

BBS UK becomes a founding member of Bardet-Biedl Syndrome International

BBS Multi-Disciplinary Clinics: Coronavirus & Innovation



BBS UK
Conference 2021

Fundraising
An update on BBS UK fundraising and how you can support

Rebecca Perfect - a new Advice Worker for BBS UK



Personal Perspectives

Personal perspectives from Hollie, Helen and Daniel

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Newsletter

Winter 2020

Welcome to the BBS UK newsletter, Winter Edition.

Foreword

Hello everyone and welcome to the winter edition of our newsletter. I do hope you are all managing to keep safe and well during these difficult times. Sadly, due to Covid-19, we have had to cancel all our planned events this year, including our wonderful conference weekend and our family and adult activity weekends. We have missed these opportunities to meet up with you all and we hope it won't be too long before we will get to see you again. In the meantime, please be assured that everyone at BBS UK is continuing to work hard to move our charity forward and offer support as best we can.



We have embraced technology as a way of keeping in contact with our members and so far we have hosted three webinars which have provided updates on how Covid-19 is affecting us, how to stay safe and how to take care of our mental health and wellbeing. We have launched our new 'Booklet for Schools and Colleges', an excellent resource for those supporting young people with BBS in education. We also held our AGM, via Zoom, on September 28th, which gave us the opportunity to wind up the old charity and present the reports and accounts for the new BBS UK CIO; the new Board of Trustees was also elected. Because we weren't able to introduce ourselves in person during conference weekend this year, we held a Zoom 'Meet the Committee' event at the end of October; we have also included our profiles in this newsletter to help you get to know us.

I would like to take this opportunity to ask that everyone takes the time to complete the BBS UK Membership Audit that is available to complete online. The information you provide will be invaluable in helping us plan how to provide the support you need and devise forward-thinking strategies for our beloved charity.

We have already taken the decision to hold a virtual conference next year on Saturday, 15th May 2021 and we are working hard to make sure it will be informative and fun for everyone. We would like to wish you all a very Happy New Year and we hope 2021 will be a much better year for everyone. Take care.

Laura Dowswell, Vice Chair BBS UK

BBS UK Conference 2021

With the coronavirus pandemic continuing to have a significant impact globally and looking to continue throughout the winter and into the spring, BBS UK has had to take the very difficult decision to make Conference 2021 a virtual event. At the time of writing this, a second lockdown has commenced, and restrictions are looking like they will continue throughout the next six months and it is therefore no longer possible for us to deliver the event we had planned. We understand how disappointing this will be for our membership and truly understand the need to come together as soon as we possibly can, however the well-being of our membership and team is our primary concern.

We are however excited at the opportunities offered by a BBS UK virtual conference which will be held on Saturday 15th May. In particular, we look forward to being able to welcome our wider membership from around the UK and overseas. The event will be free of charge and will feature a selection of

keynote talks about research and development in the world of BBS as well as the much anticipated and valued personal perspectives. We also hope to provide members with the opportunity to take part in workshops and Q&A sessions. We are still in the planning stages of this and will be able to provide more information over the coming months. Rest assured that as soon as we are able to do so we will organise a membership wide event to enable us all to come together socially once more. Look out for regular updates about the agenda and how to book on our website and social media channels.



A New Advice Worker for BBS UK

We are delighted to report that Rebecca Perfect has extended her position with BBS UK to become our very first Advice Worker. Rebecca joined BBS UK in 2019 in an administrative capacity and in recent months has been developing the Charity database as well as providing support to our team as needed. Rebecca is a qualified Social Worker with considerable experience within this type of role. She also has experience of working within the charitable sector and is looking forward to having more contact with our membership. As of 1st February 2021, Rebecca will be available to provide advice and information to our members across many aspects of social care and education including:



- ✓ Referrals for Social Care Assessments
- ✓ Housing Applications
- ✓ Signposting for Adaptations
- ✓ Education Health and Care Plan applications and reviews
- ✓ Benefit applications including PIP, UC, DLA etc
- ✓ Carer Support
- ✓ Mobility Training (rehabilitation officer referrals)
- ✓ Blue Badge applications
- ✓ Local grants

In addition, Rebecca will be available to provide support and information in respect of the fluctuating COVID-19 pandemic, ensuring members have access to accurate and up to date information in an accessible format and providing support with accessing local services.

Rebecca can be contacted as follows:

Telephone: 07543 165804

Email: rebecca.perfect@bbsuk.org.uk

Working days: Mon-Thursday, 10:30am – 2:30pm

BBS Multi-Disciplinary Clinics: Coronavirus and Innovation

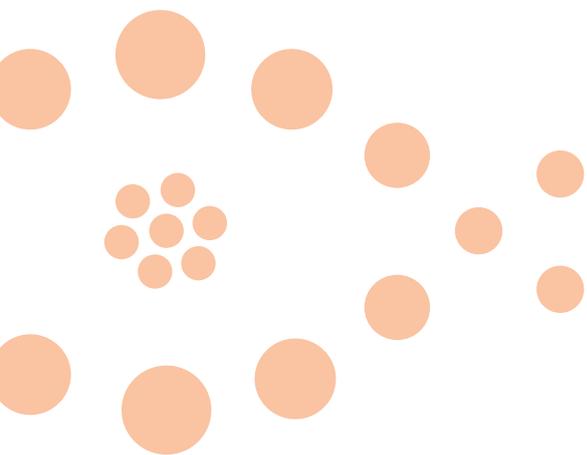
It has been a challenging year for our NHS and as a country we have collectively and publicly come together to express our gratitude and affection for this incredible institution and the inspiring individuals who work there. They have literally put their lives on the line for us this year and were once again ready to do so as COVID-19 cases rose sharply in Autumn. Our BBS clinicians are no exception and have all been part of this collective effort, through redeployment to frontline COVID-19 services, supporting frontline staff with their mental health, propping up auxiliary services and more.

Inevitably, the BBS clinics service has been impacted by coronavirus, more so in some areas than others, with cancelled clinics, redeployed staff and staff illness, and this disruption looks set to continue into the winter and beyond. We have worked closely throughout with the teams across London, Birmingham and NHS England to ensure the impact on our patients is minimal and the innovative and problem solving approach of all has once again showcased the BBS clinics service as ground-breaking and industry leading.

The BBS service had already begun developing a telemedicine service which enabled us to continue with appointments, albeit remotely. The only exception was Queen Elizabeth Hospital, Birmingham which had to temporarily

suspend clinics due to staff redeployment. As soon as it was able, it joined the rest of the service in offering remote appointments and there have been regular cross-service meetings to discuss best practice and seek ways of ensuring patients receive the best care they can.

The need for local testing (i.e. weight, BP, bloods) and the difficulties experienced with this was discussed in a meeting with NHS England and the service was invited to apply for funds for home testing equipment. The application was successful and soon, each patient will have access to a pack containing a blood pressure monitor, a set of scales and a finger prick blood test which will no doubt add value to the benefits of a telemedicine appointment.



Clinics at St Thomas', London and Queen Elizabeth Hospital, Birmingham have had some regular face-to-face appointments albeit with adaptations, and patients and team alike have valued this return to 'normality'. The photo above shows Professor Phil Beales and Dr Shehla Mohammed masked up and ready to go at the first post-lockdown clinic at St Thomas' Hospital, London.

We are advocating a flexible and individual approach, recognising that some individuals will not be able to travel to clinics or would rather not. It is becoming clear that over the coming weeks individual clinics may also need to be cancelled at short notice and we are fortunate to be able to offer telemedicine appointments as an alternative.

Needless to say, there is a great deal of organisation needed to bring each clinic together, face-to-face or virtual, and we ask that all patients commit to communicating their needs with us early on. Appointments are in extremely high

demand yet late cancellations risk appointments being left unfilled which is incredibly frustrating for the team. Not only do empty spaces impact on appointment follow-up times, but each appointment costs the NHS several hundreds of pounds; we all have a duty to protect this invaluable resource.

It really has been a challenging year and this looks set to continue for some months, however the support we continue to receive from our NHS funded specialised service has been exceptional. Our heartfelt thanks go to the BBS teams at Birmingham Children's Hospital, Queen Elizabeth Hospital, Birmingham, St Thomas' Hospital, London and Great Ormond Street Hospital, London for their continued commitment to our patient group.

Tonia Hymers
Service Manager



Vitamin D

Why do we need vitamin D?

Vitamin D is important along with calcium and phosphorous to maintain healthy bones, muscles and teeth. There has been lots of recent research demonstrating other important affects vitamin D may have on our health, but more research is needed.

Where do we get vitamin D from?

Sunshine, not food, is our main source of vitamin D. In the UK it is made by the action of sunlight on our skin between April and October. It is found in a limited number of foods, including oily fish like salmon and sardines, red meat and fortified foods e.g. some breakfast cereals and some yoghurts. It is also found in many dietary supplements.

How we check vitamin D levels at BBS clinics?

Since 2013 vitamin D has been one of the standard blood tests at the Great Ormond Street BBS clinic. The result is usually available about a month after the clinic appointment. If a vitamin D supplement is needed we contact the patient's GP. Any child with an 'insufficient' level requires a 'maintenance' vitamin D dose to be prescribed, so-called because it is enough to maintain an adequate vitamin D level. For lower 'deficient' vitamin D results, a higher 'treatment' dose is needed for a short period and this is then followed by an ongoing maintenance dose.

What do we know about vitamin D levels in BBS?

An audit from 2016 to 2017 found that 25% of the children attending our clinic required a vitamin D supplement. Our results are similar to the general UK population. Only 5% of our population needed a higher 'treatment' dose.

We looked at our vitamin D results again between April 2017 to October 2019. At the initial appointment, 26.5% of children required a supplement. Only 8% needed a higher 'treatment' dose.

Between these dates we saw some children twice and so were able to look at what happened to a few of the children who were

given vitamin D. When we saw these supplemented children for the second time we found that children with a deficient concentration had better vitamin D levels and the majority that had insufficient vitamin D levels showed an improvement after treatment. As a result of this audit we have rewritten our 'treatment' prescription letter making it clearer that ongoing maintenance vitamin D is required.

Are there guidelines for vitamin D supplements?

Yes, there are guidelines for supplementation in children and adults in the UK. A supplement may be required in the winter or all year round. In the GOSH clinic, we look at your vitamin D blood test result and advise whether a supplement is required.

Do I need a vitamin D supplement?

This year it would be sensible to take a supplement if you were inside more than usual during the summer months. Contact your GP or BBS team for some advice about whether you should take vitamin D and what is a suitable supplement for you.

Can I have too much vitamin D?

Yes! Taking too much vitamin D can be dangerous. Make sure you take only one vitamin D containing supplement unless they have been prescribed. Make sure your doctor and dietitian know about all of the supplements you are taking. (Remember to count cod liver oil as a supplement). In certain medical conditions, a different amount of vitamin D may be recommended to standard supplement doses.

Confused or need more information?

Contact your BBS clinic team or GP to discuss. For any families seen at GOSH please get in touch if you have any questions, I'd be delighted to help.

Sarah Flack

Principal Dietitian

Great Ormond Street Hospital for Children NHS Foundation Trust

Bardet-Biedl Syndrome International

Bardet-Biedl Syndrome UK are excited to announce we recently became a founding member of Bardet-Biedl Syndrome International (BBSI), together with other BBS groups from France, the Netherlands, Italy and the United States. The inaugural meeting was held on Saturday 3rd October and the following people/organisations participated:

Véronique Héloir:	President of the BBS Association, France
Francis Lestel:	Representative of the BBS Association, France
Bendert de Graaf:	President of the BBS Association, Netherlands
Dawn Patricia Hatcher:	Vice President of the BBS Association, Italy
Timothy Ogden:	President of the BBS Association, USA
Alex Ogeka:	Vice-President of the BBS Association, USA
Nate Arnesen:	Treasurer of the BBS Association, USA
Dr Robert Haws:	Head of CRIBBS, Marshfield Clinic, WI, USA
Tonia Hymers:	Representative of BBS UK
Professor Phil Beales:	Professor of Medical & Molecular Genetics, UCL Great Ormond Street and Institute of Child Health, London, UK

The goal of BBSI is three-fold:

1. To develop an international website, to host resources and provide a contact point to enable/facilitate countries with or without a BBS association to collaborate
2. Bring together all international researchers and doctors for collaboration and joined up working
3. To obtain funds to influence a BBS focused research agenda

The Association will be created under French laws and must have between 3 and 6 office holders, which initially have been agreed as:

President:	Véronique Héloir
Vice-President:	Francis Lestel
Secretary:	Dawn Patricia Hatcher
Treasurer:	Gregory Bouetel (Treasurer, BBS France).

BBSI will be managed by a Board which will consist of representatives from each member country/group. Each country has one vote and can provide up to three representatives.

The USA group have volunteered their services for the development and ongoing management of a website. They have a specialist currently working on the USA BBS website, who will be able to start working on the BBS International site once the USA site is completed. The website will be in English, however some pages will have translation options built in.

Dr Robert Haws and Professor Phil Beales have agreed to be 'Scientific Advisors'.

We are sure you will agree that this is an exciting collaboration and we look forward to seeing where this takes us all over the coming months and years. BBS UK are keen to hear from any members who would like to be considered as representatives of BBS UK on the Management Board of BBS International. Please contact tonia.hymers@bbsuk.org.uk for more information or to register an interest.

New Booklets for BBS UK!

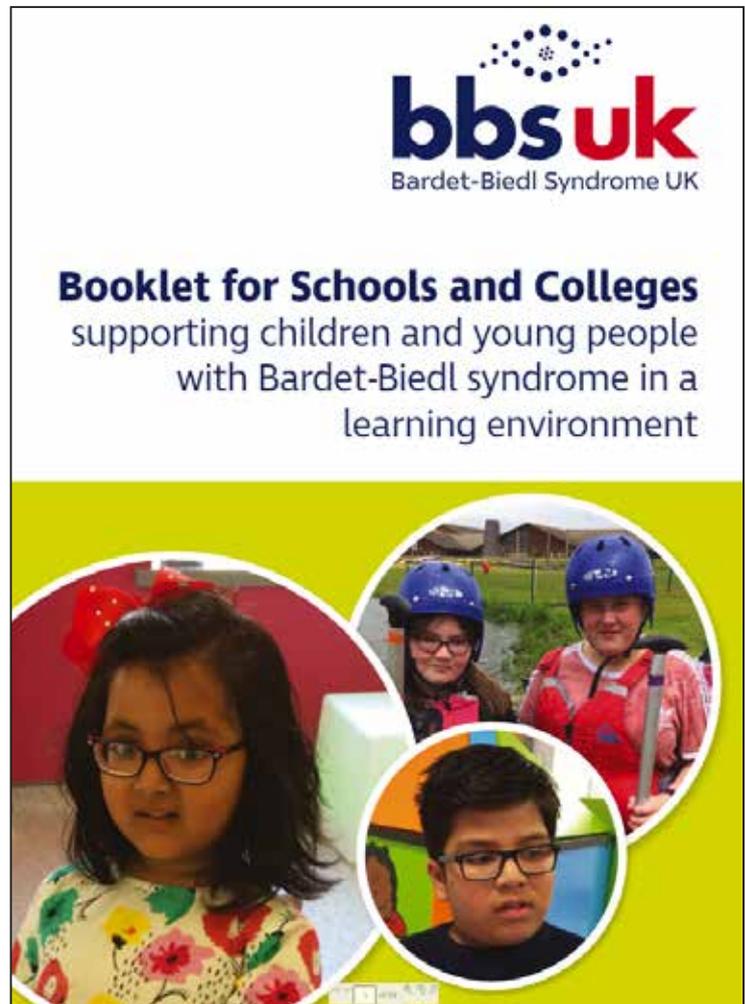
Booklet for Schools and Colleges

BBS UK have published two new booklets over the last few months. The first of these is our 'Booklet for Schools and Colleges', designed to promote a greater awareness and understanding of BBS and how it can impact on a child's journey through education from reception to age 25.

The booklet is aimed at the education professionals working with children and young people who have BBS, to enable them to more effectively support their students to reach their full potential. The booklet has been written in collaboration with education and medical professionals experienced in BBS and is based on the views of children and young people who have the Syndrome. It is packed with invaluable information and advice and includes signposting to further useful sources of support and information.

BBS UK Service Manager, Tonia Hymers said, "Young people told us that they were fed up with having to keep explaining about their needs and about BBS and some told us that they didn't feel believed because no one knows about the Syndrome. It was clear they needed support with advocating for themselves and with getting the right information across to their teaching and support team".

Each booklet has a fold out section for the child/young person to complete about how BBS affects them and the help they would like to receive. Teachers can then transfer this information to a Student Passport and share with the teaching and support team; student passport templates are available in the BBS UK website 'Knowledge Hub' via the link overleaf. A BBS UK Webinar for Parents and Carers, also available via this link, includes further information about how to use the booklet and also outlines Special Educational Needs and Disability (SEND) provision in schools. In-depth information about SEND and Education Health and Care Plans is also available to download.



The printed booklets were distributed to families in September/October, if you have not received a copy or would like additional copies, it is available to download via the following link <https://bbsuk.org.uk/education/> or contact admin@bbsuk.org.uk to receive a printed copy.

Introduction to BBS Booklet

In October we released the second of our new booklets, 'Introducing Bardet-Biedl Syndrome', which is a summary of the more in-depth Medical Information Booklet. 'Introducing Bardet-Biedl Syndrome' aims to answer some of the most commonly asked questions raised by those diagnosed with BBS, their parents/carers and extended support network and briefly outlines some of the key symptoms and care pathways available.

The booklet can be downloaded from the 'Knowledge Hub' on the BBS UK website in PDF, Word and Audio format and will soon be available in Urdu and Turkish. We are also adapting the publication into EasyRead which will make it accessible to those with a learning disability.

If you would like to request a copy of the booklet or require an alternative format, please contact us via admin@bbsuk.org.uk and we will be happy to help.



BBS UK Membership Audit and Strategy Planning

COVID-19 has disrupted our lives on many levels and that includes at BBS UK. Our events, and activities have had to be cancelled and even now we are still unable to plan the return of our valued social and activity weekends and annual conference. This enforced pause has been used wisely and we have taken the opportunity to look at our organisation and think about how we can develop and become stronger to enable us to better support our membership into the future.

The business term for this is 'Strategy Planning' and the aim is to:

- ✓ Understand the wants and needs of our service users
- ✓ Enable us to meet the needs of our membership and maximise our impact
- ✓ Provide purpose and direction
- ✓ Enable us to understand who we are, how we relate to the outside world, how we make the most of opportunities, respond to change, and face up to threats
- ✓ Clarify our priorities and identify what our resources should be focused on
- ✓ Focus on the medium to long term (3 to 5 years), with identified pathways to achieving our goals

Central and key to this process is the first item in the list above: 'understand the wants and needs of our service users'. To achieve this we have designed a membership survey to assess how Bardet-Biedl syndrome affects all those diagnosed and its impact on quality of life. It also aims to assess how well our members have been supported by the Charity, where the gaps are and what they would like from BBS UK in the future. We are making this survey available to all of our members to complete and we really hope as many of you as possible do so. We are so grateful for the support you continue to provide and cannot emphasise enough how valuable your suggestions and comments are for the development of our services.

This survey does not ask for and will not record specific identifiable data, i.e names and addresses, however we understand that some answers may inadvertently enable

identification. Individual questionnaires will not be shared with any other party or used for any other purpose. The collated, anonymised results from this membership audit will be used to improve our services and will be shared with our membership. The completed questionnaires will be treated with the utmost care, all data will be stored securely and according to the requirements of the Data Protection Act 2018 and GDPR.

It is so important to us that we hear your views to enable us to understand where we need to focus our resources over the coming months and years. The survey can be accessed via the homepage of our website which is www.bbsuk.org.uk.

If you would like further information or would like help with completing a survey, please contact Liz Loughery on 07421 312731 or via email, liz.loughery@bbsuk.org.uk.

Fundraising Round-Up

Friends of BBS UK

Thank you so much to all of you who have signed up to be a 'Friend of BBS UK'. Your regular gift allows us to plan strategically because we know, day in day out, that we will have a steady income that we can rely on. In our last newsletter we ambitiously aimed to sign up '100 Friends in 100 Days' and we haven't yet reached our target. If you are thinking about becoming a 'Friend' and would like more information, take a look at the enclosed leaflet. If you would like to speak to someone about the scheme, drop us an email and we will call you; contact details can be found opposite. By making a regular monthly donation, you will be helping us to forward plan and make long-term developments to our support and services for you, our members and your families. It need only be a small monthly gift, but for a small charity like BBS UK, every pound really does make a big difference.

Recognition for your efforts

Did you know BBS UK send out 'Thank you' cards and certificates to recognise fundraising efforts and special achievements? Children especially love them as they can take them to show and tell mornings at school. If you know someone who has raised funds or awareness for BBS UK, of any age, let us have their name, who they are and what they have done (with their postal address) and we will surprise them with well-deserved recognition.

Christmas Cards

All the entries in our Christmas Card competition have received a certificate and those highly commended received small gifts too. Well done to Ollie and Tom Sloane, Jessie and Bradley Springle, Aneeba Ahmed and Heidi Jenkins. The winning entry was from Tom Sloane; his card showed Frankie, his guide dog, getting into the Christmas spirit! Christmas card sales have been incredible and we have already sold over 600! If you would like to purchase a pack or two, they are £7.50 for 10 which includes VAT and postage costs.

Summer 2020

The BBS BBQ in August was probably the wettest day of the year and subsequently was a

wash out for all but a few hardy souls. Thank you to those who did take part, your soggy sausages raised around £250.

Even though many of us were restricted by social distancing or shielding, we were humbled that so many of you found ways to support BBS UK with your fundraising initiatives:

- Tom Horn, supported by friends, family and work colleagues at Utility Bidder, Corby, ran the Virtual Marathon in October 2020. He ran 26.21 miles in 4 hours 44 minutes and 29 seconds and after all the waiting, training and injuries, he smashed his target and finished up with a well-deserved beer.
- Claire Springle ran the Great North Run for BBS UK, with her friend Roland on 13th September and raised over £250 for BBS UK.



- Emma McGibbon, clinical psychologist for the GOSH clinics service, successfully swam the Channel with colleagues raising much needed funds for BBS UK in the process. A great achievement.



- Maylee Mason and a reluctant Liam Wilkie raised over £1,600 doing a 40 metre bungee jump into a gorge at Kilikrankie.



Caitlin and Niamh supporting their Dad from the ground!

Your fundraising efforts don't have to be extreme, though a lot of our supporters' appear to be!

The Sloane family continues fundraising all year round with quizzes, sales of upscaled items, bunting and more recently, face masks. Aneeba Ahmed continues to make bracelets and had her hair cut shorter for BBS UK. Judith Aylott raised funds by selling her amazing hand-crafted handbags on our Facebook page, as did Sandra Dale by selling exquisite knitted cuddly toys.

Families and friends have kindly sent us donations from £10 - £1,250 and with Gift Aid these all add up. Thank you so much, we are extremely grateful to you all.

What's Next

With continuing restrictions in place due to the coronavirus pandemic, we must think of raising funds in a different way. We know how inventive our members can be when it comes to fundraising (and we would love to hear your ideas) but here are a few suggestions to get you started:

- Set yourself a '7 things to achieve in 7 days challenge' and ask friends and neighbours to sponsor you.
- Make BBS UK your Amazon Smile setting on your Amazon page and when you buy, Amazon will send us a donation: Ask your family and friends to do this too.
- Go for a sponsored walk, run or jog in the park. Get fit for 2021!
- Get out the children's bingo wheel and have a competitive game of bingo on Zoom for friends or family.
- Play Beetle or Dominoes at home with the people you live with and charge a penny a point.
- Buy a large tube of smarties and save extra change in it through January, then send in the savings through Just Giving.
- Run a seasonal quiz with your family online via Zoom (20 Questions and Answers will be available on our website for a small donation).
- In the Spring we shall be running a 'virtual garden, patio, balcony or houseplant competition'. More about that next time!

Participants can make their donations via our Just Giving page:

<https://www.justgiving.com/lmbbs>

For more information about any of the above or for general fundraising support, email fundraising@bbsuk.org.uk

Wishing you all a peaceful, healthy and happy new year from the BBS UK Fundraising Team.

Christine Saxon

BBS UK Fundraising Coordinator

Having a Baby in Lockdown

by Daniel Evans

I was always told the three most stressful things to do in life are moving-house, getting married and having a child. I had already done the first two things as I had moved-house and gotten married and now we have had a baby.

In September Anna told me that she was pregnant which we were never sure was going to happen because of our disabilities, but with the help from the women's hospital Anna managed to fall pregnant.

As the months went on, we were never sure how we would cope with us living on our own and with our disabilities, but the midwives and the doctors at the hospital were very helpful and supportive and tried their best to find as much support for us as they could.

In March, who would ever have thought we would have a pandemic and things were shutting down, which meant we never got to go to any antenatal classes, but with the support of my mum, we did virtual stuff over the phone so we could practice changing nappies and clothes which helped us.

Anna was due on the 28th of April. A few weeks before we got to this date, we still did not know what was going to happen and whether I could attend. What the midwives managed to do was help us prepare for the birth without worrying about the pandemic, so it was agreed that Anna would stay in hospital for a few days after the birth to get the support with doing all the practical things with the baby,

either in a room on her own, or on the ward, depending on what was available, with the support of the nurses. Anna also had a learning disability nurse to support as well.

The 28th came and went and there was no sign of the baby during that week. There were times in the middle of the night when we thought Anna was going into labour, but she wasn't thank goodness! We were not sure what help would be available if I called in the middle of the night and I'm not very good in having to get up in the middle of the night to think.

On May 1st at lunchtime, Anna's labour started. I phoned the hospital and they told her to go in. Unfortunately I was not allowed to go with her, even though I tried to explain about Anna's disability. I was fortunate that I have a friend that works at the hospital so she was able to go in with Anna. When Anna arrived at the triage desk, instead of asking where I was, the team were upset and asking where Emily the guide dog was! This was because we were all a fixture at the women's hospital and Emily came with us every time. They said they had a bed set up if Anna wanted her in the hospital with her, but we explained that she was with a friend being looked after.

They kept Anna in the hospital. I was not allowed to be with her whilst they were waiting for a room and a bed, as the plan was to induce her as she was late. My friend was allowed to stay with her until she went into active labour which was good. I was allowed to take Anna's hospital bag to the hospital and my friend took it from me.

I sat by my phone waiting for the call to go and I had a neighbour and my carer on standby to run me up to the hospital. That weekend the team were very busy and they still hadn't found a room for Anna at that time in the

delivery suite. Eventually in the early hours of the morning on May 3rd, they found a room in the delivery suite but before they induced her they realised that she had already started to go into labour. I was given a call at 7am to say I could go. In normal times you are allowed to have two birthing partners but in these strange times the hospital are only allowing one, but with the understanding of our disabilities they were happy for our friend to stay with us.

When I arrived Anna was on gas and air and later that morning she had an epidural which made her comfortable but the baby took her time in moving! Around lunchtime they had to break Anna's waters to try and get things moving. When they did, she started to be in a lot of pain and eventually they worked out that the epidural wasn't working and, in the afternoon, they decided to put a new epidural in which worked.

Throughout the day the nurses were very good with Anna and the doctors were as well and every time they came in to check, I thought it will happen shortly but she pushed for over two hours and the baby still would not come out. Eventually after some assistance at 7:35pm the baby was born and she weighed 7lb 3oz and we called her Lily Brigid Evans. The hospital staff found a side room for Anna on her own and I left the hospital at 10pm and went home.

I kept in constant contact with Anna and a learning disability nurse to make sure that everything was ok and they were very impressed with how Anna was getting on; she managed to start breastfeeding right from the start with the support from the hospital.

It was agreed that my Mum would stay with us for the week to help with anything we needed.

Anna was coming home on May 6th but unfortunately that day did not go according to plan as early that morning in my house our fuse board blew. Whilst we were waiting for it to be fixed we were advised for Anna to stay at the hospital which the hospital were happy with. Once it was fixed, we went to collect

Anna and Lily and the staff explained everything to me and my Mum and Anna eventually came home, but she said she felt very weird leaving the hospital.

Over the first week we were very fortunate as the midwives came every day to check on Anna and also we had support from the breastfeeding team at the hospital plus the breastfeeding lady from the Children's Centre. When my Mum left at the end of the week we still had support from the midwives and the breastfeeding lady as well as having support from our carers.

Over the last six months we have managed to cope as best we can with the support from the Health Visitor as well as the breastfeeding lady plus our carers. We also managed to get some extra hours from the social workers to support us as well. Lily started teething very early on so this has affected us getting sleep but apart from that we have managed to cope extremely well with my Mum and Dad coming back every other week on a Saturday to help us as well.

Lily has started on baby rice and has taken to this very well. Anna is still breastfeeding and has done an amazing job in doing this and will continue for a few more months and she is also expressing milk using her hand pump.

If you would like to keep up to date with the progress of how we are getting on with the baby, you can follow us on our Facebook pages as pictures and stories go onto there.

On behalf of everyone at BBS UK, many congratulations to Daniel and Anna on the birth of beautiful Lily Brigid Evans and thank you for sharing your story.

My BBS Journey

By Helen Stirland



My name is Helen Stirland and I was officially diagnosed with BBS1 in 2011 at the age of 45. I was born with an extra finger on my left hand and an extra toe on each foot. I had these amputated when I was 10 months old.

I attended mainstream school where I had additional help with reading and I always had a weight problem (being constantly on diets) but apart from these things I didn't suffer from anything else. I was not a very confident person (not making friends easily) and I was bullied at school. When I left school, I attended catering college and my confidence improved.

When I got my first job in my early twenties, I attended a 'well-being day' and from that I had to see the firm's optician who referred me to the ophthalmology department at my local hospital in Doncaster. After that I was referred to a specialist at Sheffield Hospital where I underwent numerous tests. The outcome of this was the specialist thought I had something called LMBBS, but he advised me to carry on

my life as normal and to attend the opticians on a yearly basis. At this stage in my life I thought I had no visual problems. When I left my first job in catering I re-trained at the local college to do Business Admin and in 2001 I obtained a position with the Prison Service in an admin role.

Up to 2010 I was working and driving but noticed small changes in my vision and at this time I was wearing distance glasses. I had two small car accidents which made me realise something was wrong with my vision and went through my GP and was referred to the local hospital. They informed me that my peripheral vision was really bad and immediately stopped me from driving which came as a big blow. The Consultant also informed me that I would be registered as 'slightly visually impaired'. This upset me tremendously as I was unsure how I would continue to get to work as there was no regular public transport. After consulting my Manager, he informed me about 'Access to Work' which I pursued and therefore was able to use taxis to and from work.

My sister was concerned that my condition could also have been inherited by her two children, so she looked further into this from a medical point of view as she is a nurse. Through her investigations she found the LMBBS support group who I contacted and in 2011 I attended my first multi-disciplinary clinic at Queen Elizabeth Hospital in Birmingham. During this clinic I met two members who told me about the annual conference, so I made enquiries and attended my first conference that year. I have been attending every year since then (apart from this year due to Covid) and it was greatly missed.

In 2014 after attending a clinic and my peripheral vision had got considerably worse, I was re-registered as 'severely visually

impaired'. On returning to work I informed my Manager about this and was referred to the Occupational Health Department where measures were put in place for adaptations to be made so I could carry on at work.

I was referred to the Sensory Team attached to the local council and they visited me at home and gave me all the support, filling out the relevant forms and issuing me with a long cane and providing me with training. At first I found it extremely hard accepting that I needed to use the cane, especially at work but I needed to as my night vision was bad. Eventually after using it on a regular basis I accepted that if I wanted to carry on being independent then I would have to come to terms with using it. Now I look upon the cane as my friend and cannot go anywhere without it.

In 2017 I made the decision to apply for ill health retirement as it was getting to the point where I was not able to carry out my normal duties to the best of my ability even though all necessary help had been given to me. After all the medical checks and relevant paperwork had been completed, this was granted, and I left this job after sixteen years of service.

After finishing work it took me a long period to adjust to having no regular routine and I found it very hard to know how to fill my days. However, in 2018 I attended a Sight Conference held in Doncaster with two visually impaired friends and the Sensory Team. Through this conference I found out about the Partially Sighted Society which is based in Doncaster. I went along there and eventually became a volunteer which gave me a purpose again. Also, as I have an interest in history, I got involved with the Friends of the Doncaster Museums and also joined their committee where I took on the role of Fundraiser and Disability Officer.

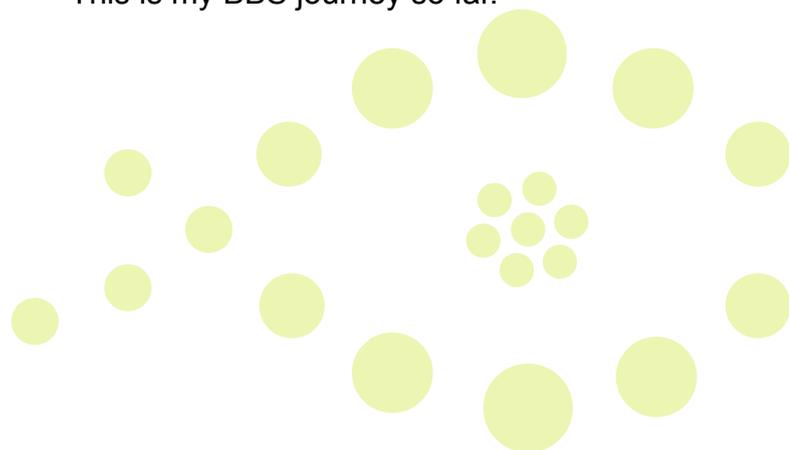
Through various groups I now have a wide circle of friends and my confidence over the years has continued to grow. I have many

friends in the BBS family which is a great help as we can support each other through good and bad times.

This year has been especially hard for everyone during the Covid pandemic. Personally, I came to live with Mum during the national lockdown period as we normally do a lot of things together. As I only live three doors away we usually have our main meal together unless either one of us is out with friends. I struggled with not being able to socialise with friends and attend my various groups but as lockdown eased, I started going out a little bit more until recently.

As you will appreciate the thing I have struggled with most is accepting not being able to do things that I used to be able to do especially as I was diagnosed with BBS so late in my life. It has been in this last three years that my eyesight has deteriorated so quickly that I get frustrated with the simplest of things that I have to get help with. I am consulting with friends for help and advice of how they cope in doing simple things with their eyesight being affected earlier in their life. I think, for me, one of the biggest difficulties I have is asking for and accepting any help. During the last three years I have suffered emotionally in accepting my condition so, therefore, I have had some counselling and support from friends and family. I now accept that my eyesight could get worse or stay the same, so just have to take each day as it comes and look for new ways in doing things.

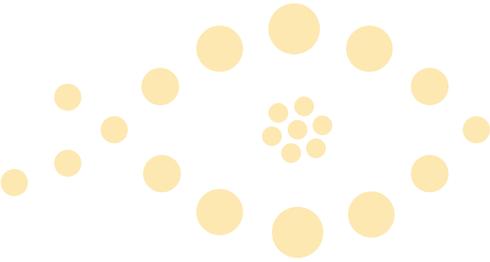
This is my BBS journey so far.



My holiday in lockdown - by Hollie Sales



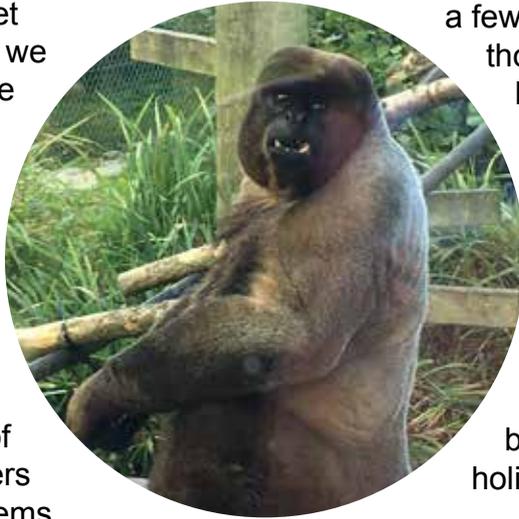
We went to Portland Bill in Dorset in October for a week; we stayed in a cottage with a hot tub. We arrived late evening on Saturday 10th October, we couldn't get in until 5pm and had to leave at 9am on the following Saturday, due to them deep cleaning because of Covid. The place was clean and tidy, some facilities weren't available but everything we needed was there. The place and scenery were absolutely incredible and stunning. We had a lovely view of Portland Bill Lighthouse right outside our front room window.



We went to lots of beaches, Portland Bill, Lulworth Cove, Weymouth and Monkey World. We just don't realise what lovely places we have in the UK.

We had to wear our masks the whole time around Monkey World, as even though it's outside, it's quite enclosed, and the monkeys can catch

Covid too. It was so quiet there, but it was nice as we could get up close to see the monkeys, instead of loads of people in our way, so that was an advantage although it was very weird, as it's normally packed.



a few people in there, so it wasn't as bad as I thought. Now I've done it, it made me feel better to go into another one but only when it's quiet; small steps. We always carry hand sanitizers wherever we go. We didn't go to any restaurants, as I'm not quite ready for that yet. We cooked at our cottage all week.

We had a lovely time, but still felt the uneasiness of Covid in the background but it won't stop me going away on holiday again!

We went into a couple of shops, they had sanitizers in places, one-way systems and social distancing, which was all good. Some were busier than others. When it's busier and there are lots of people it makes me feel a bit uncomfortable.

We went to pubs but mainly sat outside, as I feel more comfortable doing that, we did venture inside one, as it was raining, I was very nervous and anxious but there were only



Meet the Board of Trustees

Our virtual AGM was held at the end of September and members elected a new Board of Trustees. Trustees give up their time voluntarily, making sure the Charity is run legally and is financially compliant, sustainable and carrying out its objectives. Usually, the AGM takes place as part of the conference weekend at the Hilton hotel in Northampton where we can chat to you all and get to know everyone a bit better and share ideas. Due to COVID-19, that wasn't possible this year, so it was decided to hold a virtual Zoom meeting on the evening of 29th October to give everyone the opportunity to meet the Trustees in a different way. We would like to thank everyone who was able to attend, we enjoyed chatting with you and it was a super opportunity to answer any questions you had for us. The newsletter is another opportunity for us to introduce ourselves and we hope you feel you know us a little better once you have read our profiles.



Stefan Crocker: Chair

I currently hold the post of Chair of the Board of Trustees for BBS UK and it is my responsibility to provide leadership and support to the Committee to ensure the effective

governance of the Charity. It is also up to me to ensure the Board works closely to achieve its agreed objectives and that all work carried out is in accordance with the vision and key values of the Charity.

I was diagnosed with BBS in 2003 when I was investigating laser eye surgery for my astigmatism. I was referred to Moorfields in London where they diagnosed me with BBS. I endeavour to inspire BBS adults via education as I graduated in 2014 in Politics from the University of Portsmouth.

I like to encourage fellow BBS adults to travel around the country. I'm very confident on both

over and underground railways and visit friends dispersed around the country. In my spare time I love to go to concerts and have been to many over the past few years. Concert highlights include Eric Clapton, Fleetwood Mac and Iron Maiden.

I find my position as Chair fulfilling and enjoy all aspects of role. I am particularly grateful for having the opportunity to get involved in the organisation of the BBS UK conferences and activity weekends, to make them as successful as possible for our members. At the conferences I also enjoy acting as an ambassador for the organisation and being someone that attendees can comfortably approach to chat to or ask questions.



Laura Dowswell: Vice-Chair

I've been married for almost 30 years to the ever patient and laid-back Nick and we have two children – well they're not really children

anymore! Maria is 27 and Greg is 24. We live right by the sea at The Witterings on the south coast and enjoy the beach life in summer and winter.

We became involved with BBS UK after Maria was diagnosed with BBS at age 14 and we attended our first conference 11 years ago. We were amazed at the amount of support and information we got and the lovely, hopefully lifelong friends we made.

As a family, and of course BBS does affect the whole family, we were determined to 'give something back' and I now provide support to families at the annual conference as well as being a trustee and the Vice Chairman of the Charity.



Abbie Geeson: Treasurer

I joined BBS UK in 2015 and I am currently the Treasurer. I feel very passionate about the organisation and would like to support the Charity in any way I can for as long as I can

as it is such a vital support network to us all. I'm mum to Jessica who is 21 years old and we live in Brighton. Jessica was diagnosed with BBS back in 2014 after many years in and out of hospitals with various health conditions and since her diagnosis, everything has made more sense to us. Around the same time, I was also diagnosed with multiple sclerosis. As Jessica and I both have a long-term health condition it can be a challenge, but we have so much support and we have been welcomed into the BBS family and have truly embraced it. I have worked in the charity and voluntary sector for fourteen years and I have valued learning more about BBS UK and very much enjoy working alongside a great team of trustees and employees for an invaluable charity.



Dianne Hand: Secretary
George, the youngest of my two sons, was diagnosed with BBS at the age of 14 in 2010. After receiving tremendous emotional support and learning so much about BBS from people connected with the Charity,

I wanted to show my gratitude by supporting others and being a trustee enables me to do that. I am proud to have just become the Secretary of BBS UK and I am looking forward to what I am sure is going to be a very rewarding challenge, working alongside a super team of people who work hard for our wonderful charity. I live in Stockport and for many years I was a teacher and special needs co-ordinator in a primary school, so I have been able to pass on useful advice and information.



Emma Oates: Trustee
I'm married to Steve and we have two children. Our son, Tom, is 23 and our daughter, Katie, is 22. Both Tom and Katie have BBS.

Myself and my husband run a touring caravan park in a pretty village in North Yorkshire, where we live with our dogs, cat and an array of 'outdoor' animals. My husband Steve also runs a plumbing business.

We have been involved with the Charity since 2010 and have attended most conferences since then.



Rachael Foley: Trustee
I'm 40 and live in Stockport. I was diagnosed with RP in 1996, and BBS in 2012 following genetic testing.

I attended mainstream school and college, studying Leisure and Tourism and worked in a travel agent for a year before moving on to work for Stockport Council working in various roles such as: conference organising, campaigns and social media; crime and disorder, anti-social behaviour and domestic abuse. Unfortunately, due to several restructures within the Council, and my deteriorating eye-sight, in July 2017 I retired on medical health grounds.

I have since become involved in blind football, goal ball and sound tennis; and have gained a Level 1 Tennis Association Coaching Certificate. I'm also the vice-chair of Greater Manchester VI Bees, a voluntary community group for blind and visually impaired people. I provide help administering their Facebook page and with the organisation and planning of social and sporting events.

I'm also the joint Vice-Chair of RNIB, North West Network Committee. I am a Community Connect Volunteer and Sight Loss Ambassador for the RNIB, raising awareness in the community. In conjunction with the RNIB, my friend Lynne and I have recently set up a telephone social group called "Natter up North", to bring people together, make friends and stop social isolation, particularly during the testing times of the COVID-19 pandemic.

I enjoy socialising with friends and family, going on theatre trips, concerts and eating out and I am a Manchester City fan.

I have played an active role with the BBS UK Board since 2017 and was elected as a trustee in April 2018. As I have the Syndrome myself, I am aware of the difficulties that members and their families may be encountering.



Judith Aylott: Trustee

I am mother to two boys, who each have three children, so I am grandmother to six! Two of my youngest son's children have BBS.

I have had a varied life! After leaving school I

trained as a Fashion Buyer for a London Department Store, and then was a Department Manager at Habitat. After having my two sons and becoming a single parent, I retrained at a local college and have held various administrative posts including 12 years as a PA to the Director of an educational company. After remarrying and moving to Cambridgeshire, I worked at the Police Headquarters in the Criminal Records Bureau department. I didn't go to university, having attended the University of Life! I involve myself with local events, belonging to the Women's Institute and have served on the Parish Council for 14 years.

I attended my first BBS UK conference in 2017, with my son and his family; I was so moved that I decided I wanted to do something to help the Charity. I started by selling my handmade items, but felt this wasn't enough, so became an 'Observer' on the Committee in 2018. I have since become a trustee.



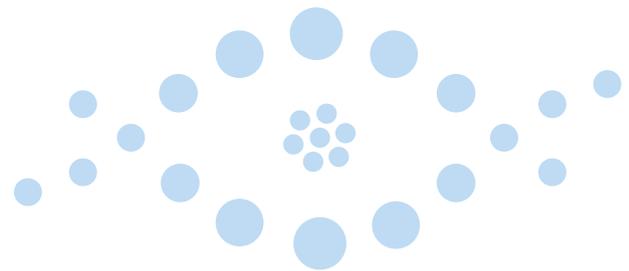
Virginia Cashin: Trustee

I have two grown-up daughters and in 2014 Lydia, my youngest who was 22, was diagnosed first with rod-cone dystrophy and then with BBS later that same year. The information available on the BBS UK website

was invaluable in leading us to request an appointment at a clinic, and the support and warm welcome we received there and at our first conference were wonderful. Lydia often says how much it has all helped her to understand herself.

We live in Berkshire and I work as a PA in a secondary school. I enjoy dancing in my spare time. I was an 'Observer' on the Charity's Committee in 2019 before becoming a trustee this year.

If you would like to contact any of the Trustees, please email us at admin@bbsuk.org.uk



Final Call for Membership forms

We are still compiling our database following BBS UK's transition to a Charitable Incorporated Organisation (CIO). As previously mentioned, we require all current members to complete a form to give us permission to continue to contact you and for you to remain a member of BBS UK. Simply put if you want to keep hearing from us – we need your permission!!

We would like to thank all those who have returned their forms so far – it is greatly appreciated. To date we have received and inputted information for over 300 members.

Whilst this is a significant number it indicates that there are still many people who still need to return their forms, to keep hearing from us. If you are unsure whether you have returned your form please email Rebecca Perfect on rebecca.perfect@bbsuk.org.uk or call 07543 165804 and we can check on the database to see if we have your details.

As always, membership remains free and ensures that our valued members receive communications such as regular newsletters, our annual conference report, updates on treatments and research and fundraising news, as well as invitations to a variety of BBS UK events and activities.

BBS UK Seeking New Trustees!

Trustees play a pivotal role in setting the strategic direction of the Charity, as well as ensuring the Charity delivers the best service it can every day. For this voluntary role, the Board are looking for individuals who are motivated, passionate about the voluntary sector with relevant professional expertise and experience, in particular across the areas of human resources, finance, management or leadership. It is essential that the Board is representative of the diverse population we support, with a range of experience and skill-set to draw upon.

The role of Trustee is a voluntary one (non-remunerative) however all reasonable out of pocket expenses are paid. The minimum requirements are:

- Attend three board meetings a year plus the Annual General Meeting (AGM)
- Participate in regular meetings/calls, via online platform (i.e Zoom) and telephone
- Engage with other Trustees via email
- Attend the BBS UK Conference.

How to apply

If you would like to express your interest in becoming a trustee, please request an application form from Liz Loughery via email at liz.loughery@bbsuk.org.uk

This role will require an enhanced DBS check to be undertaken.

Contact Details

Office Manager

Liz Loughery

07421 312 731

liz.loughery@bbsuk.org.uk

admin@bbsuk.org.uk

Tues-Thurs, 9.00am - 4.30pm

Patient Liaison Officer:

Birmingham BBS Clinics Service

Amy Clapp

07591 206787

amy.clapp@bbsuk.org.uk

Mon-Thurs, 9.00am - 4.30pm

Fundraising Officer

Christine Saxon

07512 198484

christine.saxon@bbsuk.org.uk

fundraising@bbsuk.org.uk

10 hours per week

Patient Liaison Officer:

London BBS Clinics Service

Angela Scudder

07591 206788

angela.scudder@bbsuk.org.uk

Mon-Fri, 9.00am - 3.00pm

Advice Worker

Rebecca Perfect

07543 165804

rebecca.perfect@bbsuk.org.uk

Mon-Thurs, 10:30am – 2:30pm

Service Manager

Tonia Hymers

07591 206680

tonia.hymers@bbsuk.org.uk

Mon-Thurs, 8.00am - 4.00pm



bbsuk
Bardet-Biedl Syndrome UK
www.bbsuk.org.uk
admin@bbsuk.org.uk



On behalf of everyone at BBS UK thank you to Helen, Daniel, Hollie and all those that share their stories with us, we are very grateful for your personal perspectives as I'm sure are our readers. If anyone is interested in sharing their story for future additions of our Newsletter then please get in touch with Liz Loughery (details above) – we would love to hear from you!