

Newsletter

Spring 2025

Book Now: BBS UK Conference
25th - 27th April 2025
Registration information inside!

Inspiring Experiences
Sailing, Fitness and Visiting the US
Fundraising Heroes



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Foreword

Hello everyone and welcome to the Spring 2025 edition of the BBS UK Newsletter!

I am delighted to be writing this foreword in my new role as Chair of the Trustees of BBS UK and want to say a personal thank you to our outgoing Chair, Laura Dowswell for all the support she has given me and the Charity over the last 9 years. I know I speak for all of the Trustees when I say that we will miss working with Laura very much.



It is great to be joining the Charity at this time; the number of families that we are supporting has never been higher. I know that the Advice Service and Clinics Support Services will continue to grow this year, providing much needed support across the country and the Board of Trustees and I want to ensure that we are able to reach as many families as we can. We are in the process of finalising our organisational strategy which will outline how we intend to continue to develop and improve our services and support over the next three years and I hope that we will be able to share this with you all in the next few months.

As always, this edition of the newsletter is packed full of information and updates about the work of BBS UK, and you will read about the amazing fundraising that has been taking place. I wanted to say a huge thank you to Tanja and her family, Caitlin and her family and Graeme and his golfing friends, who have collectively raised a whopping £4,460! A fantastic achievement! If anyone has any ideas for future fundraising events please do get in touch with us, we would love to hear from you.

I am very much looking forward to meeting you all at the conference in Northampton on the 25th-27th April for what will be a great weekend; I know the team have been working extremely hard in putting it together. The conference will give us all an opportunity to come together to learn from each other, hear from professionals about advances in research and socialise with the BBS UK community. If you haven't already booked your place, and would like to attend, please visit our website.

In the meantime, if you need to get in touch with us, please do, our contact details can be found on our website, or email admin@bbsuk.org.uk.

Happy reading,

Emma Hughes

Conference 2025



We will be returning to the Hilton Hotel, Northampton, across the weekend of 25th-27th April 2025, to once again welcome delegates from far and wide for a weekend of learning and sharing experiences about all things BBS.

The main event is our formal conference programme on **Saturday 26th April**, and we are working hard to put together an exciting and informative programme with something for everyone! We will hear about the latest research, including an exciting presentation from **Professor Phil Beales**, as well as updates on clinical management, personal perspectives and much more!

Dates: Friday 25th – Sunday 27th April 2025

Location: Hilton Northampton, NN4 0XW

Information about the event and how to book can be found on our website. Scan the QR code to take you to the page.



The conference day and children's trip (including lunch and refreshments) on Saturday 26th April is **free of charge** for those with BBS and their immediate family (parents, siblings, grandparents) up to a maximum of four adults. Attendees will need to pay their own accommodation costs, and we have a block of rooms on hold with the Hilton Hotel on a dinner, bed, and breakfast basis - other hotels are available nearby.

Donations are of course extremely welcome and can be made through our website or by scanning the QR code:



Additional family/friend attendees: £30
Additional child attendees: £20
Professional Delegates: £50

Livestream

We will also live stream the conference via Zoom and record the sessions for those unable to attend in person. More information and how to register will be available nearer the time.

Programme

The final programme is still in development and will be available in the next few weeks - you can expect updates on BBS research, clinical management, personal perspectives and much more; speakers confirmed to date are:

- Research update: Professor Phil Beales, ICH & Axovia Therapeutics
- Setmelanotide clinics update: Dr Elizabeth Forsythe, Gt Ormond St Children's Hospital
- Cognitive and Behavioural Profiles in BBS: Dr Gabrielle Blumer
- Future planning: Philip Warford, Renaissance Legal

In the afternoon we will split into two groups, with a separate programme for adults living with BBS, and for parents of children and young people living with BBS, to allow for more focused sessions.

Children and young people

For the children and young people, we are planning a fun day out at Drayton Manor Theme Park. For our younger guests, an on-site creche will be available.

Contact Information

If you have any questions about this event, please email admin@bbsuk.org.uk and we will get back to you as soon as we can.

Let's come together to connect, learn, and share as a community. Team BBS UK can't wait to meet you there!

Team Member Updates

Trustees

At the BBS UK Annual General Meeting (AGM), held on Saturday 9th November, members voted as follows:

- Emma Hughes was elected Chair.
- Chetan Meshram was re-elected as Vice-chair.
- Tazeem Abbas was re-elected as Treasurer.
- Dianne Hand was re-elected as Secretary for one year.
- Rachael Foley was re-elected as a trustee.
- Bilaal Ali was elected as a trustee.
- Liam Wilkie, Shehla Mohammed and Gareth Owens will continue as trustees.

Emma Hughes has worked at the Thomas Pocklington Trust, a national sight loss charity, since 2008 and has been at the forefront of the development and delivery of their services. In her role as Director of Services, Emma is passionate about ensuring that blind and partially sighted people are at the centre of all their activities. Emma believes that the voices of lived experience are essential in providing responsive, effective services and support. Emma is excited about the prospect of working alongside the trustees, staff team and volunteers at BBS UK, and to supporting their inspiring work with people with BBS, their families, and carers.



Bilaal Ali has BBS and is registered blind. He is currently in the placement year of an Accounting and Finance degree at



Sheffield Business School, is an avid sports enthusiast and plays blind football and tennis. You might recognise Bilaal as he shared his personal perspective at our online conference in May 2024.

Bilaal says, “having lived with BBS since birth, I have valuable experience in key areas like education, employment, sports, and social connections. I’m passionate about supporting others with BBS, helping them feel comfortable with the condition and enjoying their lives. I would also like to enhance the social aspect of BBS UK, through more events and meet-ups.”

You can read all our trustees’ profiles on our website.

Thank you and goodbye to Laura and Stefan

Having reached the end of their terms as trustees, we said goodbye to Laura Dowswell and Stefan Crocker.



Stefan has been a highly valued member of the Board for nine years and has served as Vice-Chair and Chair during this time. Throughout his trusteeship, Stefan has been the biggest cheerleader for BBS UK and its services, getting behind all new ventures with enthusiasm and with the aim of improving awareness and access for all.



Stefan has also been an active volunteer at social events, information days and conferences, both at home and abroad, representing BBS UK in Germany and the United States. We thank

Stefan for all that he has done for BBS UK and its membership over the years.

Laura has played a pivotal role in shaping BBS UK over the past nine years, serving first as Treasurer and then Vice-Chair, before taking up the post of Chair for the past three years. When Laura initially joined the Board, the Charity was facing a number of governance and operational challenges. The Charity was in need of experienced leadership, strong financial stewardship, and a can-do/will-do approach to take that forward.

As Treasurer, Laura's extensive experience in charitable finance ensured that we achieved the highest levels of financial transparency and accountability, which were crucial in strengthening our governance.

Laura's leadership has been equally transformative, providing invaluable support to her fellow trustees and staff team. In her role as Chair over the past three years, Laura has led the Board and supported Tonia and the team to successfully deliver on our three-year strategy and has been instrumental in our current strategy review; under Laura's leadership, the Charity has grown stronger, more resilient, and better prepared for the future.

On behalf of the Board, staff team and membership, we give a heartfelt thank you for her years of service, for her leadership, and for the passion and commitment she has given to BBS UK.

Goodbye to Samantha Serido

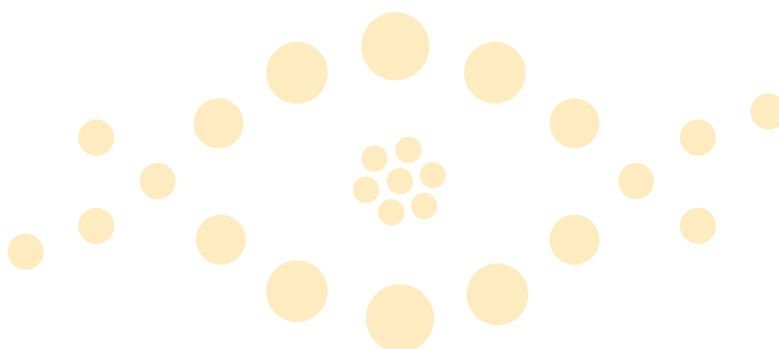
In September, we were sad to say goodbye to Samantha as Fundraising and Communications Officer. During her time at BBS UK, Samantha submitted many grants and trusts applications, as well as developed fundraising promotional materials and fundraising and communications strategies.

Before her departure, Samantha said: "It has been an honour to contribute to the success of our BBS UK community. I am particularly grateful to all who shared their personal stories with me, whether sharing them while completing incredible fundraisers for our charity or while helping to build awareness about life with BBS. It has been truly inspiring to see how a community that is considered rare and small in size, is able to deliver such huge support to one another."

Thank you Samantha and we wish you all the best in the future!

Annual General Meeting

On 9th November 2024, we held our Annual General Meeting (AGM) on Zoom where members gathered online to vote on key decisions. Thank you to our members, staff and trustees who gave up their time to attend. The minutes from the AGM are available on our website, along with the Trustees Annual Report and Accounts 2023-24.



Fundraising Roundup

Looking back at 2024, we are incredibly thankful for the wonderful support we have received from you, our generous fundraisers, donors, volunteers, and supporters. We wouldn't be able to provide the essential support and information services that we do without your help and this fundraising round-up highlights some of the ways that you have helped us to bring hope and positive change to those who need it most.

Horse Riding Fundraiser



Tanja, her daughters, and friends completed an incredible 100km riding challenge during the month of June 2024 and raised a whopping £1,660 for BBS UK!

Tanja's daughter Anayah has BBS; she is pictured riding her horse and is at the front of the group photo. It was Anayah's birthday in June, so they made it extra special by going on a 10km ride over a couple of days, followed by a delicious picnic in the park.



As they approached the end of the challenge, Tanja said, "we've almost completed our challenge and are so grateful for everyone's donations and support. I want to say a special thank

you to BBS UK. I really hope the donations make a difference to you as you make a difference in so many people's lives."

Thank you to all the wonderful fundraisers who supported this challenge, human and animal: Tanja, her daughters Anayah and Alisa, Kathy, Erika, Jade, the dogs Bella and Kane, and the horses Tyson, Lilly, and Star.

Wilkie's Warriors Take on the Kiltwalk

On the 11th of August, young Caitlin Wilkie along with some of her friends and family, took on the Dundee Kiltwalk to raise funds and awareness for BBS UK.

The Kiltwalk is Scotland's largest mass walking event with people wearing traditional Scottish kilts to raise funds for charities. In the words of her family, Caitlin "took on the wee wander with her sister, Niamh and their daddy, Liam, while mum, Ash, took on the mighty stride with a few friends and neighbours." They smashed their £600 target, raising a wonderful £800 for BBS UK!



The Wilkies are avid supporters of BBS UK, with Liam, serving as Trustee and Ashley, a member of our Service User Advisory Board. Thank you to the Wilkies for your incredible fundraising efforts and dedication to the Charity!

Graeme's Golf Fundraiser

By Graeme Roache

Graeme Roache showed his support for BBS UK and our community by partnering with his golf friends to raise over £2000 for our Charity. Graeme is a member of the Service User Advisory Group and contributes to improving BBS UK's services. The following is Graeme's report about the event:

The fundraiser was organised by my dad's friend, Donnie Bellshaw, who works for JACOBS UK Limited. The company donates annually to various charities, and we asked if they would support BBS



UK last year. To our great delight they said yes! With Donnie's fantastic organising skills this led to a Golf Tournament being held at Dumbarton Golf Club, where my dad and Donnie are members.

There were nine teams of four playing an 18-hole Texas Scramble format. Entry for the day included a traditional Scottish morning roll and sausage, and a delicious two-course dinner at the end of play. A trophy was presented to the winning team, with prizes awarded for the 'longest drive' and 'nearest the pin.'

My mum and I were responsible for selling the raffle tickets. There was a huge range of prizes donated by JACOBS, family and friends, the players, and from Dumbarton Golf Club. A fun day was enjoyed by all, and we helped to raise awareness of BBS UK.

The fantastic sum of £1,850 was raised on the day. An additional £200 was personally donated by Donnie Bellshaw, which was doubled by JACOBS giving a grand total of £2,250.

The photo shows Donnie presenting me with the cheque.

Thank you to Graeme, his family, friends, and everyone at the golf club!

Grant and Trusts

We have worked hard this year to increase our income, submitting several grant applications with some success. A huge thank you goes to the funders supporting us, including:

- **The Eveson Trust**, which has provided funds towards our Transition Support Service. This grant award is vital in enabling us to offer guidance and support for young people with BBS transitioning into adulthood. More information about the success of our Transition

Support Service can be found in the Clinics Update article.

- **National Lottery Awards for All** are funding our pilot Wellbeing Support Service, in partnership with RareMinds, a specialist non-profit, rare disease counselling service.
- **The Kaye Charitable Trust** have contributed towards costs for our Advice Service, helping us to continue to help individuals with BBS to access the local support they need.
- **Sir Jules Thorn Trust** through the 'Ann Rylands Small Grants' have provided funding towards our core costs. These are the day-to-day costs associated with keeping BBS UK up and running.
- **Rhythm Pharmaceuticals** have once again kindly contributed a generous amount towards the running of our annual conference.

Corporate Fundraising



ARAG Legal Protection Ltd generously donated on behalf of their employee who is a BBS UK member, supporter. Thank you for thinking of us!

Paul Pini, the Director of Think Expo Logistics, chose BBS UK as their charity for 2024, in support of one of their clients, whose grandchild was recently diagnosed with BBS. The Think Team attended the International Broadcasting Convention in the Netherlands and counted the steps they took. They then donated 1p for every step taken. This might not seem like a lot, but the team predicted they would walk the equivalent of 40 miles in just one evening- and they were right! Collectively, they covered a whopping 71,311 steps! The company then also generously topped up their donation, bringing the total to £1000.

We can't thank Paul and his team enough for

choosing BBS UK, and for bringing us along in spirit, across the sea to the Netherlands.

Feeling inspired? Get your organisation involved!

- **Team building fundraisers:** Your company can engage in exciting team-building fundraisers for BBS UK. Imagine challenge events, donating 1p for every step walked during an office day, or training together for a group bike ride. Get creative with events such as 'Fancy Dress Friday,' where employees contribute to BBS UK to join in the fun!
- **Check out your company's fundraising resources:** Discover the potential within your company by checking if there is a company foundation or a match-giving programme. If you joined our online conference last year, you might recall the presentation on artificial intelligence and accessibility by Hector Minto and David Prince from Microsoft. We're grateful to Microsoft for not only sharing their expertise but also making a donation in recognition of their employees' time at our event—a wonderful example of how companies can support charitable initiatives.
- **Sponsor our events or projects:** Make a lasting impact by sponsoring projects like our annual conference or Clinics Support Service. Your company's support will ensure we can continue to be there for every family affected by Bardet-Biedl syndrome.
- **Choose us as your Charity of the Year:** We would be thrilled and honoured to be selected as your company's 'Charity of the Year.' Together, we can raise awareness and provide crucial support for those impacted by Bardet-Biedl syndrome.

Our Corporate Giving poster is available to download and share on our 'Support Us' page on our website. Scan this QR code:



Countdown to Conference Fundraiser

Countdown to conference fundraiser!



Pledge to raise £100 for BBS UK and you'll receive a fundraising gift bag. Whoever raises the most will win a limited edition BBS UK hoody!



The countdown to 25th April begins now! In the run-up to our annual conference, we are challenging you all to a competition to see who can raise the most money for BBS UK. Every fundraiser who pledges to raise at least £100 will receive a BBS UK T-Shirt and fundraising pack, and the three top fundraisers will receive a limited-edition BBS UK hoody, modelled in the photo by Tom Sloane.



You can choose from one of the following ways to raise money:

1. Host a meetup

It can be as simple as inviting family and friends round for a cup of tea, cakes, and a chat, or more involved, for example by organising a party.

The Sloane family host multiple afternoon teas each year. They make their own food and drinks and charge a ticket price for attendees. You could do this at your home, or get a local business involved and ask for free use of their space. You can even ask your local supermarket if they offer any schemes for discounted baking goods when raising money for charities.

And don't let the weather stop you! The Sloanes hosted both outdoor and indoor fundraising events, raising several hundred pounds over multiple events. Thank you for your continued support!



2. Get outdoors and walk, cycle, run, or ride for BBS UK!

Many of you will remember the huge success of the We Carry the Gene campaign, where the community were asked to get out walking, running, riding, and cycling while being sponsored to reach a set distance.

You can do this too by organising a sponsored walk or other physical activity, getting family and friends and even your dogs involved, and spreading awareness of BBS along the way.

We're here to help with your fundraising efforts! Team BBS UK can help you set up your JustGiving page and will provide support along the way. Contact us at fundraising@bbsuk.org.uk.



Advice Service Report

The BBS UK Advice Service is proud to reflect on another successful year of providing critical support and vital advocacy to individuals with Bardet-Biedl Syndrome (BBS) and their families navigating increasingly complex systems of care and support.



Among some of our achievements in 2024 was the launch of the Monthly Adult Zoom Socials, warmly hosted by BBS UK volunteer Maria Dowswell. These virtual meetups have fostered a welcoming and supportive space for adults with BBS to connect, share their experiences, and build meaningful relationships. Please email rebecca.perfect@bbsuk.org.uk if you are interested in joining.

We supported the transition events in Birmingham and London, and also the London Adult Event at Sight Village. These in-person gatherings proved invaluable in helping attendees access information, resources, and a sense of community.

Over the past year, the tightening of eligibility criteria for benefits and social care has had a significant impact on many within our community. The Advice Service has seen an increase in referrals from individuals seeking help with:

- Challenging decisions made by social care agencies regarding the level of support they can access.
- Navigating benefit decisions and issues with the Department of Work and Pensions (DWP).

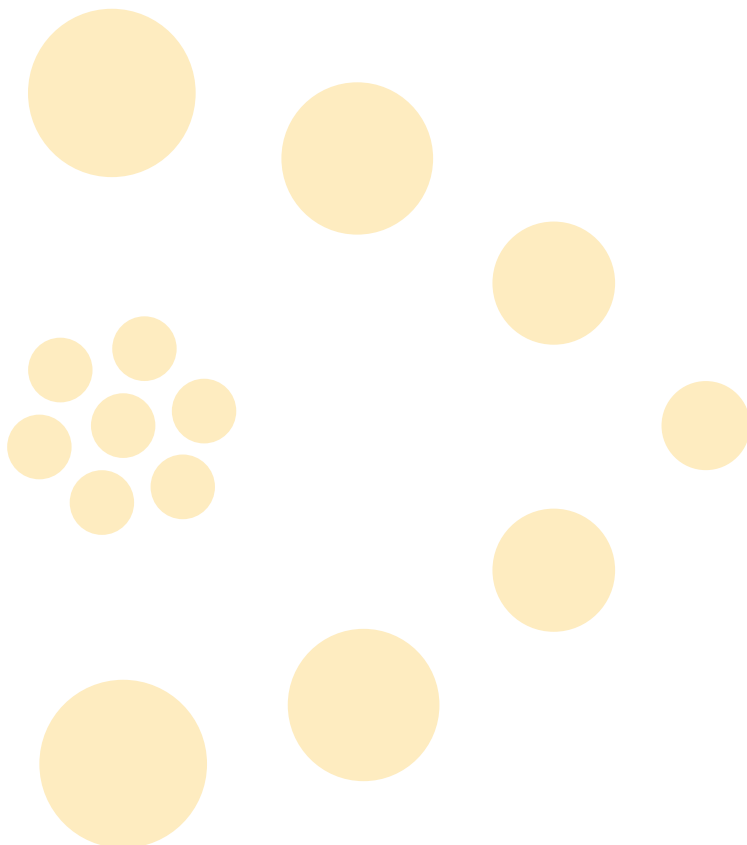
To help address these concerns, we've developed a practical guide to benefit applications, which

can be found in this newsletter, highlighting what to expect, the importance of preparation and the key requirements for reporting changes in circumstances.

This year, we also continued to offer social work placements for students from London Metropolitan University. This month, we welcomed our third social work student, Temi to our team. These training placements not only support students in building essential skills in working in the field of rare conditions, but also expand the capacity of the Advice Service, enabling it to provide personalised assistance to more individuals and families.

In the coming months, we will focus on helping individuals and families access housing, care packages, education, and transport solutions, whilst also strengthening connections with local service providers to streamline access to vital support.

We are excited to see many of you at the BBS UK Annual Conference in April.



Tips for Applying for Benefits

By Shirin Memi, Advice Service

Before completing the application form

- **Accept its a lengthy process.** Start early, plan ahead, and don't expect to get through it in one session.
- **Read the instructions before completing the application.** Go through the entire application before you start, to ensure you understand all the requirements.
- **Gather all evidence and supporting documents before completing the application.** Allow plenty of time for this; you may need to contact the professionals involved in your support and care for their input.
- **Review the eligibility requirements.** Make sure you qualify for the benefit before applying. Some benefits will have income limits, residency requirements, or other criteria that could affect eligibility.
- **Pay attention to deadline dates.** Make sure you are aware of the date by which you must return the form or supporting evidence to DWP. A deadline date for returning forms is usually accompanied with the application form.
- **Request any supporting evidence in advance.** Professionals may be busy and unable to provide evidence at short notice – some may need weeks rather than days to complete the request, and you may need to chase.
- **When requesting medical evidence** such as a letter or report, ask if it can be written specifically to address any difficulties in the application form. For example, a physiotherapist could write about any one-sided weakness that makes it difficult to get washed or dressed independently.

Completing the application form

- Make sure everything you say when applying is **correct, accurate, honest, and transparent.** The DWP will check with evidence providers, and providing incorrect information can lead to delays, rejections, or penalties.
- **Keep a copy for yourself.** Just over half of the applications made are successful - a copy of your application will be essential for any assessments and future appeals. Keep copies of all supporting documents.
- **Double check** the application before submitting.
- **Submit early.** Try not to wait until the deadline date. An early submission can avoid issues with deadlines.

REMEMBER: Once you are in receipt of a benefit, it is your responsibility to inform the benefit agency of any change in your circumstances, which may include:

- Moving in with someone, or moving to somewhere different
- Getting a new job
- A change in your work hours
- A child reaching 16 years old
- A change in another benefit
- Staying in hospital/residential care for a period of time
- Getting married
- Starting or stopping education or training
- Starting a residential school placement, i.e., the RNC.

If in doubt, check with the Department of Work and Pensions. Failure to report a change in circumstances means you could be overpaid, and will have to pay back the money, and in some cases pay an additional penalty.

Useful Organisations:

Turn2us

Turn2us is a national charity offering information and support to people experiencing financial insecurity. The number to call is 0808 802 2000.

There is lots of information on their website that could be helpful to you, including a free Benefits Calculator and Grant Search. These can help you find out what support you may be eligible for, including welfare benefits or charitable grants you don't have to pay back. www.turn2us.org.uk

Gingerbread Single Parent Helpline

Gingerbread provides advice and practical support for single parents, including advice on benefits and tax credits. You can call free on 0808 802 0925 or visit www.gingerbread.org.uk

Disability Support Project

The Charity provides support for advice and guidance for benefits, tribunals, applications, form filling, financial planning, and mental wellbeing. Call the helpline on 01527 351021, or visit the website at www.dspuk.org.uk

Advice Local

Advice Local has an online search facility to find a local advice agency by postcode. www.advicelocal.uk/find-an-adviser

Citizens Advice

www.citizensadvice.org.uk/benefits

Find your local Citizens Advice centre

www.citizensadvice.org.uk/about-us/contact-us/nearby

MIND

The MIND welfare benefit helpline supports anyone with mental health problems struggling to navigate the benefits and welfare system.

Call 0300 222 5782 or go to: www.mind.org.uk/information-support/tips-for-everyday-living/money-and-mental-health/claiming-benefits

Find your local branch of MIND

www.mind.org.uk/information-support/local-minds

Carers UK

www.carersuk.org/help-and-advice/financial-support

Carers UK: 020 7378 4999

Carers Scotland: 0141 378 1065

Lines are open Monday to Friday, 9am – 6pm (including Bank Holidays).

For more complex queries or more detailed guidance, Carers UK suggest you contact them on the email Helpline advice@carersuk.org

Age UK

The Age UK advice line is open 365 days a year.

It gives advice to older adults about benefits, retirement, hospital stays, and care homes. Please call Age UK Advice Line on 0800 678 1602

Lines are open 8am-7pm.

The website has information about cost-of-living support for older people, explains benefit entitlements and how to claim:

www.ageuk.org.uk/information-advice/money-legal/benefits-entitlements

Find out what services and support your local Age UK offers near you

www.ageuk.org.uk/services

SCOPE

Information on Disability Benefits including how to find your local Disability Centre.

www.scope.org.uk/advice-and-support/benefits

Benefits and Work

Advice on claiming ESA support and other benefits. Please note some benefit guides need to be purchased.

www.benefitsandwork.co.uk

Rethink

Mental health charity that provides a diverse range of mental health services and support groups.

<https://www.rethink.org>

Mental Health & Money Advice Service

The Mental Health and Money Advice service helps and supports people understand, manage, and improve their money and mental health issues.

www.mentalhealthandmoneyadvice.org/en/welfare-benefits/

Money Helper

This is a free service provided by the Money and Pensions Service. They offer free and impartial money and pensions guidance.

www.moneyhelper.org.uk/en

Useful Online Resources:

www.benefitsguide.co.uk

A free to use resource which provides clear and comprehensive information on all available UK welfare benefits as well as guidance on benefit applications, appeals, emergency assistance & legal help.

www.gov.uk/browse/benefits

Government website which gives information relating to all benefits and entitlement and how to claim.

Benefits calculators

You can use an independent, free, and anonymous benefits calculator to check what you

could be entitled to. This will give you an estimate of:

- the benefits you could get
- how much your benefit payments could be
- how your benefits will be affected if you start work or increase your hours
- how your benefits will be affected if your circumstances change - for example, if you have a child or move in with your partner

Turn2us benefits calculator

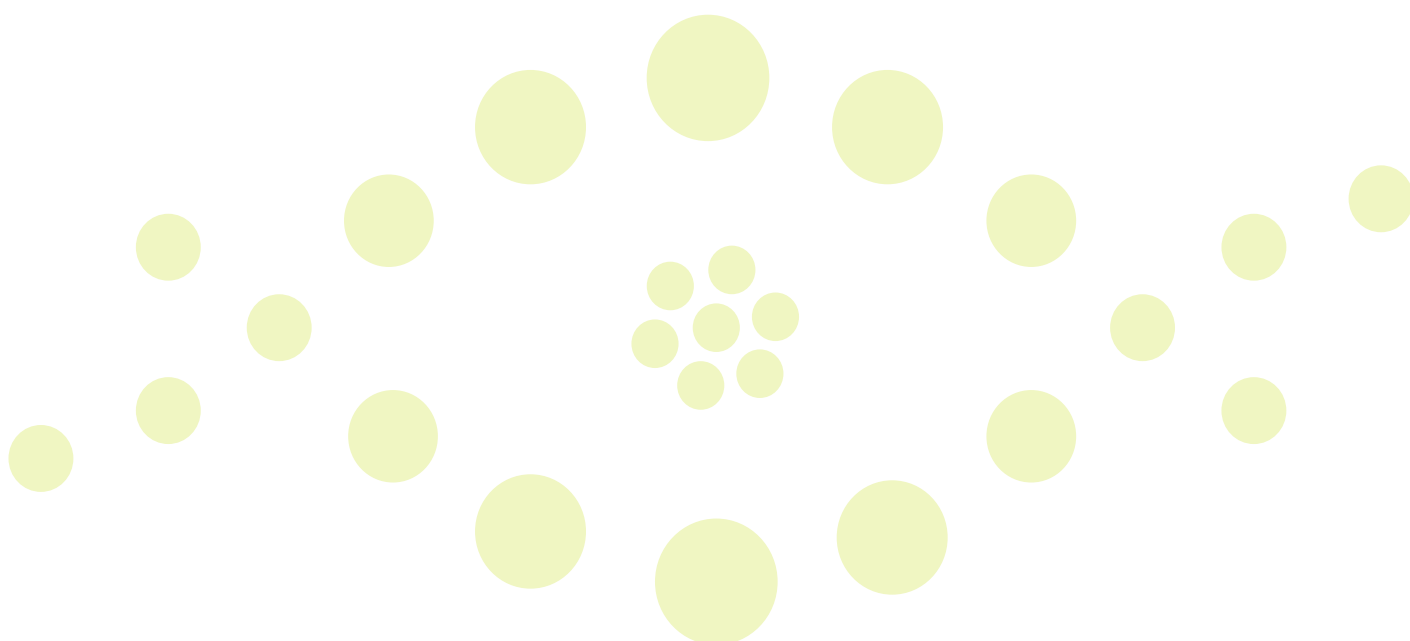
Turn2us is a national charity offering information and practical help to people facing financial insecurity.

www.benefits-calculator.turn2us.org.uk

Policy in Practice better off calculator

The Better Off Calculator allows you to see how much benefit you are eligible for and how much better off you might be in work. You can also see how changes in your household budget can affect your income.

www.betteroffcalculator.co.uk/login



BBS UK Clinics Support Service



The NHS commissioned BBS clinics offer both face-to-face and virtual appointments to those living with Bardet-Biedl syndrome. At each clinic, patients are seen by an ophthalmologist (eyes), nephrologist (kidneys), dietician (food and nutrition), clinical psychologist, geneticist (genes), speech and language therapist and endocrinologist (hormones). BBS UK is contracted to provide support and facilitation services, helping people living with BBS and their families before, during and after the clinics.



After a well-deserved break over Christmas and New Year, the team are back and ready to provide more top-tier support to the community. Feedback from the clinics has been overwhelmingly excellent and we thank everyone for taking the time after their appointment to complete the online form. Here's some positive feedback from the last quarter!

"Fantastic support from start to finish. Thank you."

"I appreciate everyone that was at the clinic you are so supportive and helpful."

"Really well coordinated, it ran smoothly, and the support team were helpful and empathetic."

We continue to see an increase in new referrals, with higher numbers of new families coming to the clinics. This means more people with BBS are getting the support they need. The four centres are doing all they can to ensure timely appointments for everyone, including making good use of telemedicine appointments.





Blood Tests: All blood tests now take place in the Blood Test Centre, located on the ground floor of the South Wing. BBS clinic appointments are still held at the Rare Disease Centre on the first floor. Patients will receive a text from Swift Queue, with the time of their blood test, which is also on the appointment letter from GSTT. On arrival at the Blood Test Centre, patients just need to give their name—no forms or paperwork are needed!

MyChart: Patients who are not yet signed up for MyChart, the electronic patient record system, should let the team know at their next clinic appointment and they will help set it up. They can also set up proxy access if someone else helps to manage appointments, reports, and other information.

Patient Amenities: Breakfast, lunch, tea, coffee, and water is provided for patients on clinic days. Patients should let the team know of any dietary requirements in advance.

For GOSH patients:

Kath Sparks is the Clinical Nurse Specialist at GOSH and is available through the MyGOSH app for any queries or support.

Location Change: Friday morning MDT appointments have moved from the second floor to the first floor (right next to the Blood Test Unit) in the same building.

Meals and Refreshments: Unfortunately, due to Trust policy, we are unable to provide lunch at GOSH, so patients should bring food and drinks for the day! We will, however, continue to provide tea and coffee. If you have a travel cup, we'd love it if you could bring it along to help us stay green!

For BCH patients:

Birmingham is a Clean Air Zone, so if you are travelling by car, please check on the 'Brum Breathes' website to see whether charges will apply (www.brumbreathes.co.uk). Hospital attendees have been granted an exemption from charges until further notice; exemption vouchers can be collected from the main hospital reception during your visit. BBS UK cannot cancel any

BBS clinic appointments are a truly precious resource for the ongoing care of everyone living with BBS and take a lot of time and resources to organise.

We urge all patients to attend their scheduled appointment, however we understand that cancellations are sometimes inevitable. If you do need to cancel or rearrange, please contact your Patient Liaison Officer as soon as possible, so we can offer the appointment to others. If you are open to last-minute cancellations, please let us know.

Thank you to the BBS clinicians and everyone involved in the clinics for their hard work and dedication!

News updates

For GSTT patients:

We are excited to introduce Katie White, our new Clinical Nurse Specialist (CNS). If you need to get in touch with Katie, you can call her on **07546 762 686** and leave a message. She will be happy to assist you.

charges or fines issued by Birmingham City Council, so please plan ahead to avoid issues.

Travel Expenses

You may be eligible for help with travel costs if you are on a low income and need to make a journey to receive NHS care after being referred by a GP or hospital doctor. Full information about how to claim travel costs can be found on our website by scanning the QR code.



For those on Universal Credit or Employment and Support Allowance (ESA) please note the following:

- **Universal Credit (UC):** You will need access to your online UC journal to share the necessary information with the cashiers.
- **ESA:** Make sure your document is up to date. It should be less than six months old and include your current address.

If you don't have the right documentation with you on the day, we can provide you with an HC5 (T) form to complete and make a claim after the clinic date. Please note that reimbursements can take up to 12 weeks to process, and the cashiers have a limit on how much they can cover.

Transition Events

Moving from children's clinics to adult services is a big step, and it's natural for young people and their families to feel a bit nervous about the change. New doctors, a new schedule, and even a new hospital—there's a lot to think about! That's why we've been holding transition events to make the process as smooth and stress-free as possible for our young patients.

These events are a chance for young people and their parents or carers to learn all about the adult clinics, explore the hospital, and meet some of the friendly staff they'll see there—all with the BBS UK team on hand to guide and support them every step of the way.

In November, we hosted a young adults' transition event for Birmingham service patients, and it was a great success. To kick things off, we arranged a fun and informal social evening of bowling the night before the clinic. It gave everyone a chance

to relax, connect, and show off their bowling skills before heading to their first adult clinic appointment at University Hospital Birmingham. We were thrilled to see 12 young adults successfully transition from children's services to adult services during this event.

For our first transition event for London, four patients arrived the day before their clinic appointments and took part in a walking tour of the hospital. The tour included key areas like the Blood Test Centre, Eye Department, and Patient Transport, as well as a visit to the Rare Disease Centre where BBS appointments are held. Along the way, they met friendly staff members and even had the opportunity to speak with the Eye Clinic Liaison Officer for GSTT.

To unwind, the group enjoyed an activity on Southbank, playing Crazy Boules together in a supportive and lively atmosphere. It was wonderful to see everyone, including parents and BBS UK staff, bonding over the game. The evening wrapped up with a shared meal before everyone returned to the patient hotel. The next day, their first BBS appointments in adult services ran smoothly, with the group continuing to encourage and support one another throughout.

For Birmingham service patients who cannot attend due to work/college commitments, we are looking to provide an alternative, in the form of a virtual tour and a meet the team virtual session.

We are already looking ahead to welcoming our next group of young people this summer. We hope they'll find the free event just as helpful and enjoy building connections with their BBS peers. Transitioning to adult services doesn't have to be daunting—with a little preparation and support, it can be an empowering and even enjoyable experience.

BBS UK have a Transition Handbook for young people living with BBS called 'My Life My Future.' This invaluable booklet has proven to be a useful toolkit of information to support older children with their independence and self-advocacy. It is also a useful resource for supporting conversations about transition and future opportunities and choices with younger children. You can find this on our website.

Service User Advisory Group

The BBS UK Service User Advisory Group (SUAG) was formed in January 2023 so that service users of the Charity can share their experience and knowledge. Membership of the group is representative of our community, including people with BBS, parents and carers of those with BBS, from England, Scotland, Wales, and Ireland, whilst also ensuring cultural representation.

The group is chaired by BBS UK trustee, Rachael Zimble, with support from trustee, Dianne Hand, and BBS UK Operations Assistant, Natalie Braunton.

What does SUAG do?

SUAG reviews BBS UK's services and provides feedback and suggestions on how to improve them and make them even better.

We are sometimes asked by external organisations, for example, researchers, sight loss charities, or pharmaceutical companies, to review documents or processes, which SUAG will also support. The work of SUAG will inform BBS UK decision making and support improvements and strategy development.

Since January 2023, the SUAG members have met four times on Zoom and provided feedback and suggestions to BBS UK about our services and projects including:

- Conference
- Newsletter

- Medical Information Handbook
- Community Audit
- BBS UK's three-year strategy

We are immensely grateful to SUAG members for volunteering their time over the past year and for sharing their suggestions for how to ensure our services are right for our community. The members reflected on the past year during the most recent meeting and had this to say:

"The group is absolutely critically important. BBS UK have always heard opinions from trustees and staff, but now we have the view of service users around the UK and Republic of Ireland."

"It is great to touch base and connect with the group and feel like you're providing something for the community."

"It's good to chat to people throughout the year who are in a similar situation."

Would you like to join?

We currently have four spaces in the group and to ensure a diverse range of service users and views, we are currently looking for **male** applicants, including those living with BBS, and fathers of children with BBS.

If you are interested, please email [Natalie Braunton](mailto:admin@bbsuk.org.uk) at admin@bbsuk.org.uk.



BBS UK Ireland Regional Group

A key focus for the Ireland Regional Group core team in 2024 was seeking ways to improve access to healthcare services for BBS families living in Ireland. We want to share the progress made, seek your input on shaping our activities for the coming year, and encourage everyone to get involved.

Lindsay McKnight, Anne Smith, and Tonia Hymers have been meeting regularly with Jackie Turner, Genetics Counsellor at the Clinical Genetics Centre at the Mater Hospital, Dublin, to address two critical goals:

1. **Enhancing Access to BBS Clinics in the UK via the Treatment Abroad Scheme**

We've received conflicting reports about the availability of repeat visits to the UK BBS clinics. While some families have accessed multiple appointments, others have faced barriers after their first visit. Ensuring fair and consistent access for all families is a priority. If you have navigated the system to secure return visits, we need your insights to help us identify best practices and advocate for equitable access.

2. **Developing a BBS Multi-Disciplinary Clinic in Ireland**

We're working towards establishing a dedicated BBS clinic within Ireland to reduce reliance on overseas services. Earlier this year, we conducted a review of services to inform the business case. Your input has strengthened our position and will be instrumental in discussions with healthcare decision-makers.

Representation in the HSE Rare Disease Strategy

We're proud to have Gillian Stafford representing BBS and other rare disease communities on the Health Service Executive (HSE) Rare Disease Strategy Steering Group. Gillian's involvement ensures that the voices of individuals with BBS are heard at a national level. While progress has been slow, the draft strategy is expected to be ready for review shortly. This is an important step forward in addressing the broader challenges faced by those with rare diseases.

Fundraising: Supporting Our Mission

In 2023, Team Ireland achieved incredible success with the Carry the Gene campaign, demonstrating the strength of our community; this year, we are shifting our focus to corporate fundraising.

Many companies offer staff giving or corporate sponsorship programs, and we'd love your help to tap into these opportunities. If you or a family member works at a company that supports charitable giving, consider nominating BBS UK as a beneficiary. We've created a comprehensive information leaflet to make the process easy – please reach out if you'd like a copy. Every contribution makes a meaningful difference in supporting our work.

Community Engagement

As we start planning activities for 2025-26, we are keen to hear your thoughts and ideas for the Ireland Regional Group:

1. **Ireland Regional Group Social Zoom**

We meet regularly via Zoom to connect with each other, share our experiences, and help shape the group's activities. Whether you have suggestions for events, advocacy priorities, or areas where you need support, we want to hear from you.

2. **Social Media**

We have a (private) Regional Group Ireland Facebook page which enables us to keep in touch with each other on a day-to-day basis. We would love to engage with more of you there – search for 'BBS UK Regional Group: Ireland' on Facebook.

How You Can Get Involved

The Ireland Regional Group thrives on the passion and commitment of its members. Here are some ways you can support our efforts:

- **Attend the Social Zooms:** Share your ideas and help us plan meaningful activities for 2025.
- **Advocate for Fair Clinic Access:** Share your experiences of accessing the UK BBS clinics to help us build a case for equitable access.
- **Fundraise or Donate:** Explore corporate sponsorship opportunities or participate in community fundraising initiatives.

- **Spread the Word:** Encourage others in the BBS community to get involved and join our events.

A Heartfelt Thank You

We are truly grateful to the BBS UK Ireland Regional Group core team for their dedication and hard work. Their efforts are making a tangible difference in the lives of BBS families across Ireland. But we can't do it alone. Your support, feedback, and participation are essential to driving progress.

Let's work together to make 2025 a year of growth, connection, and impact for the BBS community in Ireland. Whether it's attending an event, sharing your story, or exploring new ways to contribute, every action matters. Thank you for being a part of this journey with us.

For more information about any of the above, please contact: Lindsay.mcknight@bbsuk.org.uk or admin@bbsuk.org.uk

Running for Mary:

Michael's Marathon Mission



This year, the London Marathon will hold special meaning for our community, as one of the runners will be taking on the challenge for a deeply personal reason. Meet Michael O'Sullivan, who is lacing up his running shoes to represent BBS UK in support of his sister, Mary.

Mary, now 34, has Bardet-Biedl Syndrome, which has shaped her life in profound ways. Diagnosed as a young child, Mary faced significant challenges, including a kidney transplant at just eight years old and a lifelong struggle with her eyesight. Today, Mary relies on her cane to navigate the world, but her resilience and determination continue to inspire everyone around her.

For Michael, BBS UK has been a cornerstone of support for their family. Reflecting on their journey, he shared:

"BBS UK has been a constant support to our family through the years, creating a sense of

community and providing huge reassurance to my parents when Mary was first diagnosed. We've attended the annual conference in Northampton since I can remember. Raising funds for this Charity is my way of giving back to the organisation that has done so much for us and countless others."

Michael's connection to Mary is as strong as ever. Weekly walks have become their shared ritual, helping Mary rebuild her confidence and combat isolation. His commitment to her well-being is the same drive fuelling his marathon goal: raising £5,000 to support the work of BBS UK. Michael plans to reach this target through generous donations and a fundraising event, inspired by his sister's strength and the sense of community the Charity has given them.

Michael also hopes his run will inspire other families navigating the challenges of BBS. He says, "It would mean a huge amount to my family to help repay our gratitude and contribute to the incredible work BBS UK does to support families like ours."

We are honoured to have Michael representing us in the 2025 London Marathon. His dedication not only highlights the power of family but also demonstrates the importance of the BBS UK community. We hope you will join us in cheering Michael on every step of the way.

If you'd like to support Michael's fundraising efforts, you can donate through the JustGiving page by scanning the QR code.



The New World of Gene Therapy

Condensed and reprinted with kind permission from RARE Revolution Magazine.

Why is everybody talking about gene therapy?

Gene therapy is one of the fastest growing areas of healthcare. Rather than just treating symptoms, gene therapy has the potential to get right to the core of a disease by modifying a person's genes. This can be achieved by replacing a disease-causing gene with a healthy copy of the gene or inactivating or removing a disease-causing gene.

Gene therapy research began over 40 years ago and while progress has been slow to reach patients, several products are now authorised for use in a range of conditions with that number expected to increase over the next few years.

Gene therapy offers tremendous hope, particularly in the rare disease setting where treatments are often unavailable or only provide short-lived quality of life (QoL) benefits. With gene therapy, there is potential for cure, or restoration of function, that can lead to significant improvements in quality of life. Consequently, patient communities are keen to understand what these therapies could mean for people living with these genetic conditions. But reality and expectations are not always aligned, and health systems will need to work through complex social, ethical, health and economic concerns.

Gene therapy may not offer a cure or be suitable, or indeed available, to everyone. Gene therapies frequently make news headlines because of their extremely high price tag, and even when they are approved by regulatory authorities, unfortunately, there is no guarantee of access for the individuals that most need them. Also, patients and caregivers may not fully understand gene therapy and its benefits may be overestimated.

What kind of an impact could gene therapy have on people living with a rare disease?

In many cases, it is hoped that gene therapy will result in a "cure," abolishing symptoms so that

"management" of a condition is no longer required. For some conditions this may be the case, but in other conditions, patients may overestimate the benefits and expect a "cure," but in reality, their condition is improved but not eliminated. Safety considerations are also of paramount importance and will be a factor for many people. Managing the difference between expectations and reality for all those concerned is essential.

How could gene therapy impact the wider healthcare systems?

Gene therapy has the potential to significantly improve the lives of people affected by a genetic condition; however, the cost of the treatment represents a significant hurdle. For example, a gene therapy recently approved in the US has a price tag of \$3.5 million. For institutions such as the UK's National Institute for Health and Care Excellence (NICE), funds are limited and so understanding the impact of gene therapy and its cost effectiveness is critical.

Even in countries that can offer gene therapy, not all individuals will be eligible for treatment, not just within a clinical trial but also in the real world after approval. For example, age may be a limiting factor, or in the case of gene therapy using viral vectors, previous exposure to the wild-type virus. These restrictions could create a two-tiered system, for those that have had gene therapy versus those that have not.

Despite these challenges, the huge potential for gene therapy remains, with the possibility for some previously undertreated diseases to become a thing of the past, with a real reduction in demands on healthcare services for some conditions.

Key Challenges

Gene therapy has come on in leaps and bounds, but challenges remain. The process of developing a gene therapy can be very lengthy and patients and their families sometimes have unrealistic expectations. Even if approval is granted, cost can be a huge barrier to access.

Education is key. This is complex science and can be difficult for patients, Health Care Professionals, and regulators/funding decision-makers to understand. We need to have meaningful discussions about safety and durability. We need to determine what happens if the treatment fails. What are the ethical considerations and motivation for treatment? Does everybody want to be treated in this way? The diseases that potentially could be treated with gene therapy are typically poorly served by current therapies and the burden of disease affecting patients and their families is immense.

Patient communities must be involved in the decision-making process. Open discussions, debate and education are needed, and the support of patients and patient advocacy groups can help to drive these discussions forward, championing the cause of those without a voice. The future

is incredibly exciting and the potential for gene therapy is huge for patients and caregivers, but to deliver a global solution and true health equity, pharmaceutical companies, payers, policymakers, and advocacy groups need to work together.

This article originally appeared in the 8th of March 2023 edition of RARE Revolution Magazine, written by: Amanda Henkel, practice area lead, rare diseases; Jo Fearnhead-Wymps, SVP patient engagement; Alex Morton, SVP patient engagement, MEDiSTRAVA, an Inizio company. You can visit their website at www.rarerevolutionmagazine.com and read the full article by scanning the QR code:



Sailing Vision Week

By Abi Clapp

Sailing Vision Week is an inspiring event that takes place each year, giving up to 60 blind, partially sighted, and visually impaired people the chance to develop sailing skills. It is run by Sailing Vision Trust, a UK-based charitable trust and community with a goal to make water sports accessible to everyone. Abi Clapp is a BBS UK member and service user, and she shared with us her experience at sea.

Discovering Sailing

Abi's interest in sailing began through VICTA, a national charity that provides support to children and young adults up to 29 years old who are blind or partially sighted, and their families. VICTA gave Abi a list of activities she could try, and sailing stood out to her as she had never sailed before. Abi loves new experiences, so she didn't hesitate. She was "100% sure she wanted to go" and was excited from the moment she signed up.

Life on Board

During Sailing Vision Week, Abi spent a whole week on a boat with another visually impaired participant who she already knew

from other activities, along with four experienced Sailing Vision Trust staff, who supported them at all times.

While stepping onto the boat for the first time was a bit tricky, Abi quickly adapted to life at sea. The team made sure she felt comfortable and confident while learning tasks like lifting the sail and tying specific knots.

A typical day involved getting the sail out, learning the parts of the boat, and relaxing with her



crewmates in the evening.

Abi said being out on the water was “nice and peaceful.” With just the sounds of the waves crashing and the wind blowing through her hair, Abi felt calm and happy.

Challenges and Triumphs

Of course, sailing came with its challenges. Abi found it hard to put the sail up when it was wet and slippery, and walking on the boat became harder as well. With the support of her team, she overcame these obstacles and learned valuable new skills. Despite the challenges, Abi felt a sense of accomplishment after each task, which boosted her confidence.

Favourite part

For Abi, the most fun part of the week was when they were allowed to jump off the back of the boat and swim in the sea! The water was very cold, but

it was still lots of fun.

Abi believes that activity trips like Sailing Vision Week can help build confidence and independence. The experience has certainly inspired her to try even more adventurous activities... when asked if she would go again, her answer was an enthusiastic “Yes!”

Her advice to anyone hesitant to try sailing or a similar adventure? “Try and have a go at everything.”

Abi’s story is a reminder of the power of stepping outside your comfort zone and trying something new. Sailing Vision Week gave her the chance to learn, grow, and have fun, all while being part of a supportive community that celebrates independence and adventure.

Are You a Member of BBS UK?

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

Benefits of Membership:

BBS UK’s members are at the heart of our work. Become a member and you will join a growing group of people who play a part in everything we do. For BBS UK, having dedicated members enables us to meet our aims and objectives more fully. Engaged and involved members ensures transparency, credibility, and expands our impact through increased volunteering and community engagement. The collective strength of our membership not only benefits individual members, but the BBS community as a whole.

Members also have:

- The opportunity to participate in decision-making and shape the direction of the Charity
- The chance to vote to elect BBS UK trustees and to become a trustee
- Access to BBS UK projects including the IT Equipment Fund and Chris Humphreys Memorial Fund

In addition to becoming a member, you can support 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

Membership is entirely **free** to ensure that everyone who needs our support can 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.

How will I know if I am already a member?

To become a member of BBS UK, you will have either completed an application form or consented to becoming a member when asked by one of our team. If you have been sent AGM voting information in the past, you are already a member. If you are not sure, contact us at admin@bbsuk.org.uk and we will check for you.

Talking to Children About Vision Impairment:

A Guide for Parents and Carers

This article is a condensed version of the Tough Talks resource created by RNIB, reprinted here with their permission. To access the full guide, visit the RNIB website by scanning the QR code:



Learning that your child has a vision impairment is a deeply emotional experience. For many parents, the hardest part is knowing how to discuss it with their child. These conversations, though challenging, are vital for building trust, confidence, and a sense of agency in your child. With thoughtful preparation, you can approach the topic in a way that supports your child's emotional well-being and resilience.

Starting the Conversation

There's no single correct way to talk about vision impairment. Every child is unique, and as their parent, you are best positioned to know what works for them. Consider their personality, age, and how they handle new or difficult information. Whether their condition is progressive or stable, the approach you take should reflect their developmental stage and readiness to engage in the discussion.

- **Be Honest and Clear:** While it's natural to want to protect your child, openness fosters trust. It's important to avoid providing inaccurate information or making promises you can't keep. Honesty will help your child feel secure and better prepared for the future.
- **Follow Their Lead:** Many children begin the conversation themselves by asking questions. These moments provide an excellent opportunity to address their concerns in simple terms, sticking to what they need to know. Avoid overwhelming them with unnecessary details, and let their curiosity guide the depth of the conversation.

Who Should Be Involved?

While parents often lead these discussions, other trusted adults can play an important role. Your

child may feel more comfortable speaking with someone outside the family, such as a teacher, a doctor, or another professional. Peer support can also be incredibly helpful, connecting with other families or children who share similar experiences can provide a sense of belonging and practical insights.

- **Encourage Trusted Relationships:** If your child prefers speaking with a teacher or family friend, support that choice. The most important thing is that they feel comfortable and supported.
- **Involve Professionals:** Qualified Teachers of Children with Vision Impairment (QTVIs) and eye health professionals can provide expert guidance and answer more complex questions.

When Is the Right Time?

There is no right or wrong time to have these conversations. Factors such as your child's age, developmental level, and the immediate impact of their condition can all influence the timing.

- **Immediate Needs:** If your child's condition requires urgent treatment, such as surgery, new glasses, or other interventions, it's important to talk about these changes promptly.
- **Gradual Discussions:** If there's no immediate change in their daily life, you might take time to gather advice and prepare for the conversation.
- **Ongoing Conversations:** Many children benefit from an ongoing dialogue rather than a single, formal discussion. They might process information in stages, returning with new questions as they grow older, and their understanding deepens.

Facing the Future: Helping Your Child Understand Their Condition

Talking about the future can be one of the most emotionally challenging parts of the discussion.

Your child may ask difficult questions, such as, “Will I go blind?” It is important to clarify what blindness means, as many children assume it refers to total darkness. In reality, many people with vision impairment retain some usable vision, which can support independence and everyday activities.

- **Be Honest About Uncertainty:** Vision impairment often varies widely, even among individuals with the same condition. It’s okay to admit that you don’t have all the answers about how their vision might change over time.
- **Focus on Abilities:** Highlight what your child can still do and the support available to help them navigate their world. For example, assistive technologies, mobility training, and vision impairment-friendly activities can make a significant difference.
- **Encourage Optimism:** Reassure your child that they can continue enjoying their favourite hobbies and even explore new ones, such as braille, touch typing, or audio-described media. Involving them in problem-solving can empower them to take an active role in adapting to challenges.

Practical Tips for Communication

- **Plan Ahead:** Consider preparing for conversations by writing down key points or discussing them with a trusted professional beforehand. If your child’s condition involves regular hospital visits, eye health staff can help guide these discussions.
- **Prepare Short Answers for Others:** Family members and siblings may face questions about your child’s condition from others. Equip them with simple, positive responses, such as, “She has a condition that makes her eyes work differently, but she’s amazing at using apps that help her see better.”
- **Share Age-Appropriate Information:** Tailor the level of detail to your child’s developmental stage. Younger children may only need basic explanations, while older ones might benefit from understanding more about their condition.

What If They Don’t Want to Talk?

It’s not uncommon for children to avoid discussing their condition, especially if they’re struggling to process it. Forcing the conversation can lead to frustration or emotional withdrawal. Instead, demonstrate your support through actions. Focus

on solving practical issues, celebrating their achievements, and helping them understand that vision impairment doesn’t define their potential.

- **Encourage Acceptance Gradually:** Some children take longer to accept their condition, especially as they approach adolescence or adulthood. Peer support groups or role models with similar experiences can help them feel less isolated.
- **Be Patient:** Give them time to process the information and remind them that you’re available whenever they’re ready to talk.

Supporting the Whole Family

The impact of a child’s vision impairment extends beyond the individual. Siblings and extended family members also need support and opportunities to ask questions. Including them in discussions ensures they feel informed and equipped to help.

- **Prepare for Questions:** Siblings might encounter curiosity from others, such as, “Why does your brother use a cane?” Having pre-prepared answers can make these moments easier to navigate.
- **A Family Approach:** Discussing the condition as a family can promote understanding and unity. It’s also an opportunity to reinforce positive messages and share strategies for overcoming challenges together.

Resources and Support

Navigating vision impairment can feel overwhelming, but you don’t have to do it alone. There are numerous resources and support networks to help your family.

- **RNIB Sight Loss Counselling Team:** Offers professional advice and emotional support tailored to parents and carers.
- **QTVIs and Vision Services:** Can provide expert advice on education and mobility training.
- **Peer Support Networks:** Connecting with other families through organisations like RNIB can provide invaluable insights and emotional support.

Remember, the most important thing you can do is show your child that they are loved and supported. By fostering an open, honest dialogue and focusing on their strengths, you can help them approach the future with confidence and resilience.

How Exercise Helps Me Feel Stronger and Happier

Emma May is a long-standing member of the BBS UK community and is also a member of the Service User Advisory Group. Emma's story shows how exercise can make life better, helping with both physical health and feeling good inside. Her message is simple: give it a go and find what works for you!

I'm Emma May, and I live with BBS, a condition that affects my vision, balance, coordination, and weight. Exercise has become an important part of my life. It has helped me feel stronger, more confident, and healthier—both physically and mentally.

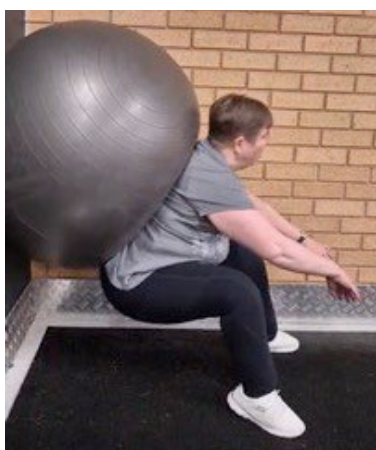
How I Got Started with Exercise

My journey with exercise began when I was very young. My parents always encouraged me to stay active, even though I struggled with coordination and balance. I couldn't ride a two-wheel bike, but I loved my tricycle. At school, I swam regularly, and when I was older, I used an exercise bike at home. My first time in a gym was through an exercise-on-prescription program. It was a big step for me, and I enjoyed it right away. Later, when we moved to Brecon, I started working with personal trainers. They've helped me build a routine that works for my body and my abilities.

My Exercise Routine

I focus on strength-building exercises at the gym. My favourite equipment includes the leg press and lateral pull-down machines, and I also use free weights. At home, I use a stepper, an exercise bike, and other equipment to stay active.

One thing I've recently started using is a large exercise ball. It has improved my posture and balance. After struggling with incontinence for years, I noticed a big improvement since adding the ball to my routine. I now sit on it when using my laptop at my desk, and it's been



a game changer for me. But make sure to get a well-made one, or you might end up on the floor like I did when the seam ripped!

My personal trainers have always been supportive, adapting exercises to suit my needs. They've encouraged me to try new things, like using the punch bag, which I now enjoy. I also like to mix things up by working out to music or following exercise routines online.

I recommend using British Blind Sport' audio-led exercise videos for those who want to work out at home. These can be found on their website at: www.britishblindsport.org.uk/active-at-home

How Exercise Makes Me Feel

Exercise has made a big difference in how I feel, both physically and emotionally. I feel stronger and have more energy. It's also helped me manage my weight, which hasn't always been easy. I still enjoy the occasional treat, but staying active helps me keep things in balance.

Mentally, exercise has been a great stress reliever. It lifts my mood and helps me feel more focused. I also enjoy the social side of it. My trainers and the gym staff are friendly, and there's often a bit of banter that makes the sessions fun.

My Advice to Others

For anyone with a condition like BBS—or any health challenge—I'd say don't let it stop you from trying exercise. Start small and find what works for you. Having a good support system, like a personal trainer, can make a big difference.

You might be surprised by what you can achieve. Exercise isn't just about improving your body - it's about feeling good in yourself and finding something you enjoy. My journey with exercise has been full of ups and downs, but it's one of the best things I've done for my health and happiness. If I can do it, so can you.



My Journey to the BBS International Conference



Stefan Crocker has been an integral part of BBS UK, serving as a trustee for nine years and regularly volunteering at conferences, exhibitions, and social gatherings. His dedication to the BBS community is inspiring, and in this article, he shares his experience traveling to the BBS International Conference in the US.

Hello, my name is Stefan Crocker, and I had the pleasure of attending the BBS International Conference in Minneapolis, Minnesota, from 26th–28th July 2024.

Preparing for Travel

I booked a direct flight from Heathrow to Minneapolis and made sure to request assistance at the airport. This turned out to be very helpful. My friendly taxi driver helped me at Heathrow by guiding me to the airport staff, who were ready to assist. They supported me through check-in, security, and took me to the gate via the passenger assistance lounge. Onboard, the crew checked in with me often to make sure I was comfortable.

A tip for fellow travellers: use bright straps on your luggage to make it stand out. It makes things quicker for both you and your assistant. I also used the Blind Square GPS app while taking taxis abroad—it was a lifesaver for finding my way.

Arriving in Minneapolis

After landing, airport staff helped me collect my luggage and find a taxi to my hotel. The hotel staff were very welcoming and even walked me to my room on the fifth floor. They explained how to contact reception if needed, though I decided to practice navigating on my own. That evening, I had dinner at the hotel restaurant. The manager read the menu aloud to me, and I tried something new: a Monte Cristo sandwich (no I hadn't heard of it either), a fried treat filled with turkey, ham, and cheese. It was delicious!

Exploring the City

The next morning, I woke up early thanks to jet lag (it was 4 a.m. in the US) though it felt like mid-morning to my body! This gave me time to get everything done before the evening's "Meet and Greet." I used a mix of Uber and Blind Square to visit local stores. Uber drivers were easy to work with, as the app lets you message them about any special assistance you need.

In one store, the staff had trouble understanding my British accent, so I used the Seeing AI app on my phone to identify products. These tools made it much easier to shop and explore.

Meeting New People

I was nervous before the 'Meet and Greet' at 7 p.m., but it turned out to be a great experience. On the way, I met Tim Ogden, the president of the Bardet-Biedl Association USA, and his family. Before the trip, I had connected with people through the 'Families of Bardet-Biedl Syndrome' Facebook group, and one of them introduced me

to more attendees. Meeting friendly faces helped me feel relaxed and part of the group.

Highlights of the Conference

The conference officially began on Friday morning with breakfast. I enjoyed fresh fruit, bagels with cream cheese or jam, and a hard-boiled egg. The sessions kicked off with introductions from Tim Ogden and Val Shephard.

Later, we heard fascinating talks by Professor Phil Beales and Dr Arleen Drack who spoke at BBS UK's online conference last year on gene therapy. These sessions were packed with information, but regular coffee breaks and opportunities to stretch, kept us all energised.

After lunch, I joined a panel about the mental health impact of sight loss. I was nervous, but Tim handed me the microphone, and I shared my story of living with BBS. It felt empowering to answer questions from the audience and connect with others.

The day ended with a fun picnic. We rode in a classic yellow American school bus to the park, where I chatted with a Canadian father whose four children all have BBS. Afterward, we returned to the hotel for more socialising. Saturday's focus was on workshops. The ballroom

was divided into smaller spaces, and we could choose from different sessions. After lunch, the pattern repeated, giving everyone a chance to explore a variety of topics. That evening, I joined a group for dinner at a local sports bar and tried a bison burger, another first for me!

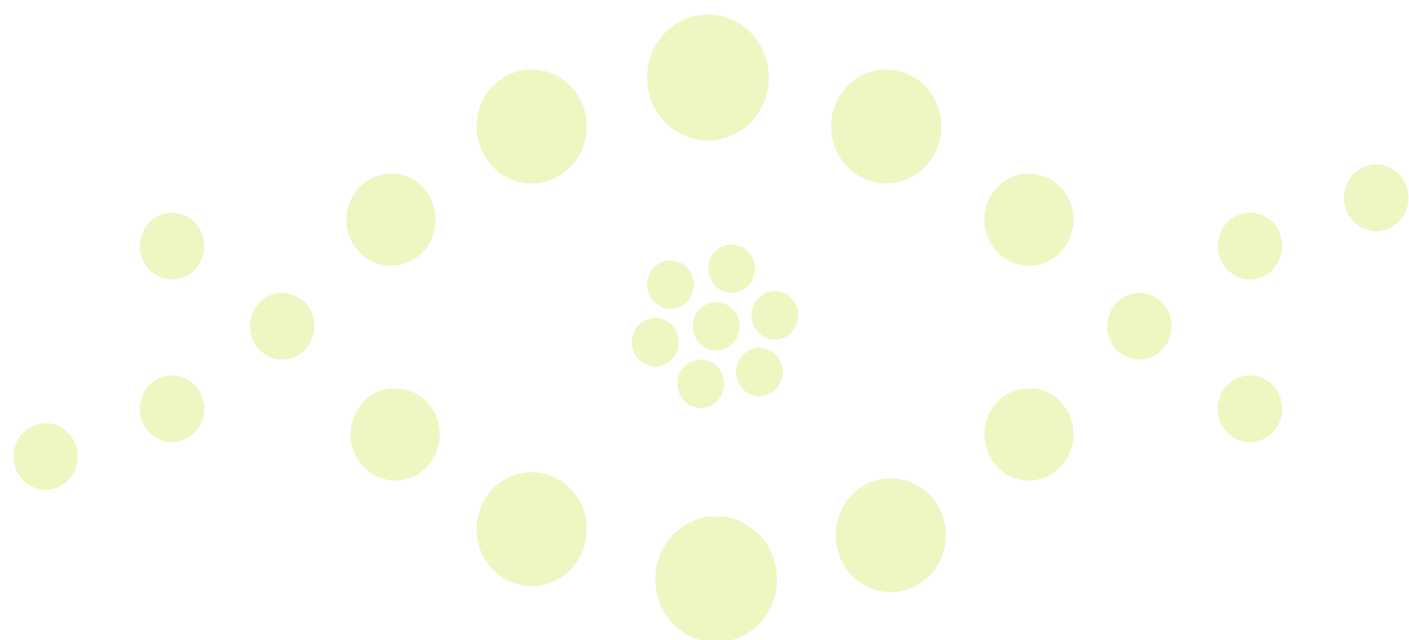
The Journey Home

On Sunday we said our goodbyes over breakfast. My Uber driver was exceptionally kind, helping me into the airport. However, there were some challenges with airport staff who struggled with English. It was a bit stressful being separated from my luggage and cane at security, but everything worked out in the end.

Once on the plane, the crew provided excellent support, and my return to Heathrow was smooth. Assistance staff there guided me to the train station, and I completed the journey home to Bedford feeling accomplished and happy.

A Message to Fellow Adults with BBS

Traveling independently as an adult with BBS can feel daunting, but with preparation and tools like airport assistance, Blind Square, and Seeing AI, it's very doable. The experience was worth every moment, and I'm already looking forward to the next BBS International Conference in 2026. I hope this inspires more adults with BBS to explore the world and join in these incredible events.



BBS UK Out and About

Team BBS UK have been out and about meeting our service users, educating people about Bardet-Biedl syndrome, making important professional connections, and learning about technology and support services that might benefit our community.

Cilia Dublin

Tonia Hymers (Operations Manager), and Laura Dowswell (former Chair), represented BBS UK at Cilia 2024 in Dublin in September. Cilia 2024 was the 6th biennial European Cilia Research meeting, and the world's largest scientific meeting for cilia research, bringing together researchers, clinicians, patients, and patient representatives from around the world.

Tonia was invited to give a presentation from a parent and patient group leader perspective and spoke about the journey of BBS UK from a small volunteer run support group to the highly valued and regarded charity that it is today. The importance of patient voice and involvement was woven throughout, as was the importance of collaboration.

Across the two days, Tonia and Laura met up with leading researchers, including Professor Phil Beales and Professor Helen-May Simera, who many of you will be familiar with, colleagues from the Ciliopathy Alliance, and from BBS organisations in the US, Netherlands, Germany, and Italy.

Attending international conferences provides invaluable opportunities to connect with others

from around the world, to drive research and improve the lives of those living with BBS.

Team BBS UK at Birmingham Children's Hospital (BCH)

Natalie Branton

In May, former Fundraising and Communications Officer Samantha Serido and Operations Assistant, Natalie Branton, went on a day trip to visit the children's BBS clinic at Birmingham Children's Hospital (BCH). It was an exciting chance for them to see the amazing care the clinic provides for people with BBS. Natalie and Samantha really enjoyed meeting the team and chatting with families, Natalie writes:

BCH is a bright, cheerful place with colourful art on the walls, a piano, and a giant bear statue in the foyer. I even stopped to take a photo with the bear!



When we arrived, we were greeted by BBS UK Patient Liaison Officers Amy Clapp and Laura Davis. They introduced us to the friendly clinic staff, including Marie McGee, who works with young people moving from children to adult services.

We learned so much about how the clinic runs. The team works hard to make sure every patient



can see all the doctors in one day. This helps families save time, avoids extra hospital visits, and reduces stress. We also spoke with families who were at the clinic. Over sandwiches and fruit pots, we heard about their experiences. It was great to see how much the families appreciated the support they received.

At first, finding our way around the hospital was tricky. The corridors felt like a maze! But after some practice, we were able to help families get to their appointments.

On our way back to the train station, Sam and I talked about how important these clinics are. They make it easier for families by bringing all the care they need into one place. Sam said:

“What I loved most about the Birmingham Children’s Clinic was how carefully everything is planned to support families. From the piano for fun, to the sensory room for quiet breaks, it was lovely to see how it all came together to help young people feel comfortable.”

I hope to meet more people from the BBS UK community during future visits to clinics!

Sight Village Birmingham

Back in July, BBS UK once again exhibited at QAC’s Sight Village in Birmingham, as part of our schedule of external events for the year.



Sight Village is an event for people of all ages who have sight loss, as well as for professionals who support them. It is also for businesses and organisations that want to improve their services

for people with sight loss. At the event, exhibitors talk to visitors and showcase their services or products.

We were supported by volunteers, Ella and Aneeba, who enjoyed speaking with visitors and visiting the other exhibitors.



Highlights from the two days included discussions on inclusive travel with Seable, healthy living initiatives with BeeZee, and networking with sight loss specialists.

Sight Village dates have been released this year, with events taking place in Cardiff (8th April), Glasgow (21st May), Birmingham (7th July), Blackpool (16th September) and London (TBC). We highly recommend attending these events, for more information, visit the QAC website: www.qac.ac.uk.

Young Person’s Activity Weekend in Birmingham

In September, we took a small group of secondary school-aged young people with BBS to Birmingham for an exciting weekend! The goal was to provide a safe, fun space where young people with BBS could meet and try out new activities.

The weekend was packed with fun, including indoor crazy golf, group meals, bowling, and a visit to The Bear Grylls Adventure, for indoor climbing, archery, and even high ropes for the bravest among us! We wrapped up the adventure with a farewell breakfast at the hotel.

Some activities were tough, but we were amazed by the young people’s determination, bravery,

and resilience. It was also a great opportunity for parents to connect and share their experiences. By the end, we were all tired but left with fantastic memories—conquering the high ropes, popcorn on the coach, and becoming Robin Hood in the archery range!



attendees at Sight Village and the Natural History Museum, a second group represented BBS UK at Sight Village, connecting with professionals, sharing information about BBS UK with the public, and learning about the many services available to people living with sight loss.

Thank you to everyone who attended and thank you to all the staff and volunteers who supported the event.

Retina UK Professional and Annual Conference

Our trustee, Rachael Zimbler had a busy and rewarding time at the Retina UK Conference.

Friday's event, the Professionals Conference, welcomed around 90 in-person attendees and many more online. Rachael connected with professionals from various organisations, including pharmaceutical representatives, eye clinic liaison officers, rehab workers, and charities including Guide Dogs, SeeAbility, the Royal Society for Blind Children, and RNIB. It was a fantastic opportunity to share information, raise awareness, and build valuable connections with teams across the UK.

Saturday's event focused on individuals living with eye conditions, as well as their families, friends, and carers. Rachael also participated in the lived experience panel, sharing her personal story about living with sight loss, how Retina UK has supported her, and how her volunteering has made a positive impact.

The conference was a huge success, filled with meaningful conversations, new contacts, and lots of awareness-raising. Thank you, Rachael, for representing BBS UK so brilliantly!



Adult Social Event in London

In November, we hosted a group of adults with BBS, in London for two days.

We enjoyed group meals, took part in fun group games, including a quiz, visited Sight Village London and explored the Natural History Museum.

Whilst a team of staff and volunteers supported



Coffee Session at Priestly Smith School: Supporting Families Together

In June 2024, Priestly Smith School hosted a coffee session in partnership with BBS UK. Priestly Smith is a specialist school in Birmingham for students aged 2-19 years, whose primary need is a visual impairment.

Laura Davis and Amy Clapp from BBS UK welcomed attendees. Families also met members of the Birmingham VI Team and Priestly Smith's pastoral team, who introduced their roles and shared how they support children with visual impairments.

Connecting Families

The coffee session offered a relaxed setting for families to:

- Meet face-to-face with BBS UK's Patient Liaison Officers.
- Visit Priestly Smith School, learn about its facilities, and meet the staff.
- Share experiences with other families, creating a sense of community.

The session was a great success in fostering connections and offering support. Parents and professionals exchanged contact details to maintain communication and build a network

of mutual support. The school distributed a signposting handbook with valuable resources for families. They also committed to adding more information to the handbook based on feedback from BBS UK and other professionals, ensuring it remains a helpful tool for newly diagnosed families. The school is continuing to strengthen its relationship with BBS UK, working together to better support families and children with visual impairments.

This session showcased the power of collaboration between families, schools, and organisations like BBS UK. Together, we can create a strong support network for children with BBS and visual impairments.



Friends of BBS UK

New Friends for Christmas

In the lead-up to Christmas, new Friends who signed up to donate at least £5 a month, received a beautiful hand-knitted gift, courtesy of our 'Knitting Nannies'. We hope everyone who signed up enjoyed their gift, as well as the knowledge that their donations help us continue our vital work for people with BBS and their families.

By raising funds for BBS UK, whether that's through a fundraising event or by making a regular monthly donation, 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. Regular gifts, no matter the size, together provide the added security of a stable income, allowing us to plan

strategically and grow our services.

To become a Friend and regular giver:

- Scan the QR code below to visit our website
- Complete the 'Become a Friend of BBS UK' online form
- Set up a monthly standing order with your bank. Make sure to use your name as a reference so we know who to thank!

Thank you to our Friends, old and new, for their generous support!



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