bbs uk
Bardet-Biedl Syndrome UK

Annual Conference Report 2021

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Welcome!

Welcome to the BBS UK Conference Report for 2021. For those of you that don't know me, I'm Laura and I am very proud to be the Chair of our fantastic charity, BBS UK.

This year, our conference was held virtually due to the COVID-19 restrictions. Although we all missed not being able to meet up in person, it did mean that we were able to welcome attendees from the comfort of their own homes, and from all around the world, including America, Germany, France, Spain, Netherlands, Ireland and Australia.

For the first time ever we held two separate sessions over two days and the Committee would like to thank all the speakers who gave up their valuable weekend time and continue to support BBS UK and explore our complex syndrome with such dedication.

Friday was the professionals' session, aimed at those working with people who have BBS. Professor Phil Beales brought us up to date with research news and we welcomed Dr Bob Haws from the BBS Centre of Excellence, Marshfield, USA, who shared information about their BBS clinics, patient registry and research. Both Phil and Bob held a Q&A session which proved very popular with a wide range of questions being asked.

Saturday's session was aimed at those living with BBS, their families and carers and again, we had a wide range of talks including information and support about anxiety and low mood, and an update on the new weight loss drug, Setmelanotide. We also heard from the BBS UK staff team about current services, a BBS clinics update, our five year strategy and fundraising.

The highlight of both days was the personal perspectives which are always incredibly interesting and resonate with so many of us. We are incredibly grateful to those who shared their journey with us in this way. Summaries of all the presentations and personal perspectives can be found within this bumper report; the full unedited recordings can be found on the BBS UK YouTube channel.

BBS UK is here to support all those living with the syndrome, their families and carers and the professionals involved in their care, and as you will read, the Charity has so much to offer in terms of information and support services so please do keep in touch, make sure that you are on the mailing list to receive news, and we really hope to see you at future events.

SAVE THE DATE: Planning has already begun for **Conference 2022** which will be held across the weekend of **9th - 11th September 2022**, at the Hilton Hotel, Northampton. Further details will be available on the BBS UK website in the coming weeks and months; we look forward to seeing you there!

Laura Dowswell
Chair, BBS UK

Update on Research and Study of BBS

Professor Phil Beales
Great Ormond Street Institute of Child Health
Guys and St Thomas NHS Trust

Professor Beales welcomed everyone to the conference and provided an introduction to his scientific and medical update. Professor Beales explained that this year is the 10 year anniversary of the highly specialised BBS service and gave an overview of the incredible journey of the service since its inception; a journey that has been welcomed by professionals and the patient community alike, and has led to significant accolades from the NHS and incentivised many other patient groups to provide services based on the BBS model.

Professor Beales presented the BBS model which is made up of six elements that comprise four different hospitals; Birmingham Children's Hospital; Queen Elizabeth Hospital, Birmingham; Great Ormond Street Hospital, London (GOSH) and Guy's and St Thomas' Hospital, London. Pivotal to the service is the work of BBS UK, who provide unparalleled support that clinicians are unable to provide. The sixth component of the service is the DNA Lab led by Dr Beth Hoskins through the Great Ormond Street Hospital genetics centre. This Lab provides a national reference service for the whole country and offers next generation genetic testing, comprising a 21 BBS gene panel.

To date, 900 tests have been completed and diagnoses have been confirmed in patients with BBS who fulfil the genetics criteria, as well as having a supportive molecular diagnosis.

Current to the end of 2020:

- 80% of families have had a diagnosis confirmed
- 224 carrier tests have been undertaken
- 31 predictive tests, including prenatal tests have been undertaken
- Approximately 200 mutations have been defined
- Approximately 22% of patients do not have an identified mutation
- Approximately 33 new referrals are received into the BBS service every year

Professor Beales presented a pie chart showing that the majority of patients are diagnosed with the BBS1 gene (32%), followed closely by BBS10 (15%).

Professor Beales reported that, with the help of colleagues, he has built a patient registry with the purpose of providing ongoing clinical improvement in terms of care and service and to facilitate audits, research and to support clinical trials. The aim is to provide this to all BBS centres and also to other colleagues within the NHS, clinical researchers and trial centres.

A robust governance structure for the patient registry has been set-up, which includes a registry committee of service leads, lay people, including those from BBS UK, and members of NHS England.

Professor Beales gave thanks to the team who have designed and built the registry, Ian Kemp, Theodora Dyakova, Kath Sparks and Dr Elizabeth Forsythe.

The registry has been devised as a secure web application and its purpose is to manage databases and online surveys. There are 307 records uploaded to date, and this figure is expected to double as other centres become involved.
Professor Beales explained that the information collated via the BBS registry can help us to better understand what the patient journey looks like, as well as the natural history of the syndrome.

For many years the syndrome was believed to be understood, however, now that more data is being collected and analysed, a much more detailed picture of the disease progression is being established. For example, 84% of patients are born with extra fingers and extra toes, and at around age two, approximately 88% of patients start to show signs of development delay and behavioural difficulties. At around four years of age, the data shows onset of weight gain and the first signs of visual impairment. The database also reveals that about 40% of patients

will develop renal problems and 26% will have diabetes. It also highlights a trend in BMI with virtually all BBS patients well above the average in this respect.

Professor Beales went on to talk about research and reported an inexorable interest in BBS with 54 publications to date this year (May 2021), with a key topic of interest being the role of cilia in BBS. Professor Beales explained that primary cilia are short membrane protrusions that stick out from the surface of virtually all the cells in our body. They function as a cellular antenna that receives information about the environment and they locally transduce that information into a cellular response. It is now understood that mutations that code a number of these defective cilia proteins, underpin ciliopathies in general, which helps to explain why retinal degeneration leads to visual impairment, why cystic kidneys appear and even the extra presence of extra fingers and extra toes. Finally, he said, they are beginning to better understand the role of cilia in obesity as well.

Obesity is an imbalance in terms of energy intake and expenditure. These imbalances are seen in patients with BBS and Professor Beales explained that this is because of a specific deletion of BBS proteins in key neurons in a part of the brain called the hypothalamus (the appetite control centre) which leads to an increase in food intake and thereby increased weight gain. Professor Beales said he was hopeful that this new evidence will give rise to the development of new therapies.

To finish, Professor Beales shared a study by Andre Stepanek's group from Charles University in Prague, that looked at immunological defects associated with BBS that may lead to previously unrecognised autoimmune features in some patients. For example, an increase in the amount of rheumatoid arthritis, type 1 diabetes, hypothyroidism and other autoimmune conditions indicating the importance of BBS proteins for immunity.

Tanja Chaudhry: *A Personal Perspective*

Hello everyone, my name is Tanya Chaudhry. I have three children, my youngest, Anayah has BBS, she is 6. I had a very straightforward pregnancy, everything was fine, no problems whatsoever. The first thing we noticed when Anayah was born, was that she had an extra digit which straight away, was a big alert for everyone, for the doctors, especially. They began trying to figure out what was wrong with her, telling me there were so many different syndromes out there and doing so many tests.

When she was around 2-3 months old, I realised that Anayah's vision wasn't right and so I contacted her doctors who referred us for genetic testing. At the time, Anayah was gaining weight, she was hungry all the time, there was something wrong with her eyes, and there was the extra digit. One of our doctors in Harrow, started linking the symptoms and talking to me about BBS as a possible diagnosis, however said that I shouldn't panic or start Googling because it wasn't certain and the doctor would rather explain everything to me instead of me seeing all the bad things about BBS. Obviously, I didn't listen. I went home and Googled it. The first thing that I came to know was about the vision loss, which for me was the worst thing that could happen, I think.

I remember the day we got a phone call from the doctor, she said, "I'm really sorry to tell you, but Anayah has the faulty gene, BBS10."

There were lots of other tests to be done, but at the time I had so many questions and I didn't know who to go to. The doctor told me about the BBS UK support group on Facebook and I straight away contacted them. We were invited to our first conference - it was mixed feelings, positive, and negative at the same time. But

finally, we had someone who could answer all the questions we had because it was new to us. We never had anything like this in the family, no disabilities.

We came to Conference and first sat in the car for a while, watching everyone from the outside. A lot of you guys used a cane, which again, was very new to us, and I thought, ‘Okay, this could be Anayah's future’. Since then, we have met many of you, and we are so looking forward to coming back to the BBS UK Conference every year. The kids were really devastated when we couldn't come in 2020 and 2021 because of the pandemic. We have made so many friends, the kids and me. You feel like you are talking to your family because you all understand what we are going through. Every time I have a question, I just come to you, or if you guys have a question you come to me, and we help each other in general. We get so much support from all of you.

Anayah has lots of health issues but I can also say, living in Luton, I have had lots of support as well. The school Anayah goes to was recommended by one of the parents I met at the BBS UK Conference. She said to me, "If Anayah needs to go to a mainstream school, please send her into that school because they have a vision impairment unit”. That was the best decision I ever made, there is so much support, I think they already had a child or two who had BBS before, so they really know what to expect when it comes to Anayah, which is great.

And not only the school, but from when Anayah was little and diagnosed with BBS, we have had people coming to our house to support us with everything. Also, the local hospital, the eye clinic, for example, our eye doctor, he knows everything about BBS as well, which is helpful, because then they do understand and you don't have to explain yourself over and over again.

The BBS clinics, even though it's a really hard day, a full day and exhausting, we still look forward to it because at least we can see some of you. Over a year, things change with Anayah and we get everything checked in one day. It's quite relaxing and a nice environment as well, so again, since day one, there has been support all the time, which made us feel really grateful.

Anayah, she has good and bad days. She has been struggling with her vision and struggling now to see at night-time as well, so we think night-blindness has kicked in. Again, the doctor from the hospital is telling us not to worry, that it might slow down or stabilise for a while - no one really knows how fast or how slow things will happen. We just take one day at a time, really. Again, without all of you, it would be so much harder and we're so grateful to have all of you on board to help us out. You're our little family or big family really, and we're really, really, grateful.

USA Center of Excellence for BBS:
Patient Registry and Research

Dr Bob Haws
BBS Center of Excellence, Marshfield

Dr Haws began his presentation by saying how disappointed he was at not being able to meet everyone at last year's BBS UK conference weekend which was cancelled because of COVID-19; he was, he said, very much looking forward to meeting everyone in the future.

Dr Haws showed a photograph of a little girl called Ashley who he first met over 20 years ago, when an ultrasound scan showed polycystic kidney disease. Ashley was born, nearly at term, with very poor kidney function and Dr Haws noted that unlike other patients with kidney failure, Ashley loved to eat, which, along with other symptoms caused Dr Haws to start looking for a diagnosis, ultimately settling on Bardet-Biedl syndrome. This was before Professor Beale's landmark paper defining the diagnosis criteria for BBS, which Dr Haws said was absolutely instrumental for helping so many clinicians to make a diagnosis.

Another landmark development, Dr Haws explained, was the understanding of the role of cilia in monitoring the flow of urine through the kidney, responding to various hormonal actions to control the amount of urine output we make. This is disrupted in BBS with those affected experiencing excessive urine output.

Diagnosing Ashley with Bardet-Biedl syndrome was a significant moment for Dr Haws, because Ashley's mother organised the BBS Family Association and encouraged him to attend their conferences, providing the opportunity to hear people's concerns, including that healthcare providers knew less about BBS than those affected and their parents, and about how fragmented healthcare was, with providers not working together. Dr Haws also spoke about the importance of family networking, with his patients and their families desperately wanting to meet others with the syndrome to ask questions and learn from each other.

Dr Haws started developing the Center of Excellence for BBS at the Marshfield Clinic in central Wisconsin, where they assembled specialists in cardiology, neurology, neuropsychology, dermatology, medical genetics, physical medicine, rehabilitation and orthopedics, dietitians, occupational therapists, speech therapists and so on, all working together with the common goal to provide the best care possible to patients with BBS.

The Center of Excellence sees BBS patients from all over the United States and also from overseas, including Australia, Saudi Arabia and territories of the United States and Canada. Families are provided with a very comprehensive binder containing all their health care information; a copy is also sent to their local healthcare providers. Whether patients can return to the center again or not, the service continues to have relationships with their healthcare providers, to ensure ongoing coordination of care.

Dr Haws went on to talk about some of the activities the Center has organised, including a Lions Camp, a week-long activity break attended by 75 individuals, taking part in archery, goalball, volleyball and dances etc. There is also a Movie Night, Book Club, Parents Support Group, Weight Watchers Group and a Young Adults Support Group, all providing a forum to talk and share experiences.

A key aspect of Dr Haws' work is the Clinical Registry Investigating Bardet-Biedl Syndrome, (CRIBBS). CRIBBS gathers comprehensive health information from patients and puts it in a repository which can be used by researchers to understand the complex features and to develop effective treatments. Dr Haws explained that the first question he wanted to answer with CRIBBS was whether kidney transplants could be effectively used in Bardet-Biedl syndrome because in the United States, if an individual's body mass index is too high, they're not allowed to have a kidney transplant.

Dr Haws' team compared the outcomes of 21 individuals with BBS who had kidney transplants from across the United States and globally. They found that at 25 years post-transplant, 50% of the kidneys were working in people with Bardet-Biedl syndrome compared to 22% in those who didn't have BBS at 20 years post-transplant

The team were able to use the registry to show that transplantation is possible in those with BBS. That particular piece of research from CRIBBS was the largest series yet published about kidney transplant outcomes and Dr Haws expressed his appreciation to all those who had shared their data in this way.

The team have also used CRIBBS to look at dental issues, weight patterns, sleep and activity patterns, seizures, skin disorders and more recently, genetics. A key question is whether one gene type of BBS is more prone towards kidney failure than another? Using the database, which now contains the data of around 650 individuals, the team found that about 7% to 8% of individuals who have BBS go on to need a kidney transplant. BBS16 is significant in this respect with every patient who has BBS16 experiencing kidney failure and requiring a transplant before 12 years of age. This knowledge is critical for future healthcare planning.

Bringing his presentation to a close, showing a photo of a group of researchers from around the world including the US, UK, Europe and Japan, Dr Haws said, "the future really is bright for Bardet-Biedl syndrome… …we can work together, to find answers, to make a difference in the lives of individuals with BBS. I'm thrilled to be able to share with you just a little bit about what we're doing here in the States. I think there's so much more, and we have wonderful opportunities to do that together."

BBS Clinics Service

Tonia Hymers, Service Manager, BBS UK

The multi-disciplinary BBS clinics service was set up in 2010 across four NHS Trusts, in London and Birmingham. BBS UK have been integrated into the service from the beginning with a third-sector contract with NHS England, to provide patient support and clinic facilitation services. In a typical year, the BBS UK team would expect to meet around 360 patients across 50 clinics and that might be face-to-face or via telemedicine. The team has two wonderful patient liaison officers, Angela Scudder and Amy Clapp, who have an in-depth understanding of BBS and its implications for those diagnosed and their families. At the time of going to print, we are very fortunate to have been joined by two excellent assistant patient liaison officers, Shirin Memi and Laura Davis who are already proving themselves to be invaluable members of the BBS UK Support Team.

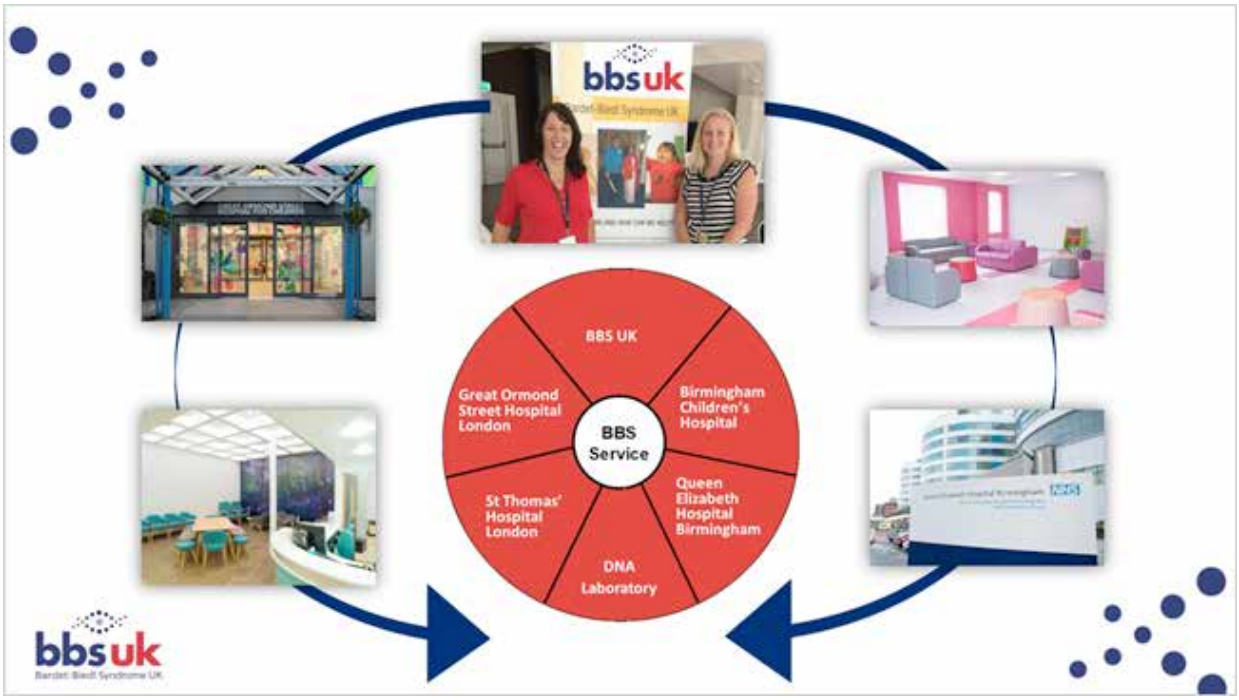
The exceptional level of service provided is holistic and aims to ensure the patient and their family feel supported to attend their clinic appointment. It can be a very stressful time for families especially newly diagnosed and our adult patients will most likely be severely visually impaired and possibly with additional mobility issues and potentially extensive health issues, so our aim is to make the process as easy as possible and help them make the most of their appointment. For example, we will liaise between patient and hospital to book the appointment, organise patient transport or hotel accommodation, and ensure the clinics team are aware of any particular issues that need to be discussed. On the day of the appointment, we may manage the clinic appointments, provide refreshments and general support and information, and spend time with each patient

to determine whether there is any additional support needed, for example with any local authority referrals and benefit applications. We can also organise travel cost reimbursement. Afterwards we gather feedback and report back to the service to support ongoing improvements.

To understand the importance and benefit of the clinics, it helps to consider the patient journey from diagnosis. Rare disease patients face an uncertain and unpredictable journey often involving multiple referrals and an endless array of tests and, once a diagnosis is made with a rare disease like BBS, there will be multiple appointments to manage which can be overwhelming and put a significant strain on the family and their resources. The lack of understanding at a local level can leave patients and their families isolated and struggling to cope and whole family support is needed. Once diagnosed with BBS, patients should be referred to the highly specialised service and from this point they will have contact with one of the BBS UK support team and a highly experienced clinical nurse specialist. They will be given an information pack of leaflets and will be directed to the BBS UK website which has a dedicated clinic section full of information and tips to help them prepare and make the most of their appointment. At each clinic, patients will see approximately six clinicians, including a geneticist, psychologist and endocrinologist and will have an appointment with each of them of around 20 to 25 minutes. Each clinic includes a multidisciplinary team meeting where any issues are discussed as a team, and the various aspects of the syndrome are considered together, which is so important. The majority of our clinicians will have seen many BBS patients and have an excellent understanding of the syndrome as a whole, which is incredibly reassuring for those attending; rare disease patients, including those with BBS, become quite used to having their condition 'Googled' by doctors in front of them.

Looking ahead BBS UK are concerned about the financial impact of Covid-19 and how it might affect the local support services that are essential to the BBS community and we are looking at what we can do to better support our membership. The BBS clinics have vastly improved understanding and knowledge of the syndrome and longer term, we are hopeful that this increased knowledge and research will soon lead to the treatments, therapies, and technologies to benefit our community.

Considering the year past and the impact of Covid on the clinics service, the introduction in 2019 of telemedicine as an appointment option to run alongside the face-to-face service has been invaluable, enabling the service to continue supporting patients during the pandemic. BBS UK have contributed to NHS England service review meetings and have held focus groups to ensure that patient voice is heard and wider learning can be had. This data has been fed back to the commissioning team and contributed to the development of a paper that aims to provide a toolkit of proposed principles and processes, representing a summary of



what good clinical care looks like for highly specialised services to follow as they develop their models of care, combining traditional and digital care services.

An obvious downside to the telemedicine service has been the lack of testing and although blood tests can be organised locally, getting this done with results ready for the appointments is not always straightforward. NHS England listened to the concerns of the different services and provided funding for home testing equipment including blood pressure machines, talking bathroom scales, a stadiometer and finger prick blood tests, which will be of enormous benefit to the patients and service once it is fully rolled out.

We are all aware of the impact the last two years has had on the NHS and our service has been affected with extended waiting times between appointments. Everyone is working hard to catch up and we can all do our bit to help by making sure we attend appointments when called or, if we need to cancel, to do so as soon as possible so that the appointment does not go to waste.

The team are very much looking forward to catching up with you all over the coming months.

For information and contact details go to www.bbsuk.org.uk.

Person Characteristics and Well Being in BBS: *Interim Results*

Dr Jane Waite
Aston University

Dr.Jane Waite is a lecturer in psychology at Aston University and co-director for the Cerebra Network for Neurodevelopmental Disorders. The Cerebra Network is interested in behavior, cognition, and emotion in people with rare genetic syndromes and over the years has profiled behaviour, cognition and emotion in 23 genetic syndromes and over 2000 individuals with those rare syndromes.

Dr Waite's talk focused on the preliminary results from ongoing research looking at person characteristics and wellbeing in Bardet-Biedl syndrome and she began by acknowledging the contributions made by many students, research assistants and clinical psychology trainees that have contributed to the research over two years as well as recognizing wider collaboration from the Queen Elizabeth Hospital, Birmingham, Anne Marie Walker, Dr Lukas Foggensteiner and BBS UK.

The Aston University site specifically focuses on the factors that affect mental health outcomes and wellbeing of people with genetic syndromes. By comparing these groups, researchers can map how changes at a genetic level can impact on development, patterns of behaviour and cognitive and emotional characteristics; identifying these can ultimately lead to improved support and clinical services.

Dr Waite clarified that she wasn't suggesting that everybody that has BBS will have certain characteristics, but that there might be a heightened risk. She also stressed that if a person has BBS, it's not a case of "There's nothing we can do about these difficulties people are experiencing, they're just part of the syndrome", understanding these pathways, enables improved intervention and support.

Dr Waite's research began with a literature review in 2018 which found a number of characteristics being described in people with BBS, including a strong preference for routines, repetitive obsessional behaviours, difficulties during social interactions, low mood, anxiety, emotional regulation difficulties, and behaviors associated with eating.

The team attended a BBS UK weekend conference and spoke to parents and to people with BBS to find out what was important to them. Adults with BBS reported that low mood, anxiety and wellbeing were important factors for them. Parents of younger children, spoke more about the preference for routines, the repetitive obsessional behaviors and difficulties with social interactions.

Dr Waite introduced some preliminary data from a small subset of participants, however, stressed that being a small subset, the data may not be representative of everybody with BBS. Dr Waite also cautioned that the results are largely focused on difficulties, and it should be understood that people with BBS do lead rich, fulfilling and meaningful lives; the results should be understood within the context that this is only a small part of the experience of living with the syndrome.

The team sent 90 questionnaire packs to parents of children under 16 years old and had 17 packs returned, a 19% response rate. The mean age of the children was 10 years old. The children were matched for adaptive ability, age, and gender, to a group of typically developing children without a diagnosis of BBS, and children with an autism diagnosis.

Around 67% of the children in the early sub-group met the cut-off for a broad autism spectrum disorder. Looking at repetitive behaviours, the team saw high levels of insistence, and sameness, and repetitive speech in people with BBS, comparable to autism, and higher than the typically-developing group. Considering activity levels, the team found no differences in overactivity, however for impulsivity, the BBS group matched those in the autism group and both groups were higher than the typically developing group. Moving on to look at mood, interest, and pleasure, they found that the BBS group were significantly lower on mood, interest, and pleasure, which is something that needs to be followed-up to truly understand what is driving that lower mood.

Moving on to talk about some of the over-16s data, Dr Waite explained that again, they have small numbers at the moment, 18 participants with a mean age of 39. Most of the participants have BBS1, a very able group of individuals who were able to self-report about vision, autism characteristics, health difficulties, intolerance of uncertainty, executive function difficulties, and anxiety and depression.

The team found that five participants met the cut-off for clinically-significant anxiety, and four for depression, which is higher than would be expected in the general population. The team found that the less the individual could tolerate uncertainty, the more difficulty they had with planning and regulating behavior, and the more likely they were to experience anxiety. Dr Waite said that they are starting to see the beginning of a model for anxiety and depression in BBS that might provide real opportunities for intervention, particularly at an early stage, to help people manage their anxiety, or develop their ability to tolerate uncertainty. She said there are numerous interventions being developed for genetic syndromes that may be able to be transferred to the BBS group based on these findings.

Dr Waite introduced Lauren Shelley, a researcher at Aston University, to explain how people can get involved in the study.

The team are looking for more parents of children to give an hour of their time to complete a questionnaire pack about their child, either online, over the phone, or by paper copy in the post; all parents will receive a feedback report about their child.

Lauren explained that it is important that as many families as possible take part, even if they don't feel they have much to report in terms of difficulties, because it will help the team to answer the question about how many people their findings are relevant for.

The team are also looking for more adults with BBS to take part, and where possible, someone who knows them well. The adult with BBS will be asked to speak to the team over the phone about their experiences and the person that knows them well will be asked to complete a questionnaire about the person with BBS, with their consent. It is also possible for the adult with BBS to take part on their own, or for just the person who knows them well to complete the questionnaire.

Lauren explained that the findings from the questionnaires in stage one of the research project, will help to inform research questions for the second stage, which will involve an assessment day. Those who take part in the stage one questionnaires do not necessarily have to complete the assessment day in stage two, both are consented for separately.

On behalf of the research team, Lauren thanked all those with BBS and their families who have taken part, and the wider research team of collaborators, for making the research possible.

If you are interested in taking part in the study, call **0121 204 4307** and leave a message to say you are interested in the BBS project. Don't forget to include your name and number. You can also email Jane at **j.waite@aston.ac.uk**.

Helen Stirland: *A Personal Perspective*

My name's Helen Stirland. I have BBS1 and was officially diagnosed in 2011 at the age of 45. When I was born, I had an extra finger on my left hand and an extra toe on both feet. My mum wasn't even told, she found out for herself that I had those extra digits. They were removed when I was about 10 months old.

I attended mainstream school and didn't really have any problems apart from extra learning with reading at junior school. I finished school and attended a college where I did a catering course for two years. After I qualified, I got a job with the British Coal Board in their catering department.

During that time in the late 1980s, when I was in my early 20s, I attended a 'well-being day' where they noticed that I had problems with my eyes. They referred me to the opticians and then I went to Doncaster hospital who transferred me to Sheffield hospital. I underwent numerous tests throughout the day. Then I saw the consultant who said he thought I had something called LMBBS and to just carry on with my life as normal.

That was a bit concerning but I thought, well, the health professionals, they know best. I went away, carried on my life and the only main problem I had was a weight problem. I have battled with that all my life and attended various slimming groups. I went on numerous diets but nothing really seemed to be that effective.

In 1994, I finished work with the coal board and went back to college, and retrained in business administration. Again, I didn't have any problems. I was able to use computers and carry on a normal life. In 2001, I joined the prison service in an administration role. I then started to have problems with my eyes mainly at night. Seeing the opticians regularly, they said there was nothing of concern.

At that point, I was still driving and still working and not having really many concerns. Then in 2010, I had two small car accidents and that was at nighttime and during the early morning when it was dusk. I thought my eyes need some further treatment. I went back to the doctors who then referred me to hospital.

At that point, the doctor said, "Your eyesight is really bad and you can't drive anymore. We are going to suggest you register as partially sighted." That was a big blow, really upsetting to be told that I couldn't drive. I thought that's my independence gone. How am I going to get to work? There was no actual support from the hospital about what registration meant and how

it would enable me in future life.

I said to the doctor and to the nurse that I didn't want to be registered but after some consideration, I went back to see them and said I would get registered as partially sighted. At that point, the nurse was very disruptive and said there is no monetary value in being registered. That really upset me more than ever because I wasn't doing it from that point of view. I was doing it for what the future could hold.

I went then back to work. My employer told me about Access to Work which would enable me to carry on with my job, by getting to work independently. I didn't know anything about Access to Work. I went through the process and eventually was able to get taxis to and from work. I also saw the occupational health department, who put measures in place which was absolutely brilliant.

I carried on as best I could and work made all the adaptations they could. As time was going on, my eyes were getting worse and I couldn't do my role properly, so in 2016, I decided to go for ill-health retirement. I actually finished with the prison service in 2017. At that point, I thought what am I going to do? I have worked for 16 years, I have a routine, I was independent, my life, I thought, had come to an end. What was my purpose in getting out of bed in the morning because I didn't know what I was going to do?

I had support from family and friends and in 2010 I was diagnosed with BBS1. When I found out about the BBS UK support group, I attended conference in 2011 and I have attended ever since and found that really useful and made very good friends and found so much support.

Like I said, I finished work in 2017 and I then found a partially sighted society in Doncaster through my sensory department team who were helping me with long-cane training. I didn't like it at first, I felt very nervous and apprehensive about using it. Now I use it all the time and it's like my friend. I can't go anywhere without it.

I live independently as best I can, but I accept now that I can't do things I used to do. Again, that has been very hard and very difficult but with the help of the Sensory Team and family and friends, I have found that I can carry on. There are things that I can do. Talking to people, you find out lots of things. You don't always get the information via your doctors or other organisations, but you do from friends and especially BBS UK support. I would recommend that if anybody hasn't attended conference personally, face to face, it is a brilliant opportunity to connect with BBS people. You get a lot out of it by making new friends, connecting with old friends, and getting a lot of information and support.

I now volunteer with the partially sighted society and at the museum in Doncaster, where I have become their disability officer and do fundraising. I can't thank my family and friends enough for all the support that they give me through adapting to BBS in my journey. I know that there are a lot of things that could alter with my eyesight, but I have just got to keep battling along to do the best that I can. Thank you for listening.

Social Isolation: *Anxiety and Low Mood*

Dr Anne Marie Walker
Dr Emma McGibbon

Dr Walker and Dr McGibbon are Clinical Psychologists working within the BBS clinics teams. Their presentation was about how to stay well and manage anxiety caused by the Coronavirus pandemic.

Dr Anne Marie Walker began by displaying a picture of the 'Capacity Cup', drawn by psychologist, Dr Emma Hepburn. The cup is pictured with the words 'burnout', 'exhaustion', 'irritability', 'anxiety' and so on, brimming out over the top, which captured lots of the different feelings people are having as we ease out of lockdown. For example, adapting to massive changes, never-ending exhaustion, huge uncertainty, pressure to socialise, and not wanting to go back to normal, can create a sense of feeling quite overwhelmed. Although some of us are excited about meeting up with people and making plans, some of us might be feeling daunted and struggling to see how things might improve or return to how they used to be.

The space inside the cup represents our emotional capacity and if it gets too full, it will overflow. Everything we do fills it up, including medical tests, going out, pressure to socialise, lower fitness levels, gaining weight. All these things take up space in our cup and, recognising how full our capacity cup is at any one time, can help us to manage our stress levels. As the cup fills, we have less emotional capacity to cope, which means that even the tiniest little thing can cause it to brim over.

Balance is the key, noticing when we are reaching the top so we can start to take things out of the cup. We can do this by talking to people we trust, asking for help, thinking of each day as a new day and starting each day afresh, making plans, looking out for ourselves, and finding things to do to reduce our stress levels, things that we enjoy. Taking good care of ourselves, figuring out what to fill our schedule with and watching out for exhaustion is important, because all these things will help us to manage our anxiety and stress.

Change and uncertainty can be very tiring and we've lived with that for a long time now. Some people might be feeling bored and lacking in motivation, and not know how to ease out of the isolation of the past year or so, feeling reluctant to start things that were cancelled in the first lockdown, or nervous about making any plans going forward. Others might be feeling lonely and isolated, struggling with the media presentations that everyone is mixing and hugging again. If you had COVID, you may still be experiencing the stress associated with that. Some of you might be grieving for people who have died, or from other losses such as a loss of a job opportunity or a sense of community.

Dr Walker mentioned how some people are struggling with how low they are feeling and that their mood goes up and down. She gave reassurance that this is normal because of the continuous stress we've all had over the last year or more. It doesn't help if other people tell you that you should pull yourself together; to cope, we need to take small steps, set small goals, and take our time, whilst recognising that it's hard. Sadness is a healthy emotional response to pain or loss and signals the need for care and compassion. It is important to seek help from your GP if you are feeling overwhelmed by your feelings, if they are stopping you from getting on with your life or having a big impact on those you live with.

Dr McGibbon spoke about why we have anxiety. We experience anxiety from the moment that we're born, and throughout our lives and it's an

important experience because it has helped us as a species to survive. It helps us to focus on problems that we need to fix. If you think about times in your life, maybe when you've had a deadline or there's been something that you've had to do, feeling a bit of anxiety can help you to really focus your mind and be quite productive about whatever it is that needs doing.

Unfortunately, sometimes we feel anxiety when there isn't a problem to be fixed and we can get into a cycle of worry and anxiety when it's not necessarily directed towards something in particular, or there isn't something that's a real threat to us.

Our brains really like predictability and we do tend to be more anxious in uncertain situations. When we become anxious, our bodies produce chemicals that activate the fight, flight, or freeze mode. Our breathing and heartbeat get faster so that we can get more oxygen and blood to our muscles so we are able to react. As blood is diverted away from parts of the body that aren't being used, such as our stomach, it can make us feel as though we have got butterflies. Our body shuts down functions that aren't needed to face the immediate threat. Sometimes we might not feel like eating and we might have a dry mouth that makes it difficult to swallow. Blood pumping around faster can make us feel more sweaty and make our faces feel flushed. These are all physical feelings that can arise when we are feeling anxious, and they're perfectly harmless – it is simply our body getting ready to face the threat.

Another aspect of anxiety is the thoughts that we might experience, for example, is it safe to go out? Have I gained too much weight? Have my friends forgotten me? These are all thoughts that might be triggered by anxiety and it's important to remember that these are not facts, these are our worry thoughts and it's important to just notice them and acknowledge them when they do arise.

Anxiety affects behaviour. It can make us want to withdraw so we can avoid things we are worrying about, or we can take on too much and feel overwhelmed. It can also make us feel a bit irritable, angry, or wound up by people who encourage us to do things that we perhaps don't want to. The avoidance cycle is a common thing to experience when we are anxious. Something triggers our anxiety, such as an invitation to go out, and our reaction might be to avoid going. As soon as we make that decision, we feel relief because the anxiety is gone, because the trigger has gone. That brings short-term relief but it can lead to long-term consequences because if we don't have that drive to go and accept the invitation, it can become a cycle of not acting and therefore missing out on different experiences and different opportunities.

Dr McGibbon rounded up the presentation with some key strategies that can support good mental health and wellbeing:

1. **Notice and accept our feelings**: paying attention to the present moment directly impacts on our mental wellbeing and health. Being aware of how we are feeling is very important.

2. **Reconnection**: feeling close to and valued by others is a basic human need and one that's essential to function healthily in society. We need to connect to feel less alone but we do need to plan and take it slowly and be aware of things like avoidance. The BBS UK Facebook groups are safe ways of keeping in touch with people and the BBS UK Patient Liaison Team is there to provide support. Some Personal Assistants might be able to help with getting out and socialising again. Specialist organisations such as RNIB, Mind and TogetherAll can offer help too. Look to your immediate circles of support for reconnecting as well as the wider circles.

3. **Address the avoidance cycle**: plan to do the things that make you feel afraid or worried, things that feel difficult but okay to do. This will give you a goal to work towards, but you will be taking it in very small steps, going up a ladder one step at a time. Sometimes it can be helpful to ask somebody to join you to help you face it, a friend or family member. Together you can think about how to manage some of these things that you're feeling afraid of. The key is planning your day or your week, building in a clear structure and a routine, and including things that you are going to find a bit tricky, and then also things that you know you will enjoy and will bring you some relief and calm.

4. **Coping with physical experiences you get with anxiety**: keep active, with regular exercise on your own or with someone else. Go out for walks and exercise at home, starting with just 5 or 10 minutes a day. Calming exercises and guided meditations can be helpful and the NHS has a lot of apps that can be downloaded to guide you with some calming and soothing exercises.

5. **Giving**: helping and supporting others can make us feel needed and valued and boost our self-esteem, and they can be small or big acts of kindness. Reach out through a text, email, or phone call, saying I'm thinking of you, or how are you? Linking up with other people who are probably also struggling as well can be mutually beneficial.

6. **New skills**: learning new skills is found to be beneficial in terms of bringing about good mental health and wellbeing, and lots of people have been taking up new skills and hobbies over the past year, for example, connecting with nature, taking up baking, cooking, exercising online. It's never too late to learn a new skill and all these things can be beneficial.

Gareth Owens: *A Personal Perspective*

I am the father of William, who is six, he will be seven in July. William was born with a cystic kidney, and we were told not to worry as it is not commonly associated with any other conditions, which as it turned out was not true. William was born with multiple digits on each hand and foot and had a couple of other issues as well. The biggest thing that I think was difficult for us in those first few months was a real lack of information and support in terms of what the next steps would be. When William was born, we were told the kidney cysts and digits could be genetic signifiers, that they would send off for genetic testing and we would get an appointment in about three months. That ejected us into a fairly scary first few weeks, which I just really remember as being fairly panicked and not the first few months you want to spend with your baby.

We had an appointment after about three months with the pediatrician at Kings and that was probably the most damaging part of the early journey, and a really unfortunate way of handling it, because he had not checked before the meeting, that those results had come in, he just read the results from the screen to us whilst we were there. He misinterpreted them and told us there was a microdeletion which might explain the kidney issue and the fingers and toes. He said it was all basically fine and that he would set up an appointment with the geneticists so they could 'talk us through the science.'

That was a huge burden off our shoulders. We told all our friends and family, 'It's fine,

don't worry about it.' We went to the genetic appointment, which we thought would be a fun science lesson - it wasn't.

We met with Muriel Holder, who was superb and really supportive throughout those first three years. She obviously realised through the notes that she had received that we had been misinformed. One of the first things she said to us was, "I've read the file and I'm sorry to say that I think this is the start rather than the end of our journey with William from the genetics perspective." That was, obviously, a huge thing to handle, out of the blue, really. We spent our first three years moving around three different hospitals, and probably 14 or 15 departments.

William had surgery three times in the first 18 months or so. That whole period was obviously really scary and difficult; William received his diagnosis just before his third birthday. Frankly, everything has gotten better since then, because of the support from the Great Ormond Street BBS clinic and having everything brought together within that central point.

In terms of the clinics and the support from the Great Ormond Street team, and BBS UK, it has been a lifesaver for us. We underestimated how absolutely emotionally exhausting that first clinic would be, because you are essentially confronting every single thing you have put off thinking about, or not quite wanted to address, all at once.

Going into that appointment, I was thinking of William as his own person who was battling the syndrome, which was all the negative things that he, as an otherwise would be normal kid, was having to fight off. One of the things I mentioned in our first appointment, was that he had an amazing memory. Professor Beales said, "Oh yes, that's quite common with kids with BBS."

At the time I just thought, 'Whatever door you open, BBS is behind it.' I thought that was William's thing, unique to him, his special skill, but actually, whatever direction you go in, you find yourself bumping up against the syndrome. That helped me to think of the syndrome as fundamentally woven into who William is for better and for worse. We have just got to accept it as a whole package, rather than something you have to fight, or something that you are in opposition to.

William is doing okay in school. After his diagnosis, we got an EHC plan together just before he went to school which was brutally difficult to get into place. You just have to get through all barriers of social embarrassment and just relentlessly push, call whatever number you can get your hands on, to get things moving, because whatever deadlines or whatever structure is supposed to be in place, you have to make a nuisance of yourself, in my experience, to push the process through. William is now at mainstream primary school with a fantastic teaching assistant, which really helps.

William loves maths but hates English. He likes reading but does not like thinking about how a story will progress. He is getting to the age where social interaction is getting a little bit more difficult and I can see him falling out of step with other children to a certain extent, which is really difficult as a parent. Equally, he has a robust attitude to everything that means he just does not let it get him down. He just keeps going with everything. The food side of things is a challenge as it is for everyone, but we do our best and so does he.

We are in a stage now where we are looking forwards and trying to work out where we want to locate according to what is best for him. Eyesight-wise, he is okay at the moment; we know that that is likely to change at some point, but we are enjoying it while the current situation is okay. The BBS UK conference is so important, so valuable because it shows how many people are working behind the scenes to help all of our kids and all BBS patients; it is such a great community to come together with and be a part of.

Update on Study of Wee

Professor Helen May-Simera
Institute of Molecular Physiology

Johannes Gutenberg
University of Mainz, Germany

Professor Helen May-Simera began her presentation with a basic introduction to cilia, explaining that our bodies are made up of many different cells which come in all sorts of shapes and sizes and come together to make organs and tissues. They have different roles and do different things. A lot of these cells have what looks like a little hair sticking out of the surface and these hairs are called cilia. These cilia do not function very well in BBS patients and Helen and her team are continuing the work that Professor Beales started over 20 years ago to figure out exactly what these cilia do.

The cilia receive and send out signals and send out little parcels called extracellular vesicles whose role is to help the cells communicate. These parcels are caught in the urine and can be studied using a very high-resolution microscope to see if they are different in a person who has BBS. This could explain why the kidneys in BBS patients might not work very well. The study involves isolating the parcels, taking pictures of what is inside them and looking at the presence of different proteins.

Professor May-Simera explained that it is interesting that the amount of protein in people with BBS seems to vary a lot but there haven't been enough control studies yet. The protein present can depend on how much you have eaten or had to drink, so ways need to be found to adjust the results and adjust the experiments to ensure a good normalisation method.

Urine can be stored and frozen and still be good to work with, providing valuable and useful information. An exciting result of the study of the extracellular vesicles, or 'parcels' was finding that when they are isolated and placed on a special 'reporter' cell using a dropper, they respond to treatment by glowing and emitting light, which can be measured.

Professor May-Simera explained that many hours and days are needed for data collection and she would have liked to have been able to show more results but Covid lockdowns this year have made things difficult. There is a long list of things to do next and the team will be collecting more urine and continuing their research.

Finding better controls has been one of the biggest challenges. Initially urine was collected from parents and siblings of those with BBS, but they now need age and gender-matched controls. Another focus is to try and match the type of BBS mutation to the amount of protein that is found, but there are many different BBS genes with different mutations and different proteins.

Professor May-Simera finished by thanking those who had contributed to their research to date; they will be attending the next BBS UK Conference and will collect more samples to take back to their lab. She also thanked her hard-working team, particularly Ann-Kathrin, Alina, and Viola who all helped with this study.

Emmy Anstee: *A Personal Perspective*

I'm 19 and was diagnosed with BBS when I was 6 years old. I've just finished my A-Levels, which was very strange with the pandemic that we've had. When I was first diagnosed, the professionals weren't too helpful as they only looked at the weight gain and didn't really associate any of it with the difficulty with my vision until I went to primary school, where they realised there was something actually wrong.

I have such a supportive family and we always try and look at the positives. We always say that we look at things in a different way. They're so, so good. At my main-stream primary school, I had some support from the VI team. In Year 3, I was taught cane skills and how to touch type, which was then really important throughout the rest of my years in mainstream school, because that's how I take all my notes. I was part of the choir from Year 1 to Year 6 and it always cheered me up if I was having a down day. I belonged to the chess club and also took part in the Year 6 drama production where I was able to be a narrator because they did the script in really big font.

I then went to the secondary school, still mainstream. When we were shown around, it was really light and bright and I said to mum, that was where I really wanted to go. There was good sport and many music clubs, I joined three choirs and percussion ensemble. I also did the Tenner Challenge, which is part of the Young Enterprise Challenge.

They adapted The Duke of Edinburgh's Award, so I could have the same experience as the others. Someone would check the route to ensure it was flat without too many obstacles.

I had a few issues during my GCSE year and didn't quite get the grades that I needed so I had to stay back for a retake year. I was so, so disappointed in myself at first but then once I just started that retake year, I really enjoyed the extra subjects of Level 2 Health and Social Care and Level 2 Finance as well as retake science, maths, and English. I was really pleased with my results as I managed to go up about four grade levels; I was so happy I cried with relief and happiness.

The school did a trip to New York and I was able to go. I shared a room with my best friends and we went to see *Wicked* on Broadway, which was amazing. Special seats were organised for me and my LSA (Learning Support Assistant) as the ones for the school were too far back. We were now in the front row! It's the one show that I remember seeing every single little detail in, which obviously is quite a rare thing with my vision problems. My LSA has been so good throughout my years at school and she's part of the reason why I've been able to do so many activities and clubs with the school. Actually, we've developed such a trust and I've told her that I trust her as much as I trust mum. I then did my two A-Levels, Level 3 Health and Social Care and BTEC Applied Science, which I've just finished.

During that time, I was able to teach Braille to another student at my school who has a visual impairment, which was a brilliant experience. It really helped develop my patience and confidence, it felt so positive and empowering. It was a great experience.

We recently discovered that I'm able to do trips with the VICTA charity and I did my first one to the Royal National College in Hereford. There were many activities, which tested my capabilities. We played Goalball and we did some climbing. I met a group of other young people who were vision-impaired or blind. Strangely I was the one with the most amount of vision and I was asked to guide and help other people! I'm night blind, so can't see a huge amount but with my guide cane, was guiding someone else who was blind. That was very interesting but very, very scary.

I also went on a 10-day Spanish exchange trip with them where I was again pushed to my limits. I rode a camel, which is the most terrifying thing I've ever done in my life, and probably something I'm never going to do again.

I screamed the entire way around. I also went with them to Belgium and a weekend in London. I was supposed to do a trip with them in 2020, but obviously, because of the pandemic, that was cancelled which was a shame because that was to go and see some of the Paralympics. I've also been part of Riding for the Disabled, RDA, since I was six and I've made so many friends through them. Being on a horse is the most wonderful feeling and it doesn't matter if you can see or not.

I've had so many volunteering opportunities. A garden club for the disabled; lab technicians at school and teaching braille. Hopefully, I can do more volunteering opportunities in the future. Now I've finished my A-Levels, I'm hopefully going to Morton College to study food science and nutrition.

There have been a few positives that have come out of the lock-down. I've started my YouTube channel, *Emmy's VIP Life*, which is where I explain or discuss loads of things linked to what it's like with the vision impairment. I've done nine videos now and it's really lovely to receive such positive comments. I feel as if I'm actually doing something that's helping people.

I've been to many of the BBS conferences which I've really enjoyed. It's just great to meet others, especially in the young people's group. Every time we meet, we have a good laugh. A big part of our condition for quite a few of us is the weight problem. I now have a treadmill and some other gym equipment. I hated it at first but it really improved my well-being.

Despite our condition, there are many positive things that we're able to do. Hopefully, next year, we can all meet up again and have the great experiences that we normally do every year by sharing what we've been able to do with each other.

Supporting Weight Management in BBS

Sarah Flack
Principal Dietitan, Great Ormond Street Hospital

Sarah Flack has been working as a dietitian with the GOSH BBS service since April 2010. She began her presentation with an overview of the service, starting with some feedback from parents that shows just how valued the specialist provision is:

"The dietitian was very approachable and understanding of the needs of children with BBS."

"The dietitian was knowledgeable and non-judgmental…"

"Dietitians outside of BBS, do not have a full understanding of the impact food has on our children."

"Great to discuss and brainstorm ideas with someone who actually fully understands the implications of BBS."

Sarah explained that there is a specialist BBS dietitian at each of the four centres and they see everybody attending the clinic. The dietitians in the children's service also take part in the mini telemedicine clinics that happen between the big clinic appointments. This mini clinic allows for a quick check-in about diet-related concerns, such as growth, weight gain, diet and activity. If there are concerns, the dietitian may also offer a separate telephone follow-up call. Referrals can be made to local dietitians for ongoing input between the BBS clinic appointments as the appointments are now typically two years apart, and clinic dietitians can be contacted between appointments.

Sarah explained that prior to joining the BBS clinics team, she had limited knowledge of Bardet-Biedl syndrome. BBS wasn't included in the syllabus when she was at university, and a survey of her department showed this was still the same – even for her younger colleagues, with 94% of her department having no experience of working with a family or adult with BBS before starting at GOSH.

So how do the dietitians learn? When Sarah first joined the BBS team, she learnt from her colleagues, including Professor Beales who shared lots of information, and so she was able to use this and apply her dietetic knowledge and experience within the clinic. The team have a great working relationship, from which Sarah continues to learn all the time.

Sarah has attended many BBS UK conferences over the years and has found this a useful experience. She is grateful to have had the opportunity to chat with adults and families living with the syndrome and learn about things that have worked, things that haven't worked and things they wished that all dietitians knew. Most importantly, all the BBS service dietitians have learned from the experience of working with those who have lived experience and at every clinic, advice is offered, along with different ideas that people might like to try. During follow-up visits, they find out what worked and what perhaps hasn't worked, and also about the adaptations that have been made and this information can then be shared with other families and adults living with the syndrome.

Sarah went on to share her learning with attending delegates, she said, "All dietitians need to know that adults and children with BBS have lower energy requirements, and this makes dietary management really challenging. Any advice around activity needs to be accessible and vision changes need to be taken into consideration."

Sarah shared case study data. Children attending the BBS clinic are asked to complete a food diary for three typical days which is analysed and a diet report is sent out. Sarah shared data taken from a 6 year old child's diary, which showed a really well-balanced diet, with plenty of fruit and vegetables, controlled portions at mealtimes and no sugary drinks.

Calorie intake was controlled, yet despite this, his weight was increasing rapidly. After a few diet tweaks and looking at how he could increase his activity, his weight started coming down beautifully.

Sarah's second example was a 12-year-old girl who was very active and again following a well-controlled diet. She had great portion control, having half a plate of vegetables at main meals. Her average intake was about 1,200 calories a day and with this she was able to maintain her weight in the healthy BMI range. This is an incredible achievement, but it isn't easy to do this.

We know from studies that about 72% to 86% of adults with BBS are living with obesity. There is limited data in literature about patterns of weight gained in childhood, but there's an excellent paper by Dr Jeremy Pomeroy that has been published in *Pediatric Obesity* this year which discusses weight changes in childhood.

Data has been collected since the start of the specialised BBS clinics at Great Ormond Street and Sarah shared some of that data to give a sense of hope about managing weight gain. She said, "In the first year, none of the children attending the clinic were in the healthy BMI range. Published results for the period between April 2017 and March 2020 show that, on average, 9% of patients have been able to maintain a healthy BMI, which is incredible. It is also exciting that about 60% of patients have been able to reduce their BMI between appointments. Of the seven children who had a healthy BMI during that period, four of those children achieved their healthy BMI by their fourth or fifth visit which showed the benefit of attending the clinics." This data can be accessed by searching for 'GOSH clinical outcomes BBS'.

Sarah summarised by saying that achieving and maintaining a healthy weight in BBS is very challenging, but it is possible to have some really good outcomes.
She said that the dietitians are always looking at how to improve the service they offer and always welcome feedback. Sarah highlighted that we should take every opportunity to educate other professionals about BBS outside of the specialist clinics and in particular, linking with schools can have a really positive impact on diet, as this can help expand both food range and opportunities for increased activity. More frequent monitoring of weight and height between appointments would be really helpful as big life events can have an impact, for example, children starting nursery, starting school or changing school, which all impact on what we eat and how and when we can move.

Sarah suggested that healthcare professionals could also write in Dietetics Today or publish a paper in *The Journal of Human Nutrition and Dietetics* to raise awareness of diet in BBS for dietitians outside of the clinics. Work is also being done on providing accessible dietary information with a focus on portion control, increasing fruit and vegetable intake, snacks, and information for schools. This will be available from the GOSH website soon.

Rhona Long: *A Personal Perspective*

I'm mum of eight-year-old Charlie, who, when he was 3, was diagnosed with BBS1. I'm so proud to be asked to speak about life with BBS and special thanks to all those who work at the BBS clinic and behind the scenes.

Charlie was born with extra fingers and toes which we had removed. They were described at the time as something that is found more and more frequently in newborn children. He was a little bit of a late developer, but again, common consensus was every child will catch up with walking and talking by the time they start school. We were a little bit concerned about his weight, so we took him to our then GP when we lived in London. It was explained as just puppy fat. Even though we saw early signs of what we now know is very typical of BBS children, none of this was linked together.

I think my personal BBS story started when we moved house and we again went to our local GP about Charlie's weight and were referred to a consultant in Colchester Hospital who happened to know somebody who was involved with BBS. She kept asking me questions about when he was born. What was he like when he was growing up? Were there any late development issues? The fact that she kept asking questions prompted me to tell her that he had extra digits when he was born even though that wasn't something that was connected to his weight or developmental delays to my knowledge. She still sees Charlie regularly.

For anyone who is diagnosed or for any parent whose child is diagnosed, it's initially quite a lonely time. It's very emotional and confusing and very overwhelming. I remember so distinctly, the very first phone call I had with Tonia. She just took me through some of the things that the BBS Clinic could help with and what the professionals look after and what they look for. I just remember thinking, why does this woman who I've never met, know my child so well? I don't understand how she knows exactly what challenges Charlie's got.

Then a few months later we went to our first BBS conference. For me, it was the first time I'd been anywhere with such a concentration of people who had guide dogs and carers. Apart from that initial feeling of being completely overwhelmed when I was thinking is this what Charlie's future is going to look like, I then relaxed and got into the spirit of what the BBS conference is all about. Since then, I just haven't looked back and I've got so much benefit from working with the whole team and knowing that they're just on the other end of the phone or the other end of an email just to make Charlie's life better every day.

The fact that the lovely Sarah Flack (Principal Dietitian, GOSH) is always on the phone whenever I need her and we can discuss ideas that I've got about his weight and his diet. We've invented 'Chocolate Friday' for Charlie, as he's not allowed chocolate during the week, but on a Friday, he's allowed chocolate! We also developed 'Promise milk' - Charlie hates drinking water, but he loves milk and when Sarah

advised he was drinking too much milk, we changed his routine so he could have one glass of milk a day at bed time and when drinking his milk he makes a promise that he'll go to bed, sleep all night and not get up until the morning until I go and wake him up (hence 'promise milk').

All of these things that we've introduced for Charlie were inspired or revised or helped by the support of the BBS team and the local professionals that I work with. The wonderful consultant at Colchester Hospital, our local pediatrician has also connected with the BBS team. They make it easier for him to have the support to get his blood tests done because it's not just a one-off, he has to have his blood tests all throughout his life. He's just been referred to King's College Hospital to have a close look at his liver. It just makes those referrals a lot easier. If I didn't have the team at BBS to help me, it would be much more of a struggle to get those referrals through my GP, because the GP wouldn't know that having a speech delay and polydactyly is connected. They wouldn't know that having problems with your kidneys and lack of motivation is connected. They wouldn't know that visual impairment and obesity is connected. They wouldn't know because they're not specialists in that area whereas the BBS team are specialists.

I think the one message I would give to those in the medical community is that the wealth of knowledge of those who work at the BBS Clinic and BBS Charity is so enormous for a rare and complicated condition, that anyone who comes across a patient who looks like they might have BBS, please do take advantage of the BBS professionals who live, sleep, and breathe BBS all the time. I want to compare it with my older sister, who's got Marfan Syndrome. She's now in her fifties and has quite a lot of health problems because of her condition, but if she'd had the level of knowledge and support about Marfan that I have about BBS, if she'd known all of that throughout her life, she could have put so many more actions in place to make her health so much easier, now that she's older. That's what I see for Charlie. I see that what we do now makes a huge difference for when he's older. I think that it's so important to find out as much as you can, absorb as much knowledge as you can so that you can support and help the people with BBS.

Recently when Charlie came home from school with his bookbag, everybody in his class had written a couple of words about Charlie. It was typed off in the shape of a train because Charlie loves trains. His classmates had written descriptions about Charlie - how he lets you get involved, he's got a big imagination, has nice glasses, he's a great friend to have and he respects everybody. This really summed him up. Charlie's also got a separate autism diagnosis. He says, "being autistic is like the gateway to be ingenious". This is my little eight-year-old boy. Despite all of the challenges, he's a confident, interesting, well-liked, adorable, caring, loving little boy who's got loads of interests. He's interested in designing, engineering, trains, space, and the Titanic. He's able to be those wonderful things because he's so well-supported, and from my point of view, I can support him to be his best because I feel supported and well-informed enough for me not to worry about every day going forward.

Update on Setmalenotide Study

Dr Elizabeth Forsythe

Dr Elizabeth Forsythe, a clinical geneticist from Great Ormond Street Hospital, London, began her presentation with an outline of the scientific basis for hunger, what is different in people with Bardet-Biedl syndrome and how weight-loss drug Setmelanotide works.

The hypothalamus in the brain balances body temperature, sleep cycle, and blood pressure, as well as hunger levels and appetite. It senses signals that come to the brain from all different parts of the body, weighs up what the best response is, and then sends out some other signals in the form of hormones, to tell the body how to react. Regulating hunger is a complex process with some signals increasing a person's appetite and some signals decreasing a person's appetite. These signals follow a pathway, and it is the melanocortin-4 receptor pathway which decreases appetite.

Dr Forsythe explained that the melanocortin-4 receptor pathway does not seem to work properly in BBS which disrupts the process of decreasing our appetite and feeling full. Setmelanotide works on this pathway and in people who have BBS it ensures there is a better balance in terms of the food intake and what the body needs.

Dr Forsythe reported that Setmelanotide clinical trials are taking place worldwide and there are three phases:

- Phase 1 involves a small group of people to check that the drug is safe
- Phase 2 involves a few more people to check the safety and dosage of the drug
- Phase 3 considers the effectiveness of the drug

A phase 3 study with two participants is currently underway at St Thomas' Hospital in London and at other sites across the world. The trial began in March 2020, but because of the coronavirus pandemic, it stopped and was restarted at the end of September 2020. Setmelanotide is a drug that one must draw up and inject every day into the tummy with the participants monitoring daily how hungry they feel.

To get unbiased results during the first 14 weeks, participants are randomly allocated to inject either the drug or a placebo. Following this, all participants are injecting Setmelanotide. Other centres around the world began the trial earlier and preliminary results are coming in.

A study of 31 patients (12 years or older) included 28 Bardet-Biedl syndrome patients and 3 Alström syndrome patients. The results were encouraging, showing a significant decrease in weight and hunger. Adults with BBS had an average weight loss of around 10%. Dr Forsythe explained that the weight of younger patients is calculated slightly differently to account for growth however, the effect of Setmalenotide in children was also good.

Feedback from the participants in the trial, has been encouraging, with one patient reporting, "The trial has really benefited my life as I am nowhere near as hungry as I used to be. The weight keeps coming off and when I got into a smaller size, it gave me so much confidence."

A second observed that "everyone says how healthy I look due to the lovely suntan that has appeared due to the drug. Everyone thinks I've been away, but obviously, I haven't, as I've been in lockdown and I've had to shield, so I say I've been to Dubai for a month."

Dr Forsythe explained that a known side effect of the drug is that participants have a change in skin color, presenting as slightly tanned.

Another participant acknowledged that, "Since being on the trial, I've seen a difference in my body shape. I feel much more confident. I don't feel the urge to keep visiting the fridge." However, that participant also added, "If you have little or no sight, you will need the support to administer the drug and complete the daily paperwork."

Dr Forsythe concluded that developments moving forward for those with BBS were exciting, preliminary results look positive and work is being done to develop a weekly formulation of Setmelanotide. She thanked the clinical team and the participants, particularly for the way in which they had managed the study during the pandemic.

Technology for Life

Hannah Rowlatt, RNIB

Hannah Rowlatt is a coordinator for RNIB's Technology for Life team. Technology For Life is a UK-wide service that supports blind and partially sighted people to find information about accessibility features and technology, to aid independence and improve quality of life in general.

Hannah began by describing the three core elements to the service that Technology For Life provide, beginning with their helpline that operates from Monday to Friday, 9:00 - 5:00, offering advice and support to blind and partially sighted people.

The second element is a team of volunteers, some of whom have sight loss themselves. Hannah explained that sometimes we just need another human to help troubleshoot the problem, which leads to the third element, volunteer home visits.

During the pandemic, the Technology For Life team were unable to provide their volunteer home-visiting service, however they continued supporting people remotely, troubleshooting technical issues by telephone and via remote computer access. Hannah and the team also offer training, enabling local organisations, partners, and professionals to develop their technical skills, which they can then pass on to the people they support.

Hannah shared the three most requested areas for support, the first of which was accessing the built-in accessibility features available on smartphones and tablets. She explained that almost every smartphone and tablet on the market will have built-in accessibility features, which tend to be visual changes, including font size, colour and contrast to suit the user. The magnification or zoom function allows the user to enlarge whatever is on-screen. The screen-reader function is usually called 'Screen Reader,' 'Talkback,' 'VoiceOver' on Apple products and 'Voice Assistant' on Android. This function enables the device to read aloud any text within a movable box on-screen, allowing blind or partially sighted users to access messaging services, social media, emails, and online banking, thereby improving independence.

The second popular topic that Hannah is often asked about is 'Apps.' She recommended two Apps in particular, 'Seeing AI' for Apple products or 'Google Lookout,' for Android devices. Both Apps use the camera function to identify and read aloud printed text. Both Apps can also scan barcodes, allowing the user to identify products. Hannah emphasized the usefulness of the Apps for identifying tinned foods, so that we can make sure we put baked beans on our jacket potato and not a tin of peaches!

Another useful App is 'Be My Eyes,' which allows the user to connect, via video call, with a sighted volunteer who will provide them with visual support. Hannah said the App could be used to locate something that had fallen on the floor or to check expiry dates in the fridge. The App has a specialist support section, allowing a user to place a call to an organisation, such as Microsoft or Google and to NHS Test and Trace, connecting the user to trained assistants who can provide support specifically relating to COVID-19.

The third topic Hannah covered was smart speakers such as the Amazon Echo, Google Home or Apple HomePod and their importance in supporting independence due to their voice control capabilities. Hannah recommended the Alexa App 'What's in my Fridge' that uses voice commands to manage an inventory of items in the user's fridge, with the option to record expiration dates. Hannah went on to mention that Amazon Echo devices can now call RNIB directly, using the command, "Call RNIB helpline."

Finally, Hannah focused on the Amazon Echo Show, which has screen-reading functionality due to VoiceView and a camera. Using the camera and the command 'What am I holding?' the Echo Show will perform it's 'Show and Tell,' function and identify the item being shown. Alexa will even respond with directions if the camera cannot properly identify the product such as 'rotate' or 'move up, move down' etc. Alexa will then identify the item and provide product information.

Hannah explained Technology for Life have around 400 volunteers across the UK with many who are willing to travel that extra mile to support people in more remote areas. Hannah finished by urging those who would like more information to go to the Technology for Life website, or to contact the Technology for Life team.

BBS UK: Service Overview

Tonia Hymers, BBS UK Service Manager

Over the past five years, BBS UK's organisational structure, governance and financial position have been strengthened and the core services and activities have been established and developed. The Charity has improved its branding, reviewed, and updated its resources and now provides a comprehensive range of information resources which can be accessed via the website.

BBS UK offers the following core services.

Advice Service: At the beginning of the year, BBS UK launched an advice service which supports BBS members with accessing appropriate local services across the areas of health, education, social care, and benefits.

Annual conference: This highly valued weekend event brings those living with BBS and their families/carers together with interested professionals, providing access to the latest information and research. Of equal importance is the opportunity to connect with others in the BBS community to share understanding and experiences.

Information Booklets: BBS UK has several information booklets available in print, Word, PDF, and audio. 'Introducing BBS' is also available in EasyRead, Urdu and Turkish:

- **Medical Information Booklet**: The aim of this booklet is to promote a greater understanding of the syndrome and the recommended care pathway. The information provided has been provided and checked by the specialist clinics teams, is supported by research and published articles, and is 'PIF Tick' accredited, which means it has gone through a rigorous information production process.
- **Introducing Bardet-Biedl Syndrome**: This booklet provides a summary of BBS and its key symptoms and treatments, with the aim of addressing some of the most commonly asked questions from those diagnosed with the syndrome, their parents, carers, and extended support network.
- **Booklet for Schools and Colleges**: This booklet has been developed to enable those working with children and young people in a learning environment to better support them to reach their full potential. The booklet was written with input from, and was reviewed by, members of the BBS Young Peoples' Group, their parents, and carers and in collaboration with medical and educational professionals who have experience of BBS.

- **My Life, My Future!** This booklet has been produced to support young people from around 10 years of age onwards with managing the changes that come with growing up with BBS and transitioning to adult healthcare services. It is a great toolkit of information to help young people on the way to becoming more independent and to have a greater say about how they would like to live their lives, as they move forwards towards adulthood.

Website: The BBS UK website has been redeveloped to improve access and contains contact information, a clinics tab and a 'knowledge hub' where BBS UK information booklets and other information resources, including webinars can be found. There is also a section containing personal perspectives which provides an invaluable insight into what it means first hand to be affected by BBS, and the impact it can have on day-to-day life.

Newsletters/Conference Report: Newsletters and an annual conference report are distributed in print, email and audio CD and provide updates across the areas of research, BBS clinics, events, health, and wellbeing, fundraising and include much valued personal perspectives.

Social Media: BBS UK has a presence on Facebook, Twitter, YouTube, and LinkedIn, with Facebook in particular providing a valued and much used private forum for our UK members to connect and share information and experiences.

Across the past year we have introduced the following pilot projects:

IT Equipment Fund: In response to the increasing need for people to access services through means of digital technology, including across the areas of education, healthcare, and for reducing isolation, BBS UK have set aside a limited fund to provide practical and financial support to help with the purchase of devices such as laptops, tablets, and computers. Several individuals have now benefitted from this support.

Slimming World on Referral: BBS UK has teamed up with Slimming World to offer 12 weeks free membership for those wishing to lose weight. A limited number of vouchers are still available.

Counselling Project: We have teamed up with The Maypole Project to offer counselling for young people who are struggling with social isolation/anxiety. There are limited spaces in this pilot project and places are accessed via the clinics team,

We are now looking ahead at the next few years and the resources and services that are going to be most needed. To support this, the Charity has undertaken a membership audit to assess and identify how BBS affects those diagnosed and impacts on their quality of life. The audit also aimed to establish how well those affected have been supported by BBS UK, to understand what the gaps are in the service provided and therefore how we should best direct our resources in the future. The data has already been useful in the early development stages of the Charity's Five-Year Plan including the development of its Vision, Mission and Values:

BBS UK Vision: Our unique experience, knowledge and understanding of Bardet-Biedl syndrome will ensure that everyone who accesses our services is supported, connected, and informed about BBS and empowered to achieve the best possible outcome.

BBS UK Mission: We are committed to promoting positive health and well-being within the BBS community; to improving knowledge and understanding amongst those affected, the medical and educational professionals and the wider public, and to supporting research and raising awareness of Bardet-Biedl syndrome.

Our Values: We have identified and established five key values that are at the heart of BBS UK and have been since it was founded more than 30 years ago:

Committed and Passionate: We are committed to achieving the best possible outcomes for our community and are passionate about improving the lives of those affected.

Inclusive and Community Minded: We value diversity and we strive to ensure opportunities are available and open to all.

Experienced and Understanding: We have deep experience and understanding of BBS and the impact it has on our community. We understand the needs of our membership.

Positive and Forward-Looking: We take a positive attitude towards challenges and we strive forwards with optimism and open to opportunity.

Collaborative: The voice of our membership is at the heart of our work, we seek out and listen to their experiences and the expertise of others and we share best practice.

These values remain fundamental to the work of BBS UK and will underpin all future developments of the Charity.

The membership audit is closed and is being analysed and summarised and it cannot be emphasised enough how important it is to us to understand directly from you, our members, how we can best direct our resources and services. From what we have learned so far and taking a broad perspective, we expect to focus on the following:

- ✓ Develop and grow our advice service and support provision
- ✓ Improve communication with our membership
- ✓ Develop a network of volunteers to provide peer support

- ✓ Develop communities including regional groups and online social groups
- ✓ Improve and develop our transition provision for young people
- ✓ Continue to strengthen the governance of BBS UK with a focus on improving diversity
- ✓ Develop a 'Professionals Supporting BBS UK' group bringing together a network of people who have the skills to support BBS UK with its work
- ✓ Seek more opportunities at a national and international level to improve awareness and understanding of BBS
- ✓ Continue to support research and understanding of BBS

The words that really stand out in our development plan is 'together,' 'support' and 'community.' Team BBS UK is made up of trustees, staff, volunteers, members, supportive professionals, fundraisers, donors, campaigners, supporting charities and organisations, and that is everyone who attended the BBS UK online Conference and who are reading this report. We can only make this happen if we all work together and your support is vital! Volunteering can take many forms, it can be fundraising, you can become a Friend of BBS UK, raise awareness locally, support the development of regional groups, support activities and events, or sign up as a 'Professional Supporting BBS UK;' if any of this sounds like something you could help with, please get in touch - our contact details can be found on our website www.bbsuk.org.uk or email tonia.hymers@bbsuk.org.uk.

Remembering Conferences Past

Catharine Auty: *A Personal Perspective*

I'm Catharine, I am 34 years old and I was diagnosed with BBS1 in March 2017.

I was born with two extra digits, one on my right hand and one on my right foot, and skin tags on the left-hand side of my body. These were removed at 18 months old. However, I still have an extra bone which makes buying shoes very difficult.

I went to mainstream schools and loved every minute of doing all the after-school activities; ballet, tap, swimming lessons, and Brownies. At about six years old, we visited the National Railway Museum in York and left when it was dark, and I missed a step. That was the next clue! At eight years old, I was diagnosed with dyspraxia or DCD, as it's now called. At 16 I went to sixth form and whilst there I fell down some steps of varying depths and broke my right ankle. To be honest, I didn't really enjoy sixth form, I think I was more interested in my part-time job as a checkout assistant at Morrison's. I left at 17 and went to York college to do nursery nursing, which I absolutely loved, and qualified as a nursery nurse two years later.

In November 2006, at the age of 19, I decided to have a gap year. I flew to India on my own from Leeds Bradford Airport, and stayed at a World Guiding Centre called Sangam in Pune. I spent two weeks there planning and running a camp for underprivileged children.
I came back home but didn't really settle. In the March 2007, the day before my 20th birthday, I flew out to Brazil with a gap year company called Quest Overseas. We lived in a Favella, learned Capoeira and to speak Portuguese and had plenty of activities. From there we moved on to Cairu, which is a little island off Salvador. For four weeks I did lots of volunteering work where we planted trees in the rainforest, helped to build a wildlife centre and taught English.

We were then supposed to have six weeks travelling around Brazil. I spent the first week learning to surf which wasn't easy, because of the dyspraxia. The second week, we went trekking in La Chapada Diamantina, which translates to the National Park of Diamonds. Whilst there we climbed about 200 feet down the mountain and I caught my foot in a crevice

and broke my other ankle! I had to come home early, and the only way that they would let me fly home was in first class, to stop the risk of DVT. After recovering, I started working as a nursery nurse with two- to four-year-olds. That was in October of 2007. The same week that I started my full-time job, I was chosen to go on a selection weekend for a project called GOLD which stands for Guiding Overseas Linked with Development.

The following January I was selected by Girl Guiding to go to Honduras for a month, but I only had six months to raise £2500, as well as working full time, but I managed it. I spent four weeks in Honduras in the August. We spent three weeks working with Guides and Guide Leaders out there, teaching them about HIV, AIDS, teenage pregnancy, and sexual health. At the end, we had a week off, so decided to go to a little island called Utila to learn to dive, snorkel and see the dolphins. Hurricane George hit while we were there, we were safe, but our washing wasn't!

In April 2010, I went to Morocco to celebrate 100 years of Guiding and we did lots of volunteering work. We started in Marrakesh doing cultural things and then moved to the Atlas Mountains and picked litter, and did crafts with the local children. At the end we went trekking on camels into the Sahara desert and slept under the stars. When we were due to come home we found out that the Icelandic volcano had erupted and

lots of flights were cancelled. We managed to fly from Morocco to Madrid and then were told to get to Santander overnight, from where we came home on a navy ship.

Next a work colleague noticed I was not seeing things in front of me. I realised my sight had worsened. A visit to the optician confirmed there were black spots at the back of my eyes.

During the next eighteen months I had all sorts of tests on my eyes. Eventually, my consultant, who had done quite a bit of research, thought it might be BBS and sent me to a geneticist. However, the geneticist, told me my fingers were too long and I was too tall to have BBS! On Christmas Eve of 2011, I received a letter inviting me to the BBS clinic, but at the time my grandma was very poorly, and I was looking for a full-time job, so I decided to take a break from investigations.

It wasn't until February 2016 that I decided to go back to see my consultant, who registered me as partially sighted. In October 2016, I went to my first BBS clinic appointment in Birmingham and in March 2017, just after my 30th birthday, I received the genetic test result to confirm that I had BBS1.

Eventually, I decided to do cane training which I didn't like, in fact, I hated it, but it was something I needed. In 2018, after having some major surgery, I realised there was a big decline in my day vision. On visiting my ophthalmologist in April 2019, I was registered as severely sight impaired or blind, which was a massive shock. I had only just spent the weekend at the BBS conference making lots of friends, and then on Tuesday, found out that I was being registered blind. It took me quite a long time to get used to the idea, and I realised I needed something to look forward to; I booked a trip to Vietnam and spent six months learning how to travel independently using train passenger assistance.

On the 7th of March 2020, I flew to Vietnam. My friends thought I was mad as everyone was panicking about Covid-19. We flew from Heathrow to Dubai, and then on to Hanoi. We were one day ahead of COVID closures and international flights into the country being stopped. I had an amazing two weeks, from homesteads and pottery-making and rickshaw rides in Hanoi, to climbing up steps in a cliff face to get to a beach and making spring rolls on the top deck of the cruise ship in the dark while cruising around Hạlong Bay.

We looked at citadels and went around herb gardens and actually planted herbs .For lunch, we made pancakes and flambéed them in Huế. On my birthday, we learnt how to fish in traditional Thai boats with bamboo sticks and ate at a floating restaurant and had the weirdest birthday cake ever in Hội An. Other highlights were going down the Củ Chi tunnels, looking at the War Museum and Agent Orange, and getting clothes made in Ho Chi Minh.

And then there was Covid. We left Hanoi to find hotels had been shut for quarantine, we were the last cruise allowed into Hạlong Bay and as we left Huế were told that lots of things were being closed down, and we would just have to go with the flow as regards to the itinerary. We had spent the night celebrating my birthday and then on the 14th of March 2020, we were basically told we had 24 hours to get masks, or we could be thrown into quarantine even if we didn't have symptoms.

We actually carried on with the holiday and spent another week in Vietnam! Their lockdown was very different to England's to begin with as everything was still open to an extent. However masks were compulsory and temperatures monitored as we entered any building, which was interesting in temperatures of 40 degrees! The journey back home took 29 hours and the UK was extremely different. It was like the country had gone mad in two weeks and when we came back through Heathrow, some people were in HAZMAT suits. My parents met me as they didn't want me to do three journeys through assisted travel as trains were being cancelled and Covid was rife.

I suppose, to summarise. I might have BBS1, I might be registered blind, but at the end of the day I can still do whatever I want to and live life to the full.

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