

The BBS UK Annual Conference 9th-11th September 2022

**Salon
Fundraiser**

**Clinics Service and
Research:**

Updates from BBS UK

**Membership
Audit:**

Results



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Foreword

Welcome to the 2022 Spring Newsletter. It certainly feels like it has been a long winter but the arrival of spring brings with it a sense of optimism and the prospect of a return to some sort of normality, after a very challenging time for us all.

On behalf of all of us at BBS UK, I'd like to wish a Happy Easter to those of you who will be celebrating it.



Over recent months, BBS UK has been busy and the staff team has been expanding. We're delighted to welcome Laura Davis and Shirin Memi to the BBS Support Clinic team and Leonie Hudson, as Admin, Engagement and Events Co-ordinator. I know they look forward to meeting you all and getting to know you. You can read their introductions in this newsletter and I'm sure you'll join me in warmly welcoming them to the team.

As the charity continues to grow in line with demand for its services, we are looking for volunteers to offer their skills and expertise on an ad hoc basis. If you would like to volunteer your professional expertise, please email admin@bbsuk.org.uk.

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

I expect that many of us are looking forward to the return of our face to face conference in September where we will be back in our old familiar 'home' of the Hilton Hotel in Northampton. The BBS UK team are already working hard behind the scenes to ensure a fantastic and varied conference programme.

Conference is a wonderful opportunity to meet and socialise with other families who truly know what it's like to live with the syndrome, and to hear from expert speakers and attend the various workshops. I look forward to seeing and chatting with many of you there.

As always, please keep in touch with us, and with each other.

Happy reading!

Laura Dowswell

Chair of the BBS UK Board of Trustees



Family Conference

Hilton Hotel, Northampton: 9th-11th September 2022

The last BBS UK 'in-person' conference was held in April 2019 and we had no idea when we said our goodbyes, that it would be 3½ years until we could all meet up again. What an incredibly difficult time it has been, but here we are, spring is bursting out all around us and we are excited to be planning BBS UK Conference 2022.

For those who will experience this opportunity for the first time, we cannot wait to welcome you to the first of what we hope will be many conferences. The weekend is packed with new experiences from the moment you walk through the door, where your friendly BBS UK team will be there to welcome you. The Hilton Hotel, Northampton, centrally located for rail, road and air travel, has been home to our conferences for over 20 years. Many of the staff have been with us from the beginning and they pride themselves on their service and exceptional attention to detail throughout the weekend.

Guests usually arrive on the Friday afternoon/evening and enjoy the opportunity to relax in an inclusive and welcoming environment. The hotel has a lovely pool, spa and gym area, a restaurant and bar, and a large bright and airy lobby/lounge, which is served by a coffee bar.

The main conference programme takes place on the Saturday and, once again, there will be an excellent mix of speakers and facilitators including Professor Beales, who will give an overview of research and development in the world of BBS. Throughout the weekend, there will be the usual opportunity for private and workshop discussions with experts in the research and management of BBS, as

well as with members of the various BBS Clinics Teams and BBS UK. For our younger members, there will be a fun-filled day trip to look forward to (accompanied by a parent/family member) and, for the little ones, a pop-up crèche.

We are extending our programme into Sunday morning for the first time this year, and will round off the weekend with a sandwich platter lunch at 1pm, before we say our farewells.

BBS UK are delighted to be able to offer this event free of charge to those who have BBS and their immediate family/carers; guests will just need to book and pay for their accommodation directly with the hotel. In addition, we are providing a limited number of 'kids go free' places (for those who have BBS and their siblings, 17yrs and under) at the Hilton Hotel, Northampton and have secured a competitive rate on a dinner, bed and breakfast basis.

Booking is in two parts:

1. Book your place at the **BBS UK Conference**: go to www.bbsuk.org.uk (News & Events / BBS UK Conference)
2. Book your **accommodation** directly with the hotel, see enclosed information.

If you have any queries, please contact Léonie Hudson

Tel: 07510 384242

E: leonie.hudson@bbsuk.org.uk

We look forward to greeting friends, old and new, in 2022.

Chris Humphreys Memorial Fund

Chris Humphreys was one of the founder members of BBS UK; she organised the conference and supported BBS families for many years. Sadly, Chris passed away a few years ago; the Chris Humphreys Memorial Fund has been created in her memory, in order to support people to attend the BBS UK weekend family conference.

Why do we have the Fund?

At BBS UK, we believe that the conference provides an invaluable opportunity for members to meet and connect with other people who have the syndrome, as well as having the opportunity to learn more about scientific developments and about living well with BBS. The Fund is there to support people who might otherwise find it difficult to attend.

Who is eligible?

Those who are attending for the first-time and who meet the following criteria:

- Currently living in England, Scotland, Wales, Northern Ireland and Republic of Ireland
- Adult diagnosed with Bardet-Biedl syndrome or
- Parent/carers of a child (0-17yrs) diagnosed with Bardet-Biedl syndrome (child must be living with applicant)
- Eligible for any of the following benefits:
 - Universal Credit
 - Child Tax Credit
 - Working Tax Credit
 - Income Based Job Seekers Allowance
 - Income Related Employment Support Allowance
 - Income Support
 - Housing Benefit
 - Pension Credit

What will the fund cover?

The fund will cover the cost of up to two parents/carers (children go free) or one adult who has BBS and a personal assistant/carer, to attend the conference weekend. This will include dinner, bed and breakfast on the Friday and Saturday nights.

How can I apply for support from the CHMF?

To apply for support, contact Léonie Hudson via email. A copy of a recent benefits letter,

no more than 6 months old, will be required to support the application. If the letter does not confirm the applicant's address, a utility bill will also be required.

What happens if I am offered support from the Fund?

If your application is successful, we will take your conference booking information and make the required arrangements on your behalf.

Additional Information

The fund is limited and will be allocated on a first come first served basis. We are able to receive applications to the Fund for as long as the Fund contains sufficient monies to be able to offer support.

For further information about the Chris Humphreys Memorial Fund, please contact: leonie.hudson@bbsuk.org.uk

FAQs

Am I guaranteed to receive support from the Fund?

No, the funding pot is limited and will be allocated on a first come, first served basis

Does the Fund cover transport costs?

No, however BBS UK may be able to offer some support in exceptional circumstances

How many people from my family/friendship group will the Fund cover to attend conference?

The fund will pay for two parents/carers to attend – *all child(ren) diagnosed with BBS and their sibling(s) go free.

The fund will pay for an adult diagnosed with BBS to attend, along with their personal assistant/carer.

Can I apply to the Fund if I have previously attended a BBS UK Conference?

No, the Fund is only available to first time attendees.

Where else can I go for support?

The Family Fund will provide funding for families with children 17 years and under, in receipt of means tested benefits; for more information go to www.familyfund.org.uk

Local authority funding may also be available, contact your social worker or local authority for support.



Regional Group Ireland

A key aim for BBS UK over the next three years is to reduce isolation and improve communication and connection, with and amongst, the BBS community. One of the ways we will tackle this is with the development of regional groups; we are excited to report that the first BBS UK Regional Group is underway in Ireland.

BBS UK has brought together a group of individuals who have BBS, and parents of children with BBS, from across the Republic of Ireland and Northern Ireland to discuss what they would like from a regional group; key aims are to raise awareness and provide connection and support for people in Ireland who are affected by BBS, in particular to support those who are newly diagnosed.

Sandy, mum to Charlie, 3, who has BBS, is a founding member of the regional group and raised awareness with articles placed in her local newspapers. These have already led to new contacts being made, with families and professionals contacting BBS UK for more information. Once the group is established, with enough contacts, the aim is to hold a BBS Information Day in Ireland.

Sandy and Charlie share their story below:

Charlie was diagnosed with Bardet-Biedl Syndrome in December 2020.

When Charlie was born, doctors had not shown any concerns about his health but



mum, Sandy, quickly spotted issues with his development: he wasn't sitting up, rolling over, babbling or attempting to crawl in line with other children his age. Sandy's concerns were brushed off, and she was told that Charlie was too young and that all children learn at their own pace.

At around 15 months, the family had their first appointment with a paediatrician and Sandy was able to share her concerns; Charlie was diagnosed with Global Developmental Delay and routine genetic tests were ordered. Unfortunately this coincided with Covid-19 which halted routine testing. The family were told that they would have to wait three years to have genetic tests because of the backlog caused by the pandemic. Charlie's development was hampered further because physiotherapy stopped and his speech and language referral was halted.

Sandy's concerns continued to grow and she

found a private genetic consultant in the Republic of Ireland, at the Crumlin Hospital in Dublin. The consultant had previously worked at Great Ormond Street in London, and was certain that he knew what condition Charlie had. He wanted to confirm his diagnosis first, however, and it was a further six weeks before the family were told that Charlie had Bardet-Biedl syndrome.



there is significant deterioration, in terms of vision or kidney function, that tests reveal the BBS diagnosis.

Raising awareness at a local level amongst professionals and those working with young people is very important to Sandy; holding a BBS Information Day will be a key aim for the Regional Group for the coming year.

Sandy remembers asking the consultant what this meant and he replied, "It means he is going to go blind."

Sandy is an optician and so this came as an enormous shock – seeing children every day with eye conditions, it wasn't something she had been concerned about for Charlie. Sandy had anticipated the possibility that Charlie may not walk or talk, and worked through possible solutions for each but she had not anticipated loss of vision.

Charlie has routine check-ups every six months for various aspects of BBS and the family have to travel to Great Ormond Street in London for these; in the last six months, they have travelled to London five times because there are no specialists in Northern Ireland.

Sandy is working with BBS UK to develop a regional group and Information Day in Ireland. She said, "There needs to be more awareness for medical professionals here, so that they can help someone like Charlie."

Sandy learned from a consultant in Dublin that late diagnosis is common in BBS in Ireland. A young person may have struggled for some time with sight loss, autism and poor communication skills; however it is only when



If you would like more information about BBS UK Regional Group (Ireland), contact leonie.hudson@bbsuk.org.uk

BBS Multi-Disciplinary Clinics

As we emerge from the Covid-19 pandemic and start to see a reduction in the impact and severity of the disease, thanks to the incredible vaccination efforts of the NHS, the longer term impact of the past two years is now being realised. Regular news reports alert us to the impact on mental and physical health, as well as to the ever-increasing waiting times for elective care. It is now more important than ever that we each do what we can to look after our own health and wellbeing.

This newsletter contains lots of ideas of ways in which we can be more active, to have a positive impact not only on our physical health, but also upon our mental wellbeing. At last year's conference, Dr McGibbon and Dr Walker spoke about social isolation and ways of managing anxiety and low mood; the recording for this can still be found on the BBS UK website by going to www.bbsuk.org.uk/news-events/bbs-uk-conference.

The pandemic has inevitably impacted on our BBS clinics service, in particular our service at Queen Elizabeth Hospital, Birmingham, which saw large scale cancellation of outpatient clinics as teams and wards were reassigned to tackle Covid-19. This means that many BBS patients will be offered a second telemedicine appointment instead of an in-person appointment, as the team work through the backlog.

We understand that this may cause frustration and concern for some, however, be reassured that patient notes are reviewed by the medical

teams, and appointments are allocated according to need.

Your patience and understanding is really appreciated by the team, as is ensuring attendance when called. Appointments are in huge demand and missed attendance at in-person and telemedicine appointments costs the NHS a considerable sum of money, and also extends waiting times significantly. Sometimes late cancellations are unavoidable, however please do try to let your Patient Liaison Officer know as soon as possible if you are unable to attend your appointment, so that it can be offered to someone else.

It really has been a challenging two years and the impact is going to be felt for some time. We have been fortunate that, throughout the pandemic, the support we have received from our NHS-funded specialised service has been exceptional. Our heartfelt thanks go to the BBS teams at all four centres for their continued commitment to our patient group. I would also like to say a huge thank you to the BBS UK staff team, who have continued to work tirelessly in support of the service and our community throughout lockdowns, isolation and home-schooling; we are blessed to have such a dedicated and committed team. If you would like to speak to someone about the clinics service or any other aspect of BBS UK's work, contact details can be found at the back of this newsletter.

Tonia Hymers
Service Manager



2021 Membership Audit:

Results

Last year, we conducted a membership audit. We have listened to what you told us and we are reflecting on your insights as we develop and review our services over the coming year.

Who completed the audit?

- 50% of people are the parent of someone diagnosed with BBS
- 37% completed it as a person themselves diagnosed with BBS
- 13% completed it on behalf of an adult diagnosed with BBS

In terms of the views that were represented:

- 80% have a genetic diagnosis and 20% have a clinical diagnosis
- 38% are under 18
- 38% are aged 18-34
- 21% are aged 35-54
- 3% are aged 55+
- 58% are registered as Severely Sight Impaired
- 19% are registered as Sight Impaired

In terms of how long ago the person was diagnosed with BBS:

- 4% were within the last 12 months
- 23%: between 1 and 5 years ago
- 27%: between 5 and 10 years ago
- 46% were diagnosed over 10 years ago

Respondents experience the following symptoms either moderately or severely:

- Obesity: 70%
- Learning difficulties: 43%
- Developmental delay: 39%
- Anxiety: 37%
- Low mood/depression: 36%
- Extreme tiredness/fatigue: 34%
- Speech and language difficulties: 32%
- Autistic spectrum disorder: 28%
- Issues with digestive system: 24%
- Urinary tract issues: 23%
- Kidney abnormalities: 16%
- Diabetes: 9%

Participants are in receipt of the following benefits:

- Personal Independence Payment: 53%
- Disability Living Allowance: 44%
- Employment Support Allowance: 22%
- Direct Payments: 18%
- Carers Allowance: 16%

Respondents outlined the following emotional and psychological impacts of having BBS:

- Anxiety: 78%
- Loss of confidence: 65%
- Stress: 63%

70%

BBS impacts moderately or severely on day to day routines

75%

BBS has a moderate to severe impact on mobility and getting around

81%

BBS impacts on relationships with family and friends

68%

describe the impact of BBS on education as moderate or severe

- Fear: 52%
- Anger: 43%

Of the following services, respondents outlined the ones they had NOT accessed but would like to:

- Counselling: 43%
- Genetic counselling: 34%
- Support with securing a volunteering placement: 31%
- Physiotherapy: 26%
- Support with obtaining employment: 24%

Respondents have engaged with BBS UK in the following ways:

- 75% have accessed information and/or support
- 73% are members
- 60% attend events
- 35% donate or raise funds

People agreed with the following statements:

- BBS UK works hard to improve the lives of those with BBS: 80%
- BBS UK is focussed on the wishes of its members and their everyday needs: 76%
- BBS UK is ambitious on behalf of people affected by the syndrome: 73%
- I trust BBS UK to do what it says it will do: 72%
- People with BBS have a say in what BBS UK does: 58%

Respondents felt that BBS UK had made the following differences to them:

- I understand the condition more fully: 80%
- I am better informed about research into the condition: 78%
- I have greater awareness of the support available: 59%
- I am more confident in managing the challenges of my / my child's condition including my/ my child's sight loss: 48%
- I am more able to lead a fulfilling life: 26%

People rated the following BBS UK services as good or excellent:

- Newsletters: 100%
- Website: 100%
- Annual conference: 88%
- Information booklets: 100%
- Support with fundraising: 92%
- Information for children and young people: 100%
- General contact and support: 95%

People have said that the following would be of interest to them:

- Updates on treatment and research: 87%
- Local/regional groups: 72%
- Social opportunities and activities: 69%
- Information on assistive technologies: 66%
- Managing everyday life with BBS: 66%
- Support with exercise and with maintaining a healthy diet and lifestyle: 50%

Other outcomes from the audit:

- 42% of respondents said that BBS affects their/their child's life mildly to moderately; 57% stated that the impact was severe to very severe.
- 14% of respondents are employed full or part-time; 10% undertake voluntary work

43%
understand BBS and how to manage it; they feel that support is in place to help them live their life or take care of their child's needs

82%
are happy or very happy with the ongoing care they receive from the BBS Clinics Service

Fundraising Round-Up

Nails by appointment!



On Saturday 6th November, Ashley Hobbs of Market Harborough opened her beauty salon up to raise funds for BBS UK from all of her booked appointments. From 9am until 9pm, Ashley gave her customers gel

manicures, asking for a £5 minimum donation in return. On top of this, Ashley and her two sons, Ollie and Charlie, organised a huge raffle of beauty treatments and products, flowers and drinks. Charlie drew the raffle numbers and, altogether, the family team raised over £350 for BBS UK.

What a great gesture...maybe your hair or beauty salon could do something similar?

Running for real!

London Marathon Events have confirmed that The Vitality Big Half will take place on Sunday 4 September 2022. The 13.1 mile running event starts by Tower Bridge and finishes alongside the Cutty Sark in Greenwich, taking in many iconic London sights long the way. BBS UK is delighted to have 4 places available for people who are aged 17 or over and wish to raise as much sponsorship as they can to support adults and children with Bardet-Biedl Syndrome. Please email Christine.saxon@bbsuk.org.uk with your contact details if you wish to take part.

Tide Lines

Tide Lines are a four-piece band from the Scottish Highlands whose members Robert Robertson, Ross Wilson, Alasdair Turner and Fergus Munro have been supporting BBS UK since May 2021. As lockdown eased in Scotland and gigs started again, their fan group, led by Donald and Carolyn Young and Linda Clark, sold Tide Lines fans t-shirts, with all profits going to BBS UK. A huge response from their fans has resulted in a presentation of a fantastic £1,055, which the Wilkie family received on behalf of BBS UK.



“The response was quite exceptional” said Donald Young, “we sold around 120 t-shirts as far afield as Europe, the USA and Australia. Due to covid restrictions, people were still uncomfortable at gigs so they were selling or auctioning their tickets and donating the proceeds to us, in some cases at four times the face value. It really was quite extraordinary.”



The band formed in 2016 and write most of their original material in English but always with a nod to their Gaelic heritage. Their sound has been described as “...Folk with rousing rock, sparkling electronics and escapist lyrics....awash with arms-aloft sing-alongs.” (The Sunday Times) and “...roaring tunes embracing anthemic lift-off and wide-eyed wonderment.” (The Daily Mirror)

The band’s latest album, Eye of the Storm, finished its first week at number 12 in the UK Album Charts in May 2020: topping both the Scottish and Independent album charts in the process. In 2019, they were named “the Rising Sound of Scotland” at the Nordoff Robbins Scottish Music Awards - one month after selling out the 1900 capacity Barrowland Ballroom in just 5 minutes.

Thank you to the band, to their fans and to Donald, Carolyn and Linda for your brilliant support.

This is me by Aneeba Ahmed



Hello, I am Aneeba and I am 30 years old. I live with my family on the north side of Birmingham. I am a BBS patient, fundraiser and volunteer. I was born with BBS and my family knew because of the extra digits I had, which I had removed. I have BBS 10 and on the 9th February 2022, I decided to do another haircut challenge to give something back to BBS UK. My sister, Hena, took me to Billy’s where I met the lovely hairdresser. I was nervous but I was happy to do it. I raised £70 for the charity and my hair was donated to The Little Princess Trust, who provide free real hair wigs to children and young people who have lost their own hair. I wanted to help that charity too because I have friends who have recovered from cancer.

Congratulations and thank you!

From January to December, the following people created a Facebook Fundraiser to raise £1,600 for BBS UK:

Steve’s 50th, Stacey, Aneeba, Margaret, Ashley, Chloe, Nicola, Rebecca, Lynette and Lindsay. Thank you all for your fabulous fundraising efforts!



Creating a Facebook Fundraiser is a quick and easy way of raising funds for us. Please talk to Christine on 07753 163045, or email fundraising @bbsuk.org.uk if you would like help with doing this. We are not contacted by Facebook when you create a Facebook Fundraiser, so please do get in touch to let us know that you are holding one.

Stunning Sunflower Notelets

Judged in September 2021, the Best Bloomin' Sunflower competition was a huge success and many BBS UK members welcomed the glowing golden flowers into their gardens, allotments, and plant pots! Some were lemon, some orange and others a wonderful deep yellow. All were tall, some taller than others and

the winner, Aneeba Ahmed, had sunflowers which reached the top storey of her house. We have produced bright and cheery notelets (blank inside for your own use), for sale at £7.50 per pack of 10 (inc. p&p) which are now available to buy. Details of how to order and pay can be found in the fundraising section of the BBS UK website: www.bbsuk.org.uk

Diet and Exercise

Tips to help you eat better and move more

Did you know more than half of us plan to do more exercise and just under half of us want to lose weight, however only a quarter of us manage to keep to our New Year resolutions? It can be difficult to stick to a new activity and food routine, but these steps to success might help you:

1. Start by thinking about your reasons for wanting to make a change and imagine how you will feel when you have achieved your goal.
2. Make small changes and celebrate each achievement.
3. Give yourself enough time to reach your goal.
4. Make sure you have the equipment or food that you need before you start.
5. Build exercise and diet changes into your usual routine.
6. It can take between 3 weeks and 3 months to settle into new habits, so be kind to yourself if you find it hard in the beginning.
7. Find a friend, family member or a group of people to support you along the way.

Are you ready to increase your activity levels?

- **Build up gradually:** Slowly increase the amount of activity you are already doing and try to limit the amount of time you spend sitting or lying down.



- **Simple solutions:** Choose the stairs rather than the lift, walk to the next bus stop or dance around the kitchen whilst you are cooking! Walking is a great way to start boosting your activity.
- **Enjoyment:** Find something you like to do and aim to do it more!
- **Track your progress:** Keep track of your activity using an activity tracker or mobile phone. 'Active 10' is a tracker App that uses your phone to monitor your walking. It tracks your steps, helps with goals, and shows your achievements. Seeing how far you have come can boost your confidence and encourage you to keep going. 'Couch

to 5K' is a great App if you want to start running. (Further information about running for visually impaired people can be found on the British Blind Sport website: <https://britishblindsport.org.uk/membership/bbs-sports/guide-running/>)

- **Celebrate your successes:** Share your achievements with a friend, your family, BBS UK, or your clinic team.
- **A boost for your wellbeing:** Think about how you feel after you have been more active. Exercise brings many health benefits, but it can also make us feel better, improve our sleep and our energy levels.

Do you want to change your diet?

- **Keep a food diary:** It can be helpful to record all the food and drinks you have had over a couple of days and how you felt at the time, for example, tired, fed up or bored. You may notice there are times when you eat for reasons other than hunger, for example because you are super tired.
- **Planning:** Creating a weekly menu can make shopping easier and help you make better food choices.
- **Food shopping:** Shop after you have eaten rather than when you are feeling hungry. When we shop on an empty stomach, extra treats may end up in our basket.
- **Plate sizes:** if you have different size plates at home, using a smaller plate can help you to eat less.
- **Routine:** Aim to have 3 main meals at regular times and watch your snack choices. Chat to your dietitian about the meal and snack pattern that would be best for you or your child.
- **Eat more veg:** Try filling half of your plate with vegetables and salads. Divide the other half with protein rich foods, including lean meat, fish, Quorn, or pulses and starchy carbohydrate foods like potatoes, pasta, rice, chapatti, yam, or bread.
- **Enjoy your meals:** Eat without distractions

so you can focus on how much you are eating and enjoying the food. Studies have shown that you may eat more if you eat whilst watching the TV or gaming, for example.

- **Drinks:** Swap drinks that are high in calories or sugar for a low-calorie drink or water. If you like fizzy drinks, try the diet/zero sugar alternatives.
- **Alcohol** is high in calories and too much of it can also lead us to making less healthy food choices. Alternating alcoholic and low-calorie soft drinks is one way to tackle this.
- **Ask for help:** Ask for support from your family or friends, maybe they would like to make diet changes so you can do this together? Ask family or your partner not to buy you food gifts for special occasions.

If you would like to make a change but are unsure how to get started, chat to your clinic team at your next appointment.

Here's wishing you a happy and healthy 2022.

Sarah Flack

Principal Dietitian, GOSH



A Personal Perspective: Katie Oates

My Fitness Journey

Hello everyone! My name is Katie Oates and I am 23 years old. I was diagnosed with BBS10 when I was 5. I have struggled with many of the common symptoms of BBS, mainly my vision (I am registered Severely Visually Impaired and I use a long cane). In 2015, I underwent a successful kidney transplant and it has been quite a journey but now my life is so much different for the better. In the past I have also really struggled with my weight and fitness but all this has changed now as I decided in 2019 to go on a weight loss program. I use the Nutra Check App to track all of my calories, in and out.

During the first lockdown, I was watching my mum and my brother doing fitness classes online. I had never been interested up to that point, but I really thought I could give it a go. Mum was really worried about me doing it as my joints are very bendy and she was worried that I was going to hurt my ankles or knees. But I insisted and so I started joining in the following week, doing between 4 and 6 online classes a week. I found it really hard at first but I soon started to feel the benefits and began to feel fitter and happier. The classes were about doing exercise using just your own body weight, meaning that it was really easy to join in because you did not need any equipment to



do it. The instructor, Stu, could give me alternative exercises to do, if I could not do the exercise that the class was doing, which was really good.



When we came out of lockdown 1, I was so enjoying the exercise that I employed a Personal Trainer, who comes to the house once a week and does a one-to-one session with me. I love it! I have been going from strength to strength in my personal training. Stu sets me goals around strength, fitness, weight and body composition, which I work towards and, when I hit them, we set new ones. This keeps me really focused.

I also started a dance class online on a Monday night which my mum's friend, Jude, takes. It is for adults with learning difficulties. It is great fun and I've met a whole new circle of friends. On top of this I take our dog, Rosie, for long walks around the village. I can manage ok in the village with my eyesight and I like to get 4 miles a day in, when possible.

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

My weight has now dropped to 79.8 kgs, so in total I've lost 25.2kgs in about 3 years.

To summarise I am in a mentally and physically better place than I ever thought I could be. Certainly before my transplant, I was so tired that sometimes I couldn't face school or even leave the house. I feel very lucky for the gift of my kidney and I'm determined to make the most of every day. I'm loving my fitness journey and all I can say is that if I can do it, you can do it too. It's hard work with BBS but it's a great feeling when you start to see and feel the benefits of being fitter and stronger.

Thanks for reading this.

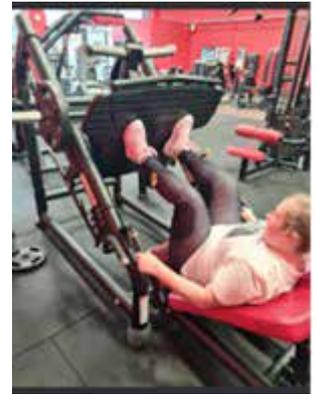
Katie. X

Slimming World

One million step challenge success!

Getting on board with increasing exercise can always be a challenge, especially hitting the recommended 10,000 daily steps target. For myself, I've always just put the emphasis on moving more and building up my steps gradually, challenging myself to do a little more each day. It's amazing how quickly you start to feel fitter - and moving more soon becomes a lot easier. Before you know it, you're smashing 10,000 daily steps!

Over the Christmas period, BBS UK and the



attendees of our BBS UK slimming zoom group, set ourselves the challenge of reaching 1 million steps between us. By working together to support and motivate each other, we not only hit the target but went on to exceed it, completing 1.2 million steps in total - what an achievement!

The people who took part in our Slimming World Zoom sessions had the following to say:

"Slimming world isn't like a diet, you can eat as much as you like with the variety of free foods."

"Slimming world is inclusive for the visually impaired – resources are available in all formats."

"Everyone attending Slimming World is all in the same boat – I have made friends, achieved goals and am fully supported by all."

"I have learnt to make healthier and better choices when meal planning and shopping."



Keira & Jacqui

"I never feel hungry."

"Don't be afraid to ask for support – the consultants are very supportive."

We are working in partnership with 'Slimming World on Referral' to offer a free 12 week membership to people aged over 16 who have BBS and would like to lose weight.

Slimming World is a plan that supports people to lose weight in a healthy, sustainable way.

If you would like to refer yourself, please call 01773 302475. This number is for our members only; the Slimming World team will talk you through the scheme and connect you with your local Slimming World representative.

Amy Clapp

Patient Liaison Officer, BBS UK Clinics

Slimming World Easter Treats – Mini Chocolate Egg Pots

Ingredients

- 500g fat-free natural fromage frais
- 11g sachet Options Belgian Chocolate instant hot chocolate
- 1 level tsp cocoa powder
- 11g sachet Options White Chocolate instant hot chocolate
- ½ tsp vanilla extract
- 4 Cadbury Mini Eggs

Method

1. Divide the fromage frais between 2 bowls. Add the Options Belgian Chocolate and cocoa powder to one of the bowls and mix well. Put the Options White Chocolate and vanilla extract in the second bowl and mix well.
2. Divide the white chocolate mixture between 4 dessert glasses, then divide the Belgian chocolate mixture over the top. Chill until ready to serve, then add a mini egg to each pot and take your pick!



Sensing Nature

Spending time in green, outdoor spaces, or bringing nature into your everyday life can benefit both your mental and physical wellbeing, according to national mental health charity, Mind. For example, doing things like growing food or flowers, exercising outdoors or being around animals can have lots of positive effects, including:

- Improving your mood, reducing feelings of stress or anger
- Helping you to take time out and feel more relaxed
- Improving your physical health
- Improving your confidence and self-esteem
- Connecting you to your local community, helping you to meet new people and reducing loneliness

For more information go to www.mind.org.uk

A study called 'Understanding Experiences of Blind Individuals in Outdoor Nature' led by

Maryam Bandukta, a PhD student at UCL, found that: 'In the context of outdoor nature, participants [with a visual impairment] described feeling independent and free when in open natural space, which inspired them to explore and get immersed in the environment without fear of traffic or colliding with other people.' The sensory experiences were found to be relaxing, as well as serving as navigation clues; for example, the tactile and auditory changes in the walking surface, the sounds of flowing water, and the feeling of the breeze and sunshine on the face.

There were also social benefits for those accompanied by sighted guides, whether through conversation about unrelated topics or about the sights and sounds around them. Having the environment described was reported as being important for 'developing mental images and making the space and experience memorable.'

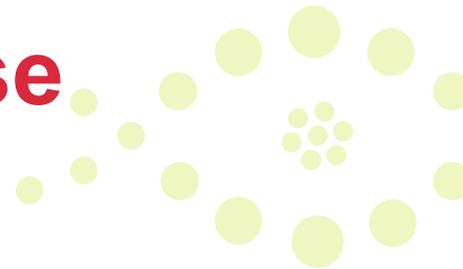
'Easing into nature with sight impairment', a booklet produced by the University of Exeter, provides a fascinating review of the results of a two-year research project that aimed to answer the question: how do people living with sight impairment experience nature during their lives? The findings have 'important implications for how we understand, enable and promote more positive, multisensory nature experiences' and highlight the importance to wellbeing of experiencing nature, for those who are visually impaired. It also suggests how any difficulties may be addressed, to promote more inclusive experiences, whilst at the same time rejecting stereotypical assumptions that may be made based on disability.

The project focused on understanding people as individuals, with diverse nature interests and knowledge. Conducted by Dr Sarah Bell at

the University of Exeter, the study was funded by the Economic and Social Research Council and ran from November 2016 to November 2018. The booklet can be accessed via www.sensing-nature.com, which has audio built in, enabling us to listen to an overview of the project, the study's interviews and also the conclusions that were drawn.

Being outside and immersed in nature is good for us, not just physically but mentally as well. As the biologist, writer and ecologist Rachel Carson wrote in her book, *Silent Spring*: "Those who contemplate the beauty of the earth find reserves of strength that will endure as long as life lasts. There is something infinitely healing in the repeated refrains of nature - the assurance that dawn comes after night, and spring after winter."

The Benefits of Exercise for Our Brains



At our 2018 Conference, Dr Elizabeth Forsythe - who is an MRC Clinical Research Fellow at UCL, Great Ormond Street Institute of Child Health - gave a powerful talk which is definitely worth revisiting.

Dr Forsythe spoke about what she had learned in the previous couple of years about how we can optimise our brain function. She explained that we have two hippocampi, one on each side of the brain and that they are important for memory, learning and for regulating our mood. Dr Forsythe went on to say that we know that the hippocampus is slightly smaller and a little bit unusually shaped in many people with BBS which, she said, makes sense because we've heard from BBS patients that they may be struggling with school, to learn new skills or have issues with mood.

We used to think that the brain is what it is, you've got the brain you've got and there's nothing really you can do about it but the good news is that it is possible for all of us

to positively impact our brains. The average brain has around 100,000 billion neurons and, in the hippocampus alone, we make 700 new neurons every day. Eating more healthy food (such as fruit and vegetables), reducing our alcohol intake, learning new skills, exercising more, reducing stress and improving sleep quality are all things that will improve our turnover of cells. This is important for everyone, not just those who have BBS, because as we age, the turnover of cells reduces.

In 2016, Dr Forsythe felt inspired to look at how we could improve the size and the function of the hippocampus, particularly in people with BBS. She and her team invited a group of children with BBS into Great Ormond Street Hospital for an MRI scan of the brain and for an assessment to check their memory and fitness. The children were then divided into two groups and they went home and followed a programme: half carried on with their normal routine and the other half worked really, really hard with a personal trainer.

The fantastic thing was that when they re-tested the children, they found big changes in the children who were fitter: they had better blood flow to their brains, they had bigger hippocampi and they were getting better at completing the memory game. What was particularly exciting about this was that, although the team already knew that you can improve the size of your hippocampus (and therefore improve your memory) through exercise, they didn't know that if you were born with a small hippocampus, you could still make a positive difference.

Another positive side effect that emerged from this trial was that lots of the participants said:

'I feel more physically confident, I feel more confident to take on sport, I feel more confident to go into a gym.' In particular this was true for the people who had embraced it as a whole family.

Dr Forsythe said, "The past two years have given me a real insight into the benefits of exercise and I think all of us recognise that it's really, really hard to make a significant lifestyle change. Anybody who has ever been on a diet knows how difficult that is, but it is possible."

The full audio recording of Dr Forsythe's talk can be downloaded from the BBS UK YouTube Channel – it is definitely worth a listen.

Couch to 5K

Taking up running can seem like a scary prospect, especially if you feel out of shape or unfit. NHS Couch to 5K will help you gradually work up towards running 5km in just 9 weeks.



Ross Gallagher taking part in the Tralee parkrun

What is Couch to 5K?

Couch to 5K is a running plan for absolute beginners. It was developed by a new runner, Josh Clark, who wanted to help his 50-something mum get off the couch and start running, too.

How does Couch to 5K work?

Probably the biggest challenge a new runner faces is not knowing how or where to start. Couch to 5K works because it starts with a mix of running and walking to gradually build up your fitness and stamina. Week 1, for example, involves running for just a minute at a time, creating realistic expectations and making the challenge feel achievable right from the start.

Who is Couch to 5K for?

Couch to 5K is for everyone. Whether you've

never run before or you just want to get more active, Couch to 5K is a free and easy way of getting fitter and healthier. If you have any health concerns about beginning an exercise regime like Couch to 5K, make an appointment to see a GP and discuss it with them first.

What are the benefits?

- Running improves your physical health.
- Running requires little equipment, but a good pair of running shoes that suit your foot type may help improve comfort.
- Running regularly will improve the health of your heart and lungs. It can also help you

lose weight, especially if combined with a healthy diet.

- There's evidence that running may help increase bone density in some people, which can help protect against bone diseases like osteoporosis.
- Running has mental health benefits. Taking on the challenge of Couch to 5K can help boost your confidence and self-belief, as you prove to yourself that you can set yourself a target and achieve a goal. Running regularly can also be a great stress reliever and has even been shown to combat depression.

One You Couch to 5K app

The One You Couch to 5K app gives you a choice of coaches to listen to as you run and also helps you to track your progress. You can be coached by celebrities including Jo Whiley, Sarah Millican, Sanjeev Kohli and Michael Johnson.

All of the information you need to get started can be found at: <https://www.nhs.uk/live-well/exercise/get-running-with-couch-to-5k/>

Source: www.nhs.uk

Useful Resources

VICTA

Visually Impaired Children Taking Action (VICTA) supports children and young people who are blind or partially sighted and their families across the UK. If you are visually impaired and under the age of 29, or are the parent of a visually impaired child or young person then VICTA can help. VICTA also have an extensive events and activities programme: www.victa.org.uk

Metro Blind Sport

A London-based charity; their aim is to open doors to sport for all vision impaired people, regardless of age or sporting ability. They have sessions throughout the year for archery, athletics, bowls, cricket, cycling, football, goalball, rambling, sailing, skiing, swimming and tennis: www.metroblindsport.org

British Blind Sport

Enabling blind and partially sighted people to have the same opportunities as sighted people to access and enjoy sport and recreational activities in the UK: www.britishblindsport.org.uk

Accessible Gyms

<https://www.activityalliance.org.uk/get-active/inclusive-gyms>

Mencap Sport

<https://www.mencap.org.uk/about-us/what-we-do/mencap-sport>

Paralympic Sport

<https://parasport.org.uk/>

Special Olympics GB

<https://www.specialolympicsgb.org.uk/>

UK Sports Association for People with Learning Disability

<http://www.uksportsassociation.org/>

Disability Sports Northern Ireland

<https://www.dsni.co.uk>

Disability Sport Wales

<https://www.disabilitysportwales.com/en-gb#cookieConsent>

Scottish Disability Sport

<https://www.scottishdisabilitysport.com/>

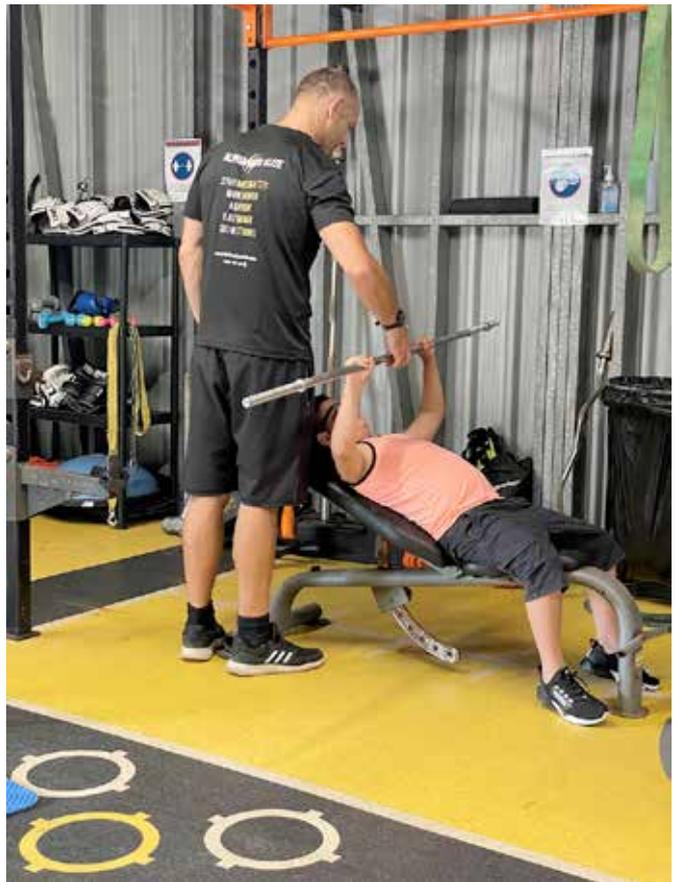
This NHS website <https://www.nhs.uk/live-well/exercise/get-active-with-a-disability/> has a long list of different sports that are available to people with disabilities, ranging from archery, rowing, cricket, karate and football; to goalball, golf, fencing, sledge hockey and snowsports.

A Personal Perspective: Eesa



Eesa, aged 9, has been training alongside his brother, Ismail, at their local outdoor sports centre, with the support of a personal trainer called Marek. When he grows up, Eesa wants to be just like his hero, The Rock (Dwayne Johnson.) The Rock has inspired Eesa to eat healthily, including lots of fruit and salads, and to drink plenty of water. Eesa allows himself one day off each week, when he enjoys a takeaway but no fizzy drinks. Eesa's favourite activity is tyre-flipping because it makes him feel strong afterwards.

Eesa loves competing with his brother, who is 22 years old. Eesa is very motivated to continue with his training and is particularly looking forward to training outside again, when the weather improves.



When he is older, Eesa would like to buy his own gym; he is a real advocate of the physical and mental benefits of exercise and would like to encourage other young people to have a go at it because he believes that they will experience positive results from doing so.



My Love Affair Starts With the Orbit

Emma May

I really enjoyed the conference in 2021. I had not been to one for a while (I always listen to the conference report) and being able to access it on Zoom was fantastic. I enjoyed all of the presentations, but I was very upset that the lovely lady from the RNIB did not mention the Orbit. Yes I do have four smart speakers and they have changed my life, however, I love my Orbit too and when I mentioned this after the conference, Tonia (BBS UK) asked me to write an article, so here goes.

Orbit is a refreshable braille display that can either be linked to a computer or used as a stand-alone with an SD card. The literature does say that you can read Kindles on it, but I haven't managed that yet; I think I need another tablet, which is something for the future. One of the fantastic benefits of the Orbit

is how convenient and portable it is – it can be quickly slipped into a pocket or a handbag, meaning that you can easily use it outdoors.

I had first seen a refreshable display device back in 1990, but they had always been way out of my league. However the price of the Orbit made me very excited as it isn't too high and I started saving. In the spring of 2020 I found out that the RNIB was giving away Orbits and I got my hands on one. After a little while, I did manage to break the charging point, but I loved it so much that I decided to buy another one. This device is definitely a game changer for fast braille users. I wondered how they had managed to cut the price so much and I was told that they took the technology that mobile phones use to vibrate, in order to make the pins go up and down.

The Orbit is small, portable, not too loud, and you can make the refreshing cells change at your own speed by using the up and down button. The battery life is quite long too. I get the books in two ways. The RNIB sent me an SD card containing two thousand books and I also download books from the RNIB readers services website to my Mac and then copy and paste them on to the card through a card reader. Having enjoyed braille for over thirty years, to have so many books to dip in and out of is a game-



Emma May with her great nephew

changer for me. The technology is so good that if you close a book, it remembers the place and you can return to the same place at a later date.

I also want to talk about my experience with the Mac computer. I bought it three years ago after a lot of soul searching. I found it very hard to get training with Jaws (screen reader software) so found the thought of re-learning a new speech system extremely daunting. My brother found MacForTheBlind in the States and, after emailing them, I finally decided to buy a Mac. After the wonderful manuals I managed to buy from the MacForTheBlind website, I've not looked back; I surf the net, email people all over the world on Google groups, listen to Apple books, and use Zoom if someone sends me the link. I used to be able to Face Time but, since the last upgrade, I have found this more difficult but I'm sure I will get the hang of doing it again. I have found that some Apps do not work on the Mac so, if I was doing it now, I would buy an iPad with an external keyboard instead.

Getting out and about

I have been lucky to have Direct Payments for five years. This is to pay for more exercise. First of all I visited the local gym once a week, but after I finished shielding I didn't want to do that. So I now go with my PA for walks in the countryside. We usually do about two to three miles a week and she is great at making sure I get the full experience, explaining the scenery and showing me trees and other things. One week, she found a large piece of twisted ivy that we carried a mile and a half back to the car for my mum to make an art installation in the garden.

BBS UK told us all about Direct Payments and encouraged us to go for it and it has been wonderful for me. Sometimes the paperwork is a bit complicated but a charity deals with all of that for us. Sometimes we go for coffee and 'naughties' but not too often. My PA also helps me to buy birthday cards and things for my mum, so that my mum doesn't have to stand outside the shop and wait for me, like she did before.

Being able to walk so far is largely down to

some amazing shoes I found online. The company is DB Wider Fit shoes and they have shoes for every occasion. I got the shoes for my niece's wedding and they were so lovely and comfortable that I smiled to myself as we walked down the line of ladies who were all complaining about their high heels killing their feet and there I was, the one with funny feet, not hurting at all!

I also have a cane that helps with rougher ground. It isn't too good at picking up small steps so my guide needs to be aware, but my mega PA is getting as good as my mum at this. I guess the ball on the end of the cane is about five or six inches across and flows over bumps. I saw it as an advert on RNIB and thought it might be a bit of a gimmick, however, I love it. You do need to have a dedicated cane for it and then a separate cane for your road work.

Well I know this is a long article but I had a lot to cover and I've tried to be as concise as I can. If anyone wants to contact me, please get in touch with BBS UK and they will put you in touch with me.



Introducing...

Hi, my name is Laura and I have been recently appointed to the role of BBS UK Assistant Patient Liaison Officer, for patients and families attending BBS Clinics at Birmingham Children's Hospital and Queen Elizabeth Hospital, Birmingham.



In 2005, I completed a degree in Digital Media at Wolverhampton University; this was followed by a career working freelance on various media projects around the UK. To develop my knowledge and understanding of children's wellbeing in education, I worked in mainstream schools and became involved in supporting children and families with specific needs. During lockdown 2020, I completed an NVQ Level 2 in Counselling.

I live in Birmingham with my partner, Paul and three children, Eva, Annie, and Leo. I enjoy visiting new places and going on outdoor adventures such as canoeing, rock climbing and cycling. I am at my happiest when I am outside, walking my Cocker Spaniel, Buck, with my family and friends. I have really enjoyed getting to know many of you over the past few months, and I am hoping to meet you all very soon, during clinic days and events arranged by BBS UK.

Hi, my name is Shirin; I joined BBS UK in August 2021 as the Assistant Patient Liaison Officer for the clinics we run at Great Ormond Street Children's Hospital and Guys & St Thomas's Hospital, both in London.



I am a qualified Social Worker and, since graduating, I have worked within the charity sector as a social worker supporting

vulnerable adults, carers, and families from BAME communities. In 2018 I returned to university and gained an MA in Post-Qualifying Professional Practice, working with universities to improve their standards in social work education.

Since joining BBS UK, I have really enjoyed my role working within the clinics and as part of the Advice Service. Every clinic I attend is a new learning experience and I feel that it is the favourite part of my job.

In my spare time, I enjoy going for long walks with my friend to try out Indian Karak Chai at local tea shops. We often walk 6-8 miles at a time. I also enjoy watching TV. I love horror movies but then I have to sleep with the light on!

I'm looking forward to meeting you all soon.

Hi there, I'm Léonie and I joined BBS UK in November as the Admin, Engagement & Events Co-ordinator.



I've had a varied career, working with children, teenagers and adults within a range of contexts including community engagement, events, teaching, training, volunteer management and fundraising.

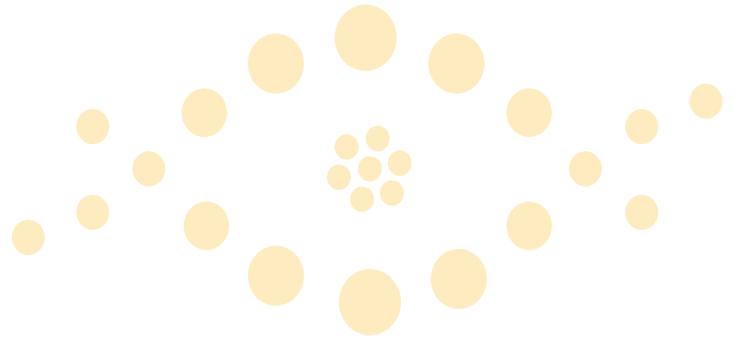
I live in Shropshire and love running in the woods or getting out into the hills on my mountain bike. I also play rounders and 7-a-side football. One of my favourite things is being part of a community pop choir; as someone who can't sing, I never thought I would 'perform' in charity concerts! I love being outside, going camping with my friends and walking on the coastal paths in Pembrokeshire and Cornwall.

I'm really enjoying learning about the great work that BBS UK does and am looking forward to meeting as many of you as possible at Conference in September.

A Review of BBS Research: 2021

Tim Ogden, President, Bardet-Biedl Syndrome Family Association (USA)

Tonia Hymers, Service Manager, Bardet-Biedl Syndrome UK



General Research Relevant to BBS Therapies

One of the most exciting possibilities for BBS treatment is gene therapy. The gene therapy landscape has changed considerably in the past few years and there have been important advances in one of the key technologies (CRISPR) that makes developing gene therapy for very rare diseases viable. Several gene therapies have been approved for patients in the United States, most notably for Leber Congenital Amaurosis (LCA), a genetic cause of retinal degeneration, and in the UK in March 2021, NICE approved a life-changing gene therapy for treating spinal muscular atrophy, making it the most expensive treatment ever approved by NICE.

Gene Therapy: BBS

As reported in 2021, Professor Phil Beales launched Axovia Therapeutics, with the aim of advancing gene therapy programs for treating early onset blindness and severe obesity in BBS. Axovia's novel gene therapy utilises an adeno-associated virus to deliver a functional copy of the faulty BBS gene in key tissues. In mouse-model preclinical studies, the novel BBS1 gene therapy modified the underlying disease of BBS, rescuing vision loss by halting retinal degeneration, stopping BBS induced weight gain and the development of obesity. Professor Beales will provide an update of progress at the BBS UK Conference on Saturday 10th September 2022.

Retinal Implants

One of the limitations of gene therapy is that it's highly specific. Not only does gene therapy have to be developed specifically for BBS, but it has to be developed for each BBS gene. However, there are other approaches

to treating blindness that are making huge progress, and apply to anyone with retinal degeneration regardless of the cause. Examples of this are retinal implants and, more recently, brain implants. There are also many studies being performed world-wide to look at various pharmacological compounds that can be taken to slow the rate of retinal degeneration.

Retina UK report that there are 'currently at least four microchip devices that are being developed and have been implanted in human eyes', however there are limitations; they are currently only available privately and are limited to restoring a basic functional sight, for example the outline of a large shape but not the ability to read. Retina UK also report that 'multiple research centres around the world are conducting tests on different versions of a retinal prosthesis and are continuously developing improved devices', which is really encouraging. Research is also developing into technology that stimulates the optic nerve or the brain directly.

New Basic Research on BBS

There were several important new basic BBS research papers published in 2021. While we have learned an enormous amount about BBS since the discovery of the initial genes, there are still plenty of things we don't understand, including why mutations in BBS genes lead to the specific symptoms of BBS.

As a reminder, all BBS genes code for proteins that are associated with primary cilia, which is a part of the cell that is important for sending and receiving messages from other cells. At last year's BBS UK Conference, Professor

Helen May-Simera updated us on her work investigating ciliary signalling mechanisms in kidney cells. The work of Professor May-Simera has shown that in BBS, dysfunctional cilia send out the wrong signals, resulting in incorrect reactions in the receiver cells.

Understanding all of the ways that cilia are involved in sending and receiving messages can help connect the dots on symptoms, but also suggest new avenues for possible treatment of certain symptoms. For example, Rhythm's obesity drug, Setmelanotide, works by boosting a certain type of signal associated with feeling full.

Dr Seongjin Seo, Dr Val Sheffield and Dr Ying Hsu produced an interesting paper in 2021 that helps to explain some of the underlying mechanisms that lead to BBS symptoms. Many of the BBS proteins come together to form something called the BBSome, a group of proteins that move 'packages' into and out of cilia. When there's a mutation in one of those genes the BBSome doesn't get assembled correctly and that affects how it does its job. Dr. Hsu, Dr. Seo and Dr. Sheffield demonstrated that photoreceptor cells act a bit differently to some other types of cells, depending on how the BBSome is impaired, and further, that the specific gene that is affected can change how well the BBSome works in photoreceptors. This is a promising avenue of research that can potentially help us to understand the varying patterns of vision loss that are specific to BBS.

In another paper, Dr. Hsu and Dr. Sheffield, along with many co-authors, studied brains from mice with BBS. They found that loss of BBS protein function leads to changes in the types of brain cells that the mice develop, which specifically affects how well the mice make 'fear memories.' This paper is important because it starts to help us understand the connections between cilia and learning differences in people with BBS.

Clinical Research on BBS

These discoveries in basic research about BBS are very encouraging because they lay important groundwork for future advances in treatment and therapy. There has also been

some research that's useful right now: clinical research on how BBS affects sleep.

One of these papers is based on data from CRIBBS and the BBS Center of Excellence at Marshfield. The team at Marshfield documents that people with BBS sleep less than recommended, especially those who have less physical activity. Since quality sleep is so important for so many other aspects of health, it's a reminder that daily physical activity is especially necessary for people with BBS. Separately, a team led by Dr. Helene Dollfus in France found that about 15% of the people with BBS who they evaluated had sleep apnoea, even though they didn't report common symptoms, like daytime sleepiness and poor sleep. The bottom line is that obvious signs of sleep apnoea may not be enough to identify sleep irregularities for people with BBS, and if there is suspicion of sleep apnoea, further testing such as a sleep study should be undertaken.

All in all, it was a very productive year for BBS research. We hope you are encouraged to know that progress is being made every week, and that a variety of researchers around the world are continuing to add to our knowledge of BBS and chart the path to our ultimate goal: effective treatment for all people affected by BBS.

Clinical Registry Investigating Bardet-Biedl Syndrome (CRIBBS)

CRIBBS is a BBS registry maintained at Marshfield Clinic in the United States, funded by the Bardet-Biedl Syndrome Family Association and currently led by Dr Bob Haws. Its aim is to promote scientific efforts to improve healthcare for individuals affected by BBS. CRIBBS has over 675 participants from across 40 countries and is growing. BBSFA President, Tim Ogden says, 'building and maintaining CRIBBS is a top priority of the association. There's nothing we could do to have a greater impact on research and treatment development.' For more information about CRIBBS and some of the research that has developed as a result, go to www.bbs-registry.org

Birmingham Sight Village: Information & Technology Event 2021

Birmingham Sight Village took place over two days on 17th and 18th August 2021 at Queen Alexandra College, Birmingham. Myself (Rachael Foley) and my fiancé, Richard Zimble, attended on 17th August.

After meeting friends at Birmingham New Street train station, we travelled in a taxi to the college wearing face coverings. Once at the college we were met by members of the Sight Village team, who registered us; we were shown where the hand sanitising stations were and allocated a member of the team to guide us around the event.

The stalls that were available for us to visit were:

- Advantage Carbon Fibre Canes (QAC)
- All Formats Transcription Services
- Associated Optical Limited
- Bristol Braille Technology
- Computer Room Services
- Dolphin Computer Access Limited
- Enhanced Vision Europe Limited / Optelec
- Focus Birmingham
- Guide Dogs
- HumanWare
- Orcam UK Limited
- Queen Alexandra College (QAC)
- Retina UK
- RNIB
- Seeing Solutions
- Sight and Sound Technology – Headline Sponsor
- Synapptic Limited
- VisionAid Technologies Limited

The event was smaller than normal due to covid but meant that it was easier to access



and the covid measures that were put in place made us feel safe around a large number of people.

Richard and I found the event really useful, both being registered blind and living with BBS. All exhibitors were very helpful and from attending the event we were able to purchase computer software, try out other technology devices/products, network with other service users who are living with a sight impairment and get updated information on a number of services that can support us in our day to day lives and with living independently.

Both Richard and I would highly recommend attending a Sight Village event to anyone living with sight loss, their carers, parents, family and friends and professionals. For more information about future Sight Village events and for dates in 2022 please visit www.qac.ac.uk



IT Equipment Fund Success!

Last year, we launched our IT Equipment Fund in response to an increasing need for people to access services through means of digital technology. Whether that be accessing online education and healthcare, paying for services online, interacting with social media or simply talking to a friend or family member over a video call. Members were asked to contact the Charity team to discuss how BBS UK could offer both practical and financial help with purchasing devices such as laptops, desktops and I-pads.

To date, the Fund has supplied 8 devices to BBS UK members, working alongside their families in order to source the most suitable pieces of equipment for them. Following a recent survey, the feedback we received indicated that all of the recipients have benefitted greatly from their new kit; members have stated that they feel far more connected with their friends, and also with other members of BBS UK. The equipment has also had practical benefits, enabling people to access online clinics, as well as other medical and

social meetings. The overall outcome has been that people generally feel much happier now that they have their IT equipment.

Dogan is one member who has benefitted from the scheme and, following his application, he was allocated a laptop. Since Dogan received the laptop, he has been using it to get in contact with his friends, to access television and online entertainment and he is planning to use it for his next virtual clinic. Dogan commented, "I can access information more easily, compared to using my phone. I can connect with people more and make more friends on Facebook. I feel happier and more connected with my friends." This is a great outcome and is exactly why we established the Fund.



If you feel that you would benefit from applying to the IT Equipment Fund, please contact Shirin at shirin.memi@bbsuk.org.uk

BBS Global Day

The first BBS Global Day was held on 25th September 2021.

BBS Global Day was a co-operative effort that was co-ordinated by BBS International, with support from the family organisations in France, Italy, the Netherlands, the UK (BBS UK) and North America (BBSFA). The aims of the day were to come together as an international BBS community to raise awareness, raise funds and to promote research.

The family associations, including BBS UK, worked with Axovia Therapeutics to produce a series of short film clips which were shared widely on social media. Our thanks go to Axovia and those members of the BBS



community who featured in the film clips.

We are now looking ahead to BBS Global Day 2022, the date of which is yet to be confirmed. By joining together as one BBS community, we are stronger and louder. Together we can improve awareness and understanding of BBS at a global level; the support and resources that we share with each other will ultimately benefit us all.

Contact Details

General Information

Léonie Hudson

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Tues - Fri, 8.30am - 5pm

Advice Service

Rebecca Perfect

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Mon - Thurs: 10:00am - 2:45pm

Fundraising

Christine Saxon

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10 hours per week

Patient Liaison Officer:

Birmingham BBS Clinics Service

Amy Clapp

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Mon - Thurs: 9.00am - 4.30pm

Patient Liaison Officer:

London BBS Clinics Service

Angela Scudder

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Mon - Fri: 9.00am - 3.00pm

Service Manager

Tonia Hymers

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Mon - Thurs: 8.30am - 4.00pm

