member currently

do not have enough social care support or are overdue a care package review, our advice is NOT to wait for the local authority to contact you. We would suggest you contact your allocated social worker to request a package review or, if you do not have a social worker, contact your local authority and request a review. Local authorities are often extremely busy and will not always prioritise contacting people or families whose needs they feel are being met.

If you do contact the local authority for extra support and they do not respond, or they give you an extremely long waiting time, then do not hesitate to contact us and we will happily support you to chase them up.

Whilst things are getting more challenging in the current cost of living crisis, we are delighted that amongst many other things we have managed to support a number of people to extend their care packages this year, and to get allocated social workers when required.

Are you a Member of BBS UK?

Please take a moment to ensure you have joined our BBS UK membership.

BBS UK's members are at the heart of our work. Become a member and you'll join a growing group of people who play a part in everything we do.

Membership is entirely **free** to ensure that everyone who needs our support is able to access it.

To become a member, visit our website www.bbsuk.org.uk and click 'Become a Member' at the top of the home page.

All members receive:

- Access to the BBS UK Advice Service
- Access to BBS UK projects including IT Equipment Fund
- Bi-annual newsletter which includes our annual conference report, information booklets and publications
- Priority notice of BBS UK events and activities
- The chance to vote to elect our trustees and to become a trustee yourself

As a member, you can get involved in BBS UK's work in all sorts of ways. There is no pressure to do anything, but if you are keen to take action, we will support you every step of the way.

Options include:

- Taking part in surveys, focus groups and government consultations
- Sharing personal experiences
- Fundraising to support BBS UK's work



Friends of BBS UK

By making a regular monthly donation or raising funds, you will be helping BBS UK to plan ahead and make long-term developments to our much needed and highly regarded support, advocacy and information services. A regular gift allows us to plan strategically as we know day in, day out that we will have a steady income that we can rely on.

To become a Friend and regular giver:

- Scan the QR code
- Complete the 'Become a Friend of BBS UK' online form
- Setup a monthly standing order with your bank. Use your name as a reference



Thank you to our Friends for their generous support!



BBS Regional Group Ireland: Information Day May 2023

The first all-Ireland BBS Information Day was held in Antrim, Northern Ireland on the 13th May 2023. People with BBS, family and friends travelled from all over Ireland to attend as well as Team BBS UK and professionals from the BBS clinics. It was a great day for collaboration, camaraderie and good fun, with a quiz and charity draw being held in the evening.

Laura Dowswell, Chair of BBS UK, welcomed everyone and introduced the speakers.

Professor Phil Beales, who has dedicated 30 years to working with and researching BBS, gave an overview of BBS, the research around the syndrome, and the BBS clinics which are NHS funded, highly specialised and offer a multi-disciplinary approach to BBS. Some of the main points were:

- Each clinic provides access to specialists in areas covering Ophthalmology, Nephrology, Dieticians, Clinical Psychologist, Geneticist, Speech and Language and Endocrinology, as well as Telemedicine Clinics for those who cannot attend in person.
- There are currently around 650 patients on the clinics' database, with 250 patients attending face to face and 92 patients attending remotely annually.
- BBS1 and BBS10 are the most prevalent variants accounting for approx. 70% of all cases.
- NHS BBS Patient Registry maintains a database of BBS patients which is primarily used for tracking purposes and for identifying potential candidates for clinical trials.
- There have been 1428 BBS related research publications to date, the majority of which have been written since the year 2000.

Katy Megahey gave her Personal Perspective, where she spoke of her life over the last 40 years, her difficulties with weight and how she worked to overcome them with the assistance and advice from a dietitian. Katy was diagnosed at the age of 6 with BBS and is now registered as blind. Katy loves concerts, day trips and shopping. She has competed in various events and is very active.



She has travelled a lot with her family, went to a performing Arts School, is part of a choir and is training in Marie Curie therapies. Her third Guide dog is currently in training.

Charlene Ferguson, Family Support Officer with the Royal National Institute of Blind People (RNIB) in Belfast, spoke about the government funded services they provide to parents and families from early years right throughout adulthood:

- 6-week information sessions
- Shape & Share events which bring children, young people and their families together with no cost attached, for peer support, to share experiences and take part in fun activities both in person across the UK and online.
- Family information days, events and education support.
- Therapies for children such as swimming, massage and many others.

Sandy Woods gave her Personal Perspective on being a parent to Charlie, aged 4, who was diagnosed in 2019 with BBS. During her moving talk, she shared her family's experiences, the high and lows, and how every day with Charlie is a gift.

Sarah Flack, Principal Dietitian with BBS UK, provided thought-provoking facts which pointed out some of the myths about what traditionally would be considered healthy foods, but which are high in calories so should be limited when

following a calorie-controlled diet. She also shared 'Easy Wins', which included sleeping well, using smaller plates and trimming fat off meat.

Rino Costanza, Scientific Affairs Manager for UK & ROI, Rhythm Pharmaceuticals gave a background to Rhythm Pharmaceuticals, and a summary of Setmelanotide, a drug licensed to treat patient obesity.

Rino explained that many BBS patients have an impaired function of the MC4R pathway, which is the key regulator of body weight and hunger. The hormone Leptin should signal to the brain when full however with a faulty MC4R this signalling is impaired, and the brain doesn't get the message that "I'm full". The Setmelanotide drug bypasses this signalling. Rhythm Pharmaceuticals are currently collaborating with BBS UK and BBS patients.

Gillian Stafford gave her Personal Perspective and detailed her struggles in her early years at school and how she was diagnosed with retinitis pigmentosa at 17 as well as her late diagnosis of BBS at 41. It was at college that she found her voice and how with the assistance from great tutors and her family she graduated with a Master's in Social Policy. She has since become an access ambassador, spoken at numerous events abroad and is an active member of an Independent Living Movement Group.

Tonia Hymers, Operations Manager, BBS UK, outlined the history of BBS UK including its mission and vision and gave an update on the services provided by the Charity. BBS UK has been supporting the establishment of an Ireland subgroup to help create greater awareness, networking and support to people with BBS and their families and/or carers.

In the afternoon, round table discussions took place where the attendees were able to ask questions of the expert speakers which proved to be a great additional source of information. Lots of activities were available for the children including



a pop-up cinema, board games, nail painting, colouring and much more, all of which added to the enjoyment of the day.

Lindsay McKnight, founder member of BBS RG Ireland closed a very successful event by thanking everybody for attending, the exhibitors, the speakers and the UK and Irish BBS groups. She rightly received recognition from the BBS Regional Group Ireland for her enthusiasm and passion in bringing this event together.

Feedback

After the event, attendees were asked to rate aspects of the day out of 5 stars, with 5 being the highest rating. Here are the results from those who completed the evaluation questionnaire:

- The Information Day overall – 5 stars (100%).
- The morning programme – 5 stars (88%) 4 stars (12%).
- The afternoon programme – 5 stars (82%), 4 stars (12%), 3 stars (6%).
- The children's/young people's activities – 5 stars (100%)
- Lunch and general refreshments – 5 stars (62.5%), 4 stars (25%), 3 stars (12.5%)



BBS UK Out and About



Queen Alexandra College

QAC Sight Village 2023: Sight Village is an exhibition for people of all ages living with varying degrees of sight loss, professionals supporting and advising visually impaired people, 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.



This year, Sight Village events were held in Birmingham, Exeter, London and Cardiff. Team BBS UK, along with some of our volunteers, exhibited at both the Cardiff and Birmingham events, we reported on the Cardiff event in our last newsletter.

Over the course of two days in Birmingham, BBS UK raised awareness of Bardet-Biedl syndrome, and the work of the Charity by talking with attendees, exhibitors and professionals, exchanging vital information about support, resources and technology available to help people with sight loss.

On day one, our trustee Rachael Foley and former chair of the board Richard Zimmler helped at the BBS UK stand, alongside BBS UK Patient Liaison Officers, Amy Clapp and Laura Davies. On day two, Aneeba Ahmed joined the team, helping to spread the word about BBS UK, as well as trying some of the technology being exhibited.

It was a great opportunity to make professional contacts, as well as meet people face-to-face and we look forward to attending more events this like in the future.

Retina UK Annual Conference: In June, Patient Liaison Officer, Angela Scudder, Operations Manager, Tonia Hymers and volunteer, Daniel Hymers attended and exhibited at Retina UK's annual conference in London.



Presentations included innovative technology and the latest updates in gene therapy and potential stem cell treatments. We connected with many professionals working with BBS patients including eye clinic liaison officers working at specialist eye hospitals and qualified teachers for the visually impaired, enabling us to share information resources far and wide. It was a very enjoyable conference, and we encourage all our members to attend – the event is also live-streamed. Presentations can be viewed on the Retina UK website.



BBS Global Day 2023

BBS Global Day is an annual cooperative effort, coordinated by BBS International, and with support from BBS family organisations in France, Italy, the Netherlands, the UK and North America. This year, BBS Global Day coincided with the BBS UK's Annual Conference on Saturday 23rd September.

During BBS Global day the BBS community is encouraged to:

- **Connect** with your national BBS association, or with BBS International.
- **Spread** the word: Post on social media using the hashtag #BBSGlobalDay. Tell your

networks why accelerating BBS research and treatment matters to you.

- **Raise** funds: Donate and encourage others to donate via your national association.

Families from around the world supported the global BBS community by pledging to walk or cycle on BBS Global Day to raise funds for BBS research and support, and by sharing their personal stories of living with BBS on social media.

To find out more, please visit www.bardetbiedl.org/bbs-global-day

Trustee Position Available

Are you passionate about making a difference to the lives of those living with BBS? Then we want to hear from you!

BBS UK is looking for new trustees, with professional experience (including in marketing, media, PR, finance or legal) to support the Charity with governance, communication and engagement. We are committed to equality, diversity and inclusion and are looking for trustees who bring a range of experiences and skills to complement our team.

As a trustee you will have:

- Induction, training and reimbursable expenses
- Opportunities to make strategic decisions and develop new skills
- Opportunities to network with like-minded organisations and professionals
- Opportunities to influence the development of innovative projects
- The chance to make a real difference to the wellbeing of the BBS community

No previous trustee experience is required.

Commitment:

The expected time commitment is three trustee meetings per year (lasting approximately 3 hours each), via Zoom or at a central location; attendance at our annual AGM; and an annual

strategy day. Regular monitoring and response of emails is also required.

Interested?

If you're interested, please email admin@bbsuk.org.uk

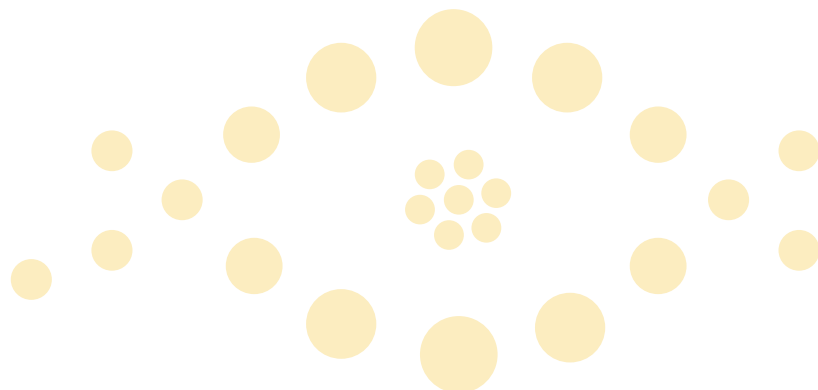
What our trustees say:

"After receiving tremendous emotional support from people connected with the Charity and learning so much at the first conference I attended, I wanted to show my gratitude by supporting others and being a trustee enables me to do that."

Dianne Hand – Secretary of BBS UK

"As a family, and of course BBS does affect the whole family, we were determined to 'give something back' and I have been a trustee on the Board of BBS UK for several years."

Laura Dowswell – Chair of BBS UK



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Patient Liaison Officer:

London BBS Clinic Service

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