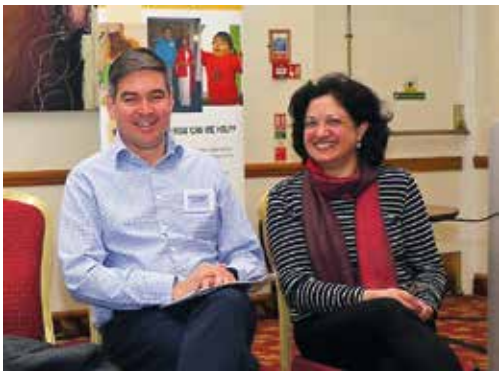
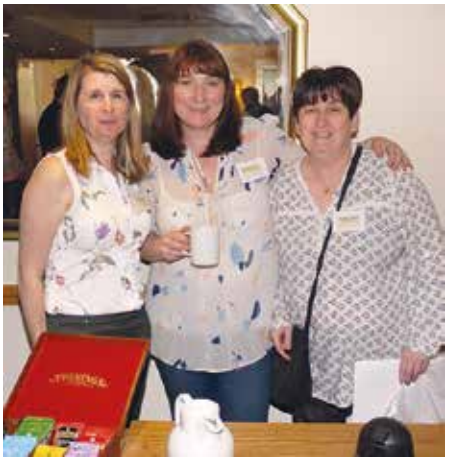
BBS UK
Conference Report 2017

Bardet-Biedl Syndrome UK

Registered Charity No. 1027384 and SCO41839





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This Conference Report contains edited excerpts of the talks and presentations from the BBS UK Conference 2017.
If you would like to listen to them all in full, go to www.youtube.com and enter 'BBS UK' in the channel search box.

Foreword

This year was BBS UK's 25th Annual Family Conference and with approximately 250 guests in attendance, it was another very successful event.

This very special weekend is what it is thanks to the commitment and passion of long-term Conference Co-ordinator, Chris Humphreys, who so very sadly passed away in June this year. Chris was the heart and soul of BBS UK for over 20 years; she gave the Charity and its members her absolute all, despite and throughout her own significant and life changing illness. Chris was the voice on the end of the helpline and was the first contact for many of our newly diagnosed families; her compassion, understanding and support gave our families the strength and courage to face their new, uncertain future, secure in the knowledge that they were not alone. Her capacity for love and interest in people was infinite, everyone she supported was important to her and they knew that. Chris retired from BBS UK two years ago, but she remained in contact with many of our families and her support continued. Chris was our families' biggest cheerleader, the support she gave was so empowering and we all benefited so much from her experience, wisdom and encouragement, my goodness she will be missed. This Conference Report is dedicated to Chris, our love and best wishes go to Phil and all the family; a full obituary will be included in the next newsletter.

Attending Conference for the first time can be daunting, but whether you are attending your first conference or your 20th, you all have so much to give each other in terms of information, support and friendship and the Hilton Hotel, Northampton, provides the perfect location for all our members, first timers or regulars alike, to make the most of this very special once a year opportunity. Many guests travelled long distances to attend the Conference, including from Ireland, Scotland, Wales and Portugal. A special mention must go to Tom, Bev and Megan, who travelled from New South Wales in Australia.

We enjoyed several very different presentations during the morning and a mix of workshops during the afternoon. We really are very blessed to be able to offer such a relevant and packed programme to our members and are extremely fortunate that so many professionals are willing to give up their weekends for us. Attending this weekend develops our learning and understanding of Bardet-Biedl Syndrome and makes us all better self-advocates, and the hope that we gain from learning about research is invaluable. On behalf of BBS UK and all of our families, I would like to take the opportunity to say a heartfelt thank you to all of the professionals who so willingly gave up their weekend time.

While the adults enjoyed their day of learning and networking, the children and young adults had a fabulous day at Drayton Manor Theme Park or bowling, while the little ones had fun in the Conference Crèche. This wouldn't be possible without our generous volunteers, many of whom return year after year, we really are so very grateful. Finally, thanks also to Becky and her team at the Hilton, Northampton for looking after us all so well, it is lovely to return each year to such willing and friendly faces.

This report commences with the Minutes of the BBS UK Annual General Meeting 2017. The Board of Trustees are all parents of children with BBS or they have BBS themselves, and they all work voluntarily on behalf of us all. They have worked incredibly hard over the past couple of years to secure a strong future for the charity and we have a great deal to thank them for. We hope you enjoy this report, full recordings can be found on the BBS UK YouTube Channel.

Planning is underway for the BBS UK Family Conference 2018, we hope to see you there.

Tonia Hymers
On behalf of BBS UK

Annual General Meeting

The Hilton Hotel, Northampton
22nd April 2017

Welcome

BBS UK Vice-Chairman and Acting Chairman, Richard Zimbler, introduced himself and welcomed delegates to the 2017 Annual General Meeting for BBS UK. Richard invited the Trustees to introduce themselves briefly, they were:

Abbie Geeson, Secretary; Laura Dowswell, Treasurer; Emma Oates, Fundraising Coordinator; Graham Longly, Trustee; Stefan Crocker, Trustee; Dianne Hand, Trustee; Margarita Sweeney-Baird, Trustee.

Apologies

There were no apologies received from the Board of Trustees

Minutes of the 2016 AGM

The minutes of the last AGM of BBS UK, held on the 23rd of April 2016, had previously been circulated and were confirmed to be a true record of proceedings. The approval of the minutes was proposed by Dianne Hand and seconded by Stefan Crocker.

Reports and Accounts

Chairman's Report: The Chairman's report was read out on behalf of the Acting Chairman, Richard Zimbler by Abbie Geeson.

'I have been the acting Chair of BBS UK for the last six months as in November 2016 Steve Burge stepped down as Chairman. Graham Longly is also retiring as a Trustee and I would like to take the opportunity to thank Steve and Graham for all their hard work and all that they have done for the Charity over the years. I would like to wish them all the best for the future. BBS UK is hoping to have three new observers appointed by the end of this year's AGM who may come on as full Trustees next year. I would also like to thank the Trustees for all of their continued hard work and contributions to the Charity.

The 2016 BBS UK Conference was deemed a great success with new and old members attending with a wealth of information, advice and good times had by all. Since our last AGM, BBS UK has carried out many activities such as a zoo trip, the Avon Tyrrell weekend and the Duxford Christmas Party for children and their families. The Charity has also had its first Adult Social Weekend staying at the Cobden Hotel in Birmingham. We went Ten Pin Bowling and had a meal at Frankie & Benny's, the feedback from those who attended thought it was a great weekend. We have around 45 volunteers that support our projects and I would like to thank them all for their help and support throughout the year as without them our work wouldn't be possible.

The clinics are being well attended with numbers continuing to grow and networks between patients being formed. I would like to thank Julie, Tonia, Angela and all of the volunteers and clinicians at all of the hospitals for the continued smooth running of the BBS UK Clinics. BBS UK has now received our Information Standard accreditation. I would like to thank Tonia for all her hard work in getting this implemented. To date we have also implemented new BBS UK policies and procedures which are now in place.

I would also like to thank all of the members for organising fundraising events and donations throughout the year. We have also been successful in being awarded grants from both Jeans for Genes for our Information Project and The Sylvia Adams Charitable Trust for their Breaking Down Barriers Project. BBS UK is planning to hold all our children, families and adult events again in 2017 and BBS UK will continue to develop our existing and new

projects. We are also continuing to take forward the change of legal structure to become a Charitable Incorporated Organisation (CIO).'

Richard finished by wishing everyone an enjoyable Conference, saying he looked forward to meeting everyone during the weekend.

Treasurer's Report: 'Income during 2016 was £63,940 compared to £59,796 in 2015, which includes restricted grants totalling £12,000 for Jeans 4 Genes and Breaking Down Barriers projects. Expenditure during 2016 is £30,813 compared to £61,311 in 2015.

At last year's AGM, the Committee gave out the message that the Charity's finances were of huge concern and if things didn't improve then it would have been unlikely that the Charity could hold the Conference as a whole weekend event. Thankfully there was an increase in income during 2016. We received one-off donations of around £18,000 with one being a legacy of £10,000. We also had marathon runners who raised a total of £5,990. However, it is unlikely that we will receive these donations during 2017 which means that we will see a reduction in income. Without these donations, and excluding the restricted grants, the income would have been £33,940 to carry out the charities core activities.

After last year's Conference, where we highlighted the Friends Scheme, lots of you kindly donated and set up Standing Orders which have resulted in an increase in income of approximately £2,000 per year (averaged over a whole year).

Expenditure during 2016 has reduced significantly compared to 2015. The annual Conference costs have reduced by approximately £6,000 following changes to the booking process and cost cutting measures. There weren't any costs incurred for the Jeans 4 Genes project during 2016 as there had been in 2015. Other costs remain comparable and are well controlled.

A realistic and prudent budget has been set for 2017 which will enable the core activities and objectives of the Charity to be carried out, but as ever we are mindful of the uncertainty of the future financial situation. Last year we spoke about the need for the Charity to build up reserves to ensure its long- term sustainability and this need still remains, and is one of our priorities. Apart from the two grants which are for specific projects, the Charity receives no external funding and relies solely on the committed and ongoing support of its members. The Committee would like to take this opportunity to offer huge thanks and extend our appreciation to all our regular donors and those who freely give up their time to fundraise.'

Fundraising Coordinator Update: 'I am Emma Oates, the fundraising coordinator for the Charity. I want to extend a little bit on the Friends Scheme which we pushed last year and which we'd like to push again this year. The financial stability of our small Charity relies on regular donations, and we are often asked why we don't charge for membership to the Charity, which would indeed provide much needed regular income, however it has always been our policy that membership is entirely free. Several years ago we set up Friends of the LMBBS, now Friends of BBS UK, a fundraising initiative, giving those members who wish to regularly support the Charity the means to do so and we have slowly built up a small loyal band of friends. We do need more friends and at this year's Conference we're giving away a BBS goody bag to everyone who completes a standing order form. Just a few pounds per month can make such a difference to a small Charity such as ours. The 'Who Are We and How Can We Help' leaflet contains a Standing Order mandate and if you are a UK taxpayer you can tick the Gift Aid reclaim box, and that gives us an extra 25p for every pound pledged. Thanks to everybody who has put the effort into fundraising this year, we're very keen to hear of anybody who has a passion for fundraising and anybody who has any skills they can offer, grant seeking skills or anything at all. It's hard work for the Trustees, so we would really like to hear from anybody who can help us in any way whatsoever.'

Approval of the 2016 Accounts

The Trustees proposed the 2016 accounts. Voting was by a show of hands and the motion to approve was passed unanimously.

Auditors

The Trustees proposed that the Charity continue to appoint Michael Bannister, of

Fryza Bannister Financials Ltd, for the coming financial year. Voting was by a show of hands and the motion to approve was passed unanimously.

Appointment of Honorary Officers

Of the current officers:

- Richard Zimbler, Vice-Chairman has been nominated and is eligible to be elected as Chairman.
- Stefan Crocker has been nominated and is eligible to be elected as Vice-chairman.
- Laura Dowswell, Treasurer is eligible and has agreed to stand for re-election.
- Abbie Geeson, Secretary is eligible and has agreed to stand for re-election.
- Emma Oates, Fundraising Co-ordinator is eligible and has agreed to stand for re-election.

No other nominations have been received for these positions and the Honorary officers were all duly elected unopposed.

Election of Committee

Of the current Committee members:

- One half of the Trustees are required to retire annually - Graham Longly and Dianne Hand are therefore retiring during the AGM.
- Dianne Hand has agreed to stand for re-election for a second period of two years.
- Margarita Sweeney-Baird has a further year to run in her present role.

No other nominations have been received for the committee vacancies and Dianne Hand was duly re-elected unopposed.

Observers

BBS UK has four Trustee vacancies this year and during 2016 we received three requests from our members to observe on the committee for 12 months prior to being nominated to stand for election. The new observers are Christian Bolton-Edenborough, James Prendergast and Rachael Foley. We therefore still have one vacancy left. If any members are interested in joining us, and would like further information please do contact Abbie Geeson, BBS UK Secretary.

Any Other Business

In the absence of any further business the meeting was closed and members were thanked for their attendance.

BBS UK Board of Trustees 2017-2018

Chairman: Richard Zimbler

Vice-Chairman: Stefan Crocker

Secretary: Abbie Geeson

Treasurer: Laura Dowswell

Fundraising Co-ordinator: Emma Oates

Trustee: Dianne Hand

Trustee: Margarita Sweeney-Baird

Observer: Christian Bolton-Edenborough

Observer: Rachael Foley

Observer: James Prendergast

From left to right: Margarita, Dianne, Stefan, Graham, Emma, Laura, Abbie, Richard

Update on Research and Study of BBS

Professor Philip Beales
Professor of Medical and Molecular Genetics; Honorary Consultant in Clinical Genetics;
President of Bardet-Biedl Syndrome UK; Founder Member of BBS UK Medical Advisory Board.

Professor Beales is based at The Institute of Child Health/Great Ormond Street Hospital where he heads the Cilia Disorders Laboratory. Together with collaborators from Europe and North America, his group have made major advances in our understanding of the causes of Bardet-Biedl Syndrome. This includes the notion that abnormally functioning cilia (small finger-like appendages on cells) lies at the heart of BBS. The challenges ahead involve understanding how dysfunctioning cilia contribute to various syndrome aspects. Professor Beales has been medical advisor to Bardet-Biedl Syndrome UK since 1996 and was made President of the charity in 2005. In 2010, he established, with the support of BBS UK, national multi-disciplinary clinics, with a comprehensive genetic testing platform for all persons with BBS in England and Scotland. Following, is an edited account of Professor Beales' presentation at the 2017 BBS UK Family Conference.

Professor Beales began by talking about the clinical service.

'We are really blessed that NHS England funded this nationally specialised clinic back in 2010. Clinics are held at Birmingham Children's Hospital, Queen Elizabeth Hospital, Birmingham, Guy's and St. Thomas's Hospital and Great Ormond Street Hospital in London, supported by BBS UK in a pretty unique partnership. There are very few of these services around the country whereby the patient support group has a direct, third sector contract with NHS England, which is something we insisted on at the beginning and is absolutely fundamental to the way these clinics are implemented and run.

While we were developing the plans for this clinical service, genes were being discovered that were associated with or causative for Bardet-Biedl Syndrome. We wanted to move our research into the NHS, and this was our opportunity. Beth Hoskins set up the BBS genetic testing service at our Great Ormond Street lab which provides a high quality service for the 21 genes that are now associated with BBS. The four clinics see 500 patients from throughout the UK and this number is growing faster than we thought it would. We had predicted there would be about 20 new patients a year but in the last month alone we've had 17 new referrals.

To support the service we are developing a new patient management system to be used at the four clinical centres that will allow us to develop and store management plans for each individual, enabling a more proactive level of care. Clinicians input their summary of the consultation on the clinic day, results of blood tests are added and then a clinical summary letter can be printed off. The benefit of bringing all the information together is going to be more valuable over time when people have been to several clinics, because there will be notes to refer back to, as well as assessment data. Eventually patients will have access to their own information which they will be able to share with their clinicians anywhere in the world. This is just the first step and we're building other components that will come in future years, such as voice recognition software and wearable sensors.

'The Patient Will See You Now' is a book by an American physician called Eric Topol. It's about the future of medicine being in your hands. There's never been a time in the NHS or in healthcare in general, when patients have

been so empowered and BBS patients, as a group, are no exception. We need to grasp all these opportunities and make sure that the NHS listens to what you have to say.'

Professor Beales moved on to talk about genetic testing.

'We didn't identify the first gene involved in BBS until the year 2000 and then it was a slow trickle after that; we're now up to 21 genes. Approximately 692 tests have been completed at our Great Ormond Street laboratory with a diagnosis confirmed in about 80%, which is a significant advance for any genetic disease. We have also provided just under 200 carrier tests for family members for varying reasons. We've also been able to successfully implement 22 predictive prenatal tests. Mutations in the genes BBS1 and BBS10 are most common in the UK - most of you should know which one you are, and that will be very important for later on when potential treatments become available.

We continue to ensure we report our scientific findings to the international community. Back in 2012, we held our very first international scientific conference, CILIA 2012 at the Institute of Child Health in London. We didn't think many people would come because it was a new conference but well over 300 people attended and there was a waiting list. Since then we've been to Paris in 2014, Amsterdam in 2016 and the next one will be in Copenhagen in 2018.

Another highlight coming from our lab, is the work from one of our PhD students, Rosie Davies. She has been working with the skin biopsy tissue donated by BBS patients. This has been immensely beneficial in helping us to understand more about how BBS occurs, what the mechanisms of disease are, and also developing the resources that might be useful for later treatments. Rosie has been able to grow in a dish, little baby "eyeballs" called optic vesicles, sphere-like structures which contain all the elements of an eye.

Using an ultra-high powered electron microscope, Rosie has been able to take a very close look at some of the layers of the retina, which is the part of the back of the eye that receives light. After 98 days of culturing these cells, we can identify all the elements of the photoreceptors and other layers of the retina, which is absolutely amazing. Rosie has developed this further to include the development of a cone and the next step is to find out whether the cone is actually working. We are essentially turning a piece of skin into an eyeball in a dish which to my mind is science fiction that has become science fact. Eventually, we should be able to use this system to determine what is going wrong in the eye and work out why people are going blind.

Katia, another PhD student in our lab, has been working with urine samples from BBS patients. She has mastered a protocol to remove the cells from the urine and grow those cells in a dish, in the lab. This has enabled Katia to compare the cells with those from people who don't have BBS. When treated in a certain way, these cells will automatically form a spheroid. The typical spheroid for someone who doesn't have BBS has somewhere between 10 to 30 different cells. There is a cavity in the middle, and pointing into that cavity are the cilia, the little finger like projections that project from the top of the cells. When we looked at patients who have BBS, the spheroids all had a very strange appearance and most of them had hundreds of cells instead of the usual 10 to 30. This research is providing us with the resources to understand how kidney disease arises in BBS and gives us something to work with when medications and drugs become available.

The pharmaceutical industries are super interested in rare diseases, including BBS. We've been approached by lots of different companies, big and small, and so far we've had discussions with Pfizer, GSK, Roche and most recently with a Japanese company called Takeda. I have also spoken to Sanofi Genzyme in Boston, where there is a unit of seven senior scientists dedicated to BBS. They really want to come and speak to us about the potential for doing clinical trials. I don't know what medicines they have in the pipeline yet because we haven't signed the confidentiality agreement, but they're taking it seriously and they want to work with our BBS community, so I will make sure that BBS UK are part of the

negotiations if the trials are appropriate.

The area where we've been making a lot of progress is gene therapy, which involves introducing the fully functioning version of the faulty gene back into an individual, for example in the eye. Victor Hernandez has been working on this with his team in my lab. Essentially, we've been using mice that have BBS: they're overweight, they have some behavioural anxiety issues and they are visually impaired which means they are an almost perfect model for us to look at. Victor has been introducing a fully functioning BBS1 gene back into one of the eyes of the mice just after they're born. We're about ten months along now and they can still see out of the eye that's been treated whereas vision in the untreated eye has deteriorated as expected!

Because BBS affects different organs, we want to look at other parts of the body and I'm really pleased to announce we've signed a deal with a funding agency called Apollo Therapeutics. We're the first research group at UCL that has been given money to develop a gene therapy programme. Not only does this allow us to do the pre-clinical part, the animal studies, proving that they all work and that it's all safe and so on, but they've also promised us that they will look towards funding the clinical trials which is really quite promising.

Looking back at some of the talks that I've given at this conference, I've been absolutely overwhelmed by the involvement that BBS conference delegates and patients have had in these advances, it's absolutely astounding. If we go back to 1999, 109 of you took part in the first paper and one of the largest surveys to date, which helped us establish the diagnostic criteria for BBS. I'm proud to say that this many years later, the paper is still held up as the main criteria in clinics all around the world for making a diagnosis of BBS. In 2000 we found the first gene for Bardet-Biedl Syndrome and then in 2003 we found the first clue that cilia were involved in one of the families that used to come to this BBS UK Conference and we haven't looked back since. In 2004 we were able to determine that several people have hearing impairment as well. In 2005, in a study of mice with BBS, we were able to see that there were problems with the ability of mice to smell. When we went back to the clinic and asked you about that, using a scratch and sniff test, 60% of you said, yes, you had problems with the ability to smell. So that again was a major interaction. We started preliminary memory studies back in 2009 and that was the first indication there were problems. The clinics started in 2010, followed by the genetic test system in 2011 and we came to you for more samples, skin biopsies in this particular instance, in 2015. In 2016, BBS UK produced an updated and much more comprehensive version of the medical booklet. The involvement and support of BBS patients makes a huge difference to the scientific community and ultimately to the patients themselves.'

Improving Memory Through Exercise

Dr Elizabeth Forsythe
Clinical Geneticist
Founder Member of the BBS UK Medical Advisory Board

Dr Elizabeth Forsythe is a clinical geneticist who has been working in the national BBS clinics in London since their inception in 2006. Graduating from Bart's and the London Medical School in 2006, she completed general medical training in London before starting general paediatrics training and gaining her postgraduate MRCPCH qualification in paediatrics in 2010. Alongside her clinical work, Dr Forsythe is researching therapies for Bardet-Biedl Syndrome in the Beales Cilia Disorders Laboratory and is completing a PhD in Genetic Therapies for the Ciliopathies. She is also a board member of the Ciliopathy Alliance and is enthusiastic about identifying new treatments and better management for people living with ciliopathies. Following, is an edited extract of Elizabeth's presentation at the BBS UK Family Conference 2017.

'My name is Elizabeth Forsythe and I'm one of the genetics doctors based in London. I want to tell you about why I am really excited about exercise and the brain. I'm not talking about Sudokus or crosswords or anything like that... I'm talking about physical exercise. About ten years ago, a small group of people with BBS took part in a study to try and understand a bit more about the brain in BBS and whether there is a difference between people who have BBS and people who don't have BBS. Our most high definition imaging technique called MRI was used to do this. What we learnt from that study was that people with BBS, on average, have a slightly smaller and often unusually shaped hippocampus which we found really interesting.

The hippocampus is involved in our long term memory formation and also with our mood and our emotions. We are querying whether the fact that it's a bit smaller could have an effect on those functions. In the scientific and medical communities, we've always thought that actually the brain is what it is and once you are grown up, you've got the cells that you've got and that's it. But the hippocampus is quite different in that it keeps generating more cells throughout life and what we've realised over time is that this particular part of the brain is affected by our lifestyle and our environment, so we can impact on the rate of growth of these brain cells.

There's been quite a lot of research into this over the last ten to fifteen years. Researchers have looked at the hippocampus in mice and they have looked at the hippocampus in people with different conditions, not just BBS. What they have found is that when you get your pulse up really high, doing something such as aerobic exercise, running, cycling or going for a swim, the speed at which we generate these brain cells can increase.

Sonia, one of the senior researchers in Professor Beales' lab, has been working on BBS for a long time and decided to look into this. She found that mice with BBS also have a slightly smaller hippocampus. A hamster running wheel was introduced to the mice which they really loved and when she looked at their brains after all the running, Sonia found that these particular parts of the brain had become much more dense and cells had grown. To potentially be able to overcome something that is genetically predetermined, not fully, but to a certain extent, is really quite amazing.

From my perspective as a clinician, I think the

main thing that convinced me that this was important for us to take forward was speaking to BBS patients in the clinics, because it's not just us talking to you, it's also us learning from you. I remember a couple of years ago, one of the mums said to me, 'I just have to tell you that my child has started this really fantastic activity programme and what I've noticed is that she's got much better physical skills, which is brilliant, but also, she's now doing better at school and we've noticed some positive changes in her behaviour.'

At one of the next clinics, I saw another set of parents who told me about a different exercise programme but what they were reporting was a similar kind of finding. They said, 'It's not just the physical side of things that's got better, it's also concentration and behaviour and so on.' I started asking about it a little bit more actively in the clinics. I spoke to Phil Beales and to Kath Sparks and actually Phil had noticed this some years prior and so we sat down, had a chat and decided that we wanted to take this forward and do something about it and bring exercise into the picture.

In September, we started running a small pilot study to get more evidence to do this on a bigger scale and to get some more information. We've got 16 children from the clinics from Great Ormond Street Hospital. Some are in what we call our intervention group and they have a personal trainer who they see once a week and hopefully that inspires them to do more exercise. Then we've also got our control group and these people continue with their normal exercise but don't have a personal trainer. We see them once at the beginning where we do a fairly comprehensive assessment, including the MRI where we look at the hippocampus and other structures in the brain. We also do a hippocampal memory assessment, which is a game that we do on a tablet where people have to remember pairs of pictures or pairs of words. We also do a fitness test because we want to find out whether there's a change over time. After people have had the intervention, and a similar time has passed for the control group, we see them again and redo the same assessments to see if there's been a change.

Different people have started at different times and we'll be finishing in September this year so I haven't got the results yet, but obviously what we're hoping for is that we will find that the people who are doing the exercise intervention will have a slightly bigger hippocampus and also that their memory might be slightly better. We are hoping that evidence from this study will give us the best chance to get funding so we can take this forward on a bigger scale.

We would really like to have a much better idea of what memory is like for people with BBS. What we know already is that some people with BBS have really fantastic memories and some people really struggle, so we want to do more memory studies to help us go forward. Kath Sparks, our BBS Clinical Nurse Specialist, did the first test run on me, and keeping in mind that I have done a lot of exams in my time, I will let you know that Kath has told everyone that I was rubbish, which is true! But the good thing is it really got me motivated to start exercising.

Our exercise team started out as just me, Phil, Kath and Sonia but we did some brainstorming and started talking to other researchers and clinicians and all of a sudden there was a huge group of us which I think is testament to the fact that a lot of people think that this could be great.

I am now on Twitter @genedrelizabeth so do follow me.'

Memory studies were carried out at the Conference with anyone who was interested in taking part, including siblings and parents of people who have BBS because it's helpful for comparison purposes. Any delegates present who had taken part in the original MRI study were also encouraged to get involved because their results could be related back to the findings that were made ten years ago.

A Personal Perspective: Maria Dowswell

The following is an edited extract from Maria's talk at the BBS UK Family Conference 2017.

'Hello, my name is Maria and I'm 23 years old. I've been to lots of BBS conferences and today I'm here with my mum, dad and boyfriend Harry. When I was little I was clumsy and fell over a lot, I also had a lot of eye problems but the doctors weren't really sure what was going on. When I was 13 my vision deteriorated and my parents took me to Moorfields Eye Hospital where the doctors thought I had a brain tumour which really scared me. I was quickly taken for CT and MRI scans which thankfully showed I didn't have a tumour. The following year I was seen by lots of doctors who couldn't get to the bottom of the problem, until finally I was referred to genetics, which was the first time we heard about BBS; I was given a clinical diagnosis at 14.

Around this time our family moved from London to West Sussex. I spent a couple of years at school there but left with few GCSEs. When I left school we decided it would be best to go to a college that specialised in teaching people with a visual impairment, which was WESC Foundation in Exeter. Unfortunately West Sussex Council didn't want to pay for me to continue my education at WESC Foundation so I ended up spending about 18 months out of education whilst we had a court case and a tribunal, which we eventually won. During this time I got bored and became jealous of my friends who were starting college which affected my self-esteem.

In April 2010 I left home and moved to the WESC Foundation about 160 miles away to start life in residential college. I was nervous and got homesick at first but realised it was a good opportunity for me. They taught me living skills such as food shopping, washing, cooking, mobility and budgeting. In September 2010 I did an equine course which I completed in 2014 with a distinction.

After finishing my studies I moved to a transition house and became even more independent. I tried to get an equestrian job but due to the remote location it was a struggle. I managed to get a working student placement at a yard but due to the location it didn't work out.

I started volunteering at the WESC Foundation charity shop three days a week which really helped my confidence grow. I was put on the till and had a lot of responsibilities. WESC Foundation opened a new charity shop and a position came up for assistant manager, I applied and was surprised and pleased when I was given the job, as all I ever wanted was a job to earn my own money and support myself. I now work 2-3 days a week which I enjoy because I am in charge of the volunteers and I engage with customers and run the shop floor.

Although I live in supported accommodation, I get very little support and live an independent lifestyle. I enjoy going to Zumba twice a week, to the gym most days, horse riding, spending time with Harry and socialising with friends. During the past few years I have struggled massively with my mental health, and have experienced anxiety and depression. I have counselling every week to minimise this. I also have panic attacks especially when I go to hospital appointments.

At the moment my health is fairly good and my vision is stable even though I don't have much left. A few weeks ago, after ten years, Professor Beale's team, discovered my BBS gene: I have BBS 14. There are currently only two other people in the world with BBS 14. I just want to say thank you to the NHS team in London who look after my health and make me feel at ease and also thank you to my parents who take me

to all my appointments.'

Q: In your experience, what is the best advice you could give to someone diagnosed in their mid-teens?

I didn't really understand at first what it all meant. My parents kind of sat down and explained it to me and I kind of got the hang of it. But at the time when I was 14 and through my teenage years, I always wanted to know what gene it was. When I found out a few weeks ago, obviously yeah, it was a shock, but I definitely think my family had more questions than I did. It was good to know which gene it was, but personally for me and my health, it didn't change anything for me because I've lived with the symptoms since age 14.

Q: You say that you exercise regularly. Can you tell us why you exercise regularly?

I really like exercising, I really like sport. I absolutely love Zumba. If I miss Zumba I get really grumpy. It's hard work, I often come out and I'm red faced and all sweaty, not a pretty sight. It's loud music and dance routines. When I first went, because it's a group of people that are all able, I thought I'm not going to be able to do it with the visual impairment, but I can do it, I'm just as good as everyone else. I stand at the front, right in front of the teacher and he changes the routine all the time, you could learn a routine and he'll add a new bit in each week. If we change round to a certain side or change direction, he'll clap instead, so it's not all visual. He has never taught anyone visually impaired before, so for him it was a learning curve as well. And the gym, I just enjoy the gym generally, it puts me in a good mood, so yeah, I just enjoy exercise, it makes me happy.

Q: A quick question. What were some of the challenges that you faced when you were living independently?

Definitely, when you're at home, and you don't know how to use something, you turn to your mum and dad. Mum and dad weren't there, they were 160 miles away, so yeah, you've either got to find someone to help you, or try and figure it out

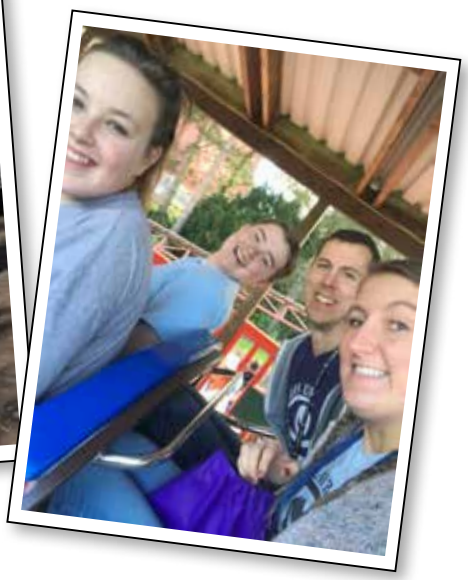
yourself. So yeah, it definitely made me grow up and mature a lot more.

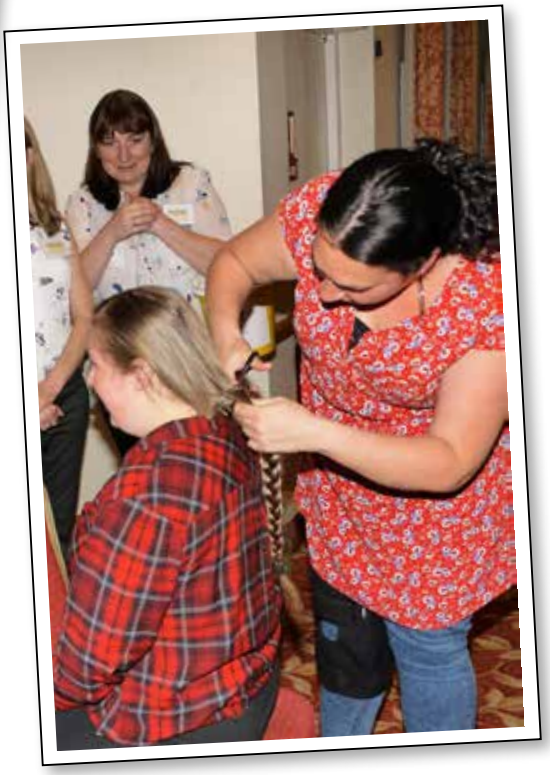
Q: I think you were saying as you've got older, you've been able to control your reactions more. Do you mean mood wise?

Kind of, I mean emotions as a teenager, as we all know, without BBS and without a disability are very hard to control. But with BBS, I don't know. I feel that like my emotions have maybe ... I've maybe matured. As a teenager, I've probably got upset or angry at the slightest little thing, and now I'm a bit calmer. Obviously with my mental health difficulties, that can be a bit hard at times, because my anxiety kicks in a lot. But generally I feel like I've found more ways to control and manage it in my own ways.

Weekend Round-Up


VANS





Wills and Trusts: Protecting Your Family's Future

Philip Warford
Chartered Legal Executive, Renaissance Legal

Philip Warford is a Chartered Legal Executive, who specialises in advising parents and carers on the legal planning aspects linked to protecting disabled and vulnerable people. He has been practising law for over 25 years and is ranked as a leading expert by prestigious publication, Chambers UK. Getting to know each and every family he works with, Philip regularly speaks at and holds legal clinics and seminars on behalf of many local and national charities including Mencap, Rett UK and Amaze. Following, is an edited extract of Philip's presentation at the BBS UK Family Conference 2017.

Philip began by outlining his presentation which aimed to cover the ways that parents and carers can protect disabled and vulnerable people and their assets through succession planning, wills, trusts, Power of Attorney (POA) and the Court of Protection.

Philip continued by listing three reasons why it wouldn't be a good idea to leave a substantial monetary gift to a child with Bardet-Biedl Syndrome. First, their means tested benefits would be compromised. Second, there is a chance that their vulnerability may be increased by inheriting a gift from their parents/grandparents. Third, there may be capacity issues and the vulnerable person may be unable to manage their finances.

Philip suggested some parents may decide to pass the gift on to their child who doesn't have BBS to manage on their other child's behalf, however this could leave the child with BBS without provision if the other child dies first and is married (where money might go to their spouse), is divorced (where money would go to their spouse) or if they fell out with the other family member (potentially lose their provision).

So what happens if there is no will? According to the law, if you do not make a will and are in a marriage or civil partnership and have children, then £250,000 worth of assets and half of the remaining assets will go to your spouse or civil partner. For example, if you have a house that's worth £500,000 and you don't make a will, your spouse will only get £375,000. The remaining £125,000 goes to your children at age 18 irrespective of vulnerability, capacity issues, or means tested benefits.

So it is important that we make a will but it is also important that we don't give anything directly to our child who has BBS – we need to organise a will and set up a trust to manage the provision.

There are three entities to a trust:

The first entity is The Trustees who will manage the trust, they can be family, friends, professional people, or a mixture and are the people that you are relying upon to do the right thing with the assets.

The second entity are The Assets. It doesn't matter what you put in the trust, largely speaking any assets can be held within a trust, cash, property and so on. Equally, whatever goes in doesn't have to stay in its current form, a property can be sold and converted into cash, or a property can be bought with cash for your child to live in.

The third entity are The Beneficiaries, the people who can benefit from the trust; you have to have everybody named in the trust who can possibly benefit.

With a Discretionary Trust, it's set up in a way that the trustees have to exercise a discretion, to take assets and give them to any of the beneficiaries as they see fit. Which means the beneficiary can safely tick the box on the

means tested claim form to say that he does not have the right to benefit from assets held in trust, because the trustees have to exercise that discretion to get assets out and to give it to him; in legal theory, they could exercise it at any moment in favour of other beneficiaries and this protects the means tested benefit.

It also protects the vulnerability and the capacity aspects because the trustees are not going to put money through the beneficiary's bank account because he may not be able to manage it, or indeed if he can manage it, it might affect his means tested benefits. But they can buy things, such as a holiday, season ticket to a football club's games, accessible equipment, property and so on. So we're protecting means tested benefits and we are providing for our child without putting anything in their name.

The problem with a Discretionary Trust is that it is taxed very badly. The government have introduced a Disabled Persons Trust, for those in receipt of DLA care component at the middle or high rate or PIP, either of the two components. The Disabled Person's Trust is exactly the same as a Discretionary Trust, it does everything that we want it to do, but it is taxed more favourably. A Discretionary Trust pays income tax at 45%, whereas the Disabled Persons Trust usually doesn't pay any income tax at all because the tax is deemed to be calculated on the disabled person and they won't have any other income.

In concluding his talk, Philip presented an example of a family that has a mum, dad, and a vulnerable adult who is 18 and has BBS. What do they need to think about? Well they need to consider making a will, setting up a Disabled Person's Trust, and then think about other issues such as mental capacity. They may need to arrange for a power of attorney if the child is 18 and has capacity, to make their lives easier. However, if their child doesn't have capacity, then they may need to think about applying to the Court of Protection to protect their child.

The Court of Protection serves to protect people who cannot make decisions for themselves. So, you only ever go to them when somebody cannot make a decision for themselves. When we look at the Mental Capacity Act and look at the presumption of capacity and about maximising capacity, if we can do that to help someone make a decision for themselves, there is no need for the Court of Protection. Where a person doesn't have capacity and we need to make decisions for them, then we might need to involve the Court of Protection.

Q: How much does it cost to set up a Trust?

The fact is that these are things that have got to be done, so you need to make sure that you get the right lawyer to do it, so it only has to be done once – make sure you ask what experience they have of a Disabled Person's Trust. If you're looking at doing a will and a trust, in the southeast, the cost could be anywhere between £1,000 and £3,000 to get everything in place. That kind of gives you a sort of ballpark figure.

Information sheets about Wills, Trusts, Power of Attorney, Court of Protection and more are available to download from the Renaissance Legal Website: www.renaissancelegal.co.uk

Understanding and managing behavioural difficulties in children and young people with complex conditions

Dr Sarah Bullen
Senior Clinical Psychologist, Great Ormond Street Hospital

Dr Sarah Bullen has a PhD in Developmental Neuropsychology and subsequently trained as a Clinical Psychologist at University College, London. She currently works within the Neurodisability and Neuropsychology services at Great Ormond Street Hospital which offer specialist assessments to children with complex neurological conditions. She has also worked in the National Centre for High Functioning Autism and in a community specialist behavioural management team in South-West London. She has extensive experience in working with children with behavioural and emotional difficulties in the context of medical or neurological conditions, intellectual disability and Autism Spectrum conditions. Following, is an edited extract from Dr Bullen's presentation at the BBS UK Family Conference 2017.

'A developmental perspective is needed when thinking about challenging behaviour as it doesn't only occur with certain conditions. How we manage our behaviour is related to the development of the brain and cognitive functions. A toddler going through the terrible twos has not developed the language skills needed to express their feelings or needs. They also do not have the social cognition needed to understand others, interpret facial expressions and tone of voice or a desire to please or be accepted by others or their peer group, characteristics which at a later stage of development have a huge effect on our behaviour.

As the child gets older, with the development of higher level executive functions, parents' expectations of their child's behaviour changes. From the teenage years these functions enable higher level goal directed behaviour, like the ability to inhibit yourself or to control your impulses. This is important as being able to plan ahead and understand the consequences of our behaviour, are functions that help us regulate our behaviour.

In the past, the term challenging behaviour was used pejoratively and put the blame on the person who was showing the behaviour. Our thinking now is to view challenging behaviour as a form of communication about an unfulfilled need or difficulty, which professionals and carers involved with a young person need to work out and unpack. There are different types of behavioural difficulties that people find challenging to manage, however it is assumed that it refers just to aggression or hitting out or having tantrums. Other types of challenging behaviour include verbal aggression, repetitive behaviour and self-injurious behaviour. Specific information about behaviour and psychological functioning in BBS is under-researched and definitely needs more work. A 2002 Great Ormond Street study on children with BBS showed that between a quarter and a half of young people who were asked, had some difficulties with behaviour or emotions. The most commonly reported problems were internalising behaviours, behaviours that a child keeps inside, like anxiety, low mood or even headaches or tummy aches that other

people cannot see. Social problems were also commonly reported: difficulties making friends or interacting with their peer group and attention difficulties like finding it hard to concentrate. Aggressive behaviour was not commonly reported in that particular group. Importantly, what this group concluded was that regardless of their level of intellectual functioning, the same behaviours were reported, indicating that the difficulties these young people were experiencing were not a function of their intellectual ability.

For the BBS child, the multiple causes or risk factors for developing behavioural difficulties can be divided into four main areas. Firstly, medical factors. A child with BBS has to cope with the distress and anxiety of their loss of vision over time and the issue of their weight management. They also have to undergo painful medical investigations and have other physical complaints associated with BBS. Multiple hospital visits mean the child misses school which can interrupt their social development and make it harder for them to fit in with their peers all of which can lead to difficulties managing behaviour.

Secondly are neurodevelopmental factors which influence the development of cognitive, motor and social skills. In BBS, intellectual and learning disabilities are the most common neurodevelopment disorders and can range from mild to severe. Social communication difficulties are frequently reported in BBS and though not everyone with social communication difficulties meets criteria for Autism Spectrum Disorder (ASD), if social difficulties are severe enough, a diagnosis of ASD may be helpful in terms of accessing appropriate support. Similarly, a diagnosis of ADHD might be given if a child has a significant and enduring problem with concentration and attention difficulties.

Thirdly are the psychological risk factors for developing behavioural difficulties specific to BBS. Problems with low self-esteem, low confidence, learning difficulties, managing their weight and their ever changing loss of vision can cause a lot of anxiety and are big things to cope with. The child's adjustment to having a condition like BBS, or any kind of chronic complex medical condition, is really important to keep in mind when we're thinking about behavioural problems. These are young people who are dealing with a whole lot more than your average person of that age. They are trying to manage all of these very complex areas of their life, while also just trying to be a typically developing young person as well.

Finally, there are ecological factors in the environment or the systems around the child which have an effect on behaviour difficulties. The medical, neurological and psychological factors are related to internal processes in the child, inside the person themselves and the ecological factor refers to factors that occur outside of the child but within their environment (e.g. relationships within family and school, wider community issues such as access to leisure facilities for children with disabilities). These four factors, internal and external are in interaction with each other and contribute to behaviour problems.

In the BBS clinic, the medical needs of children with BBS are generally well catered for, though diagnostic overshadowing can occur. This happens if a child's medical diagnosis is given priority over any additional diagnosis which results in the support for their other needs being seen as secondary or overlooked and the child's access to local emotional and communication support can then be delayed. This means that vital and important optimal times for treatment can be missed by leaving conditions untreated. Any oversight or under recognition of psychological and learning needs, particularly with children who are high functioning but with specific difficulties in certain areas that are not catered for, can aggravate psychological distress and increase possible behavioural issues developing.

Psychologists view behaviour as a communicative function and this cannot be overstated. The person is trying to tell us something with their behaviour and we need to find out what that is and what the person is gaining from the behaviour. By not understanding the function of a particular behaviour we can accidentally reinforce the difficult behaviour.

A systematic approach is used in order to understand the function of the behaviour. This approach details and assesses the different components that make up the behaviour problem and to look at the antecedents to the behaviour. What happened before the behaviour occurred? What might have been the trigger for the child, maybe they are in pain or tired or frustrated for some reason. There may also be environmental triggers. A child who doesn't like crowded spaces or bright lights is likely to have a behavioural episode in a big crowded noisy room, or any sudden change in routine can cause difficulties with a child who has social communication difficulties. So we need to question when the difficult behaviour starts and how long it lasts. Where does it normally happen? Does it happen with particular people or are there people that it never happens with? Noting where the behaviour doesn't occur is just as important as noting where it does occur.

The second part of the systematic approach is to explore the consequences of the behaviour, as the reaction the child gets to the behaviour can accidently reinforce the behaviour. What happened as a result of the behaviour? How did the adult respond? Did the response help the behaviour to go away? Did it de-escalate the situation or did it actually make things a whole lot worse? This information is then analysed using an ABC chart, which means "Antecedent Behaviour Consequence" to see what factors are coming together as triggers.

The findings of the ABC chart are used in the PBS (Positive Behaviour Support) model to implement changes in managing behavioural difficulties. The PBS model has two components. The first is the use of proactive or preventative strategies to reduce the likelihood of behavioural difficulties occurring in the first place. Here the task is to identify and modify any mismatch between the child and their environment. Challenging behaviour is often related to anxiety and the need for proactive regular calming routines and predictability. Examples of modifying the environment are the use of visual timetables, PECS (Picture Exchange Communication System), Makaton for children without verbal language, keeping tasks focused and limited to 10 minutes for children who easily become overwhelmed or find it hard to concentrate and perhaps scaffolding social interactions like turn taking to help a child with social communication with their peers.

Reactive strategies are the second part of the PBS model and are used to ensure that we don't accidentally reinforce challenging behaviour. We want to ensure that whatever we're doing calms the behaviour and reduces the likelihood of the behaviour recurring. Punishment is not used in PBS, mainly because in the long term it is not effective in modifying behaviour. Though at first in the short term there may be improvement in behaviour generally the behaviours come back stronger than before.

Reactive strategies are informally referred to as the "easier said than done strategies", nevertheless when encountering challenging behaviour it is helpful to remain calm and have a neutral facial expression and tone of voice. The underlying theory for this is that some behaviour is attention seeking and any attention whatsoever is rewarding and is best managed by responding with minimal language along with signs or visuals. Parents or teachers understandably want to explain to the child why what they did was wrong. But this is not helpful for children who are easily aroused. If a child is very anxious or emotionally aroused, the last thing they want to do is sit down and reflect on their behaviour. The reflection is best done after the child has calmed down and depending on what you think the function of the behaviour is, you might change the environment.

Distraction is also an effective reactive strategy. A change of face, change of activity, just making a joke or saying something bizarre can help diffuse the whole situation. Also allowing short breaks if you think the child is tired, but you have to ensure that they come back and finish whatever you wanted them to do, otherwise they learn that they can get out of doing things.

In order for the plan to be effective, the specific behaviours that you're trying to

change need to have the same response from everybody and consistent boundaries need to be in place across settings. It is important to ensure that all adults are aware of the child's communication needs and are working with the child in the same way, no matter where they go. There can be a situation where a speech and language therapist has done a wonderful job of teaching a child how to use PECS and nobody at the afternoon club is made aware that the child is using PECS. In this example, the child reverted back to their original form of communication, which was throwing things at people to get their attention and get their needs met.

Boundaries and discipline are often not a priority for parents of children with a medical condition or in a vulnerable state as they are understandably more concerned about managing the medical condition, but as the child returns to normal life, the more relaxed boundaries become an issue and problems can emerge. For children with additional needs, the need for consistency in boundaries is higher than with children without those needs.

The aim of the PBS approach is to develop a Positive Behaviour Support plan for the young person. As well as the proactive and reactive strategies, that plan should contain very detailed information about the child and their diagnosis and their areas of vulnerability and difficulties. It should also include their strengths and what they find rewarding and motivating and what makes them happy.

Considering the family as a whole, the psychological impact on parents of having a child with a complex condition along with behavioural difficulties is often overlooked by professionals. Parents can experience high levels of stress as they are expected to be the primary support structure for their child, and often they are not supported in their journey through adjustment and stress in relation to their child's condition.

Parental relationships can be affected by the stress of hospital visits and trying to work out what's going to happen next for their child. The parents themselves may be in different places and need time and space to adjust to the condition. Psychologists therefore need to be mindful of when is the right time to work on behaviour management with a family – it is not helpful to do this whilst parents are still in the early phases of adjustment or experiencing high levels of stress. Work may also be needed with siblings who have experienced situations that are unusual for children of their age, like a very unwell family member, or someone who's going through a particularly difficult bout of challenging behaviour. Parents are usually very mindful of the impact on siblings and it is important that professionals are as well.

A distressing factor of challenging behaviour is the impact it has on the family's access to their community. Parents of children who have severe behavioural problems often end up isolating themselves and withdrawing from their communities because of the reactions of other people. This can be either a perceived reaction or an actual negative reaction.

The clinical psychology service in the BBS clinic at Great Ormond Street Hospital aims to provide an assessment of a child's needs in relation to education, behaviour, psychological

and emotional problems and parental well-being, and signpost the family on to appropriate local services. The psychological well-being and health of the whole family needs to be considered with access to their local CAMHS, clinical psychologist or other behavioural professionals. The community paediatricians are very good for younger children and often have fantastic behaviour support groups, or run parenting support groups that can be very helpful.

It is important to have a multidisciplinary approach to managing behavioural problems. Communication is strongly encouraged between education and healthcare professionals, e.g. psychologist communicating with school. This is where the TAC (Team Around the Child) and TAF (Team Around the Family) meetings come in. Social workers can offer lots of advice and support, access to respite or activities for children with disabilities and benefits advice. School can be a focal point for accessing support, particularly if your child attends a special school and for mainstream schools, the SENCo is the number one person to go to.

In closing I've been asked how the PBS plan is communicated, especially as it seems a lot of healthcare professionals don't know they exist let alone what they contain. The questioner also wonders if there is an opportunity for a national service to provide some kind of communication card to each patient.

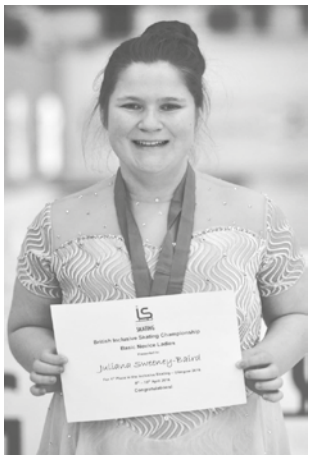
To reply, it is common that a young person has a PBS plan, that their local team is aware of, but that doesn't get translated over. It should be the responsibility of the individual psychologist or professional involved to ensure they know all of the different professionals involved with the child. In previous services I've worked in, parents were asked to provide a list of professionals involved with the child and we made a point of communicating any support plans we developed to the local professionals. As parents it's a good idea to ask the professional that you are working with to make sure that they have all the relevant contacts that they need in order to make that plan more generally accessible to everybody.'

Conference Fundraising

As you all know, fundraising is absolutely vital so that our charity is able to put on events and provide support for anyone affected by BBS. The more we can raise, the more we can do and the fundraising team would like to say a huge thank you to everyone who contributed to the fundraising efforts over the Conference Weekend. Special thanks go to Sandra Dale and Janet Wakelin for the fabulous knitted teddy raffle, Helen Petty for the Quiz prize, Ollie Sloane for the bunting and cards, Kath Sparks for the nail painting, Amy and Abbie Clapp for their brave haircuts and Judith Aylott for organising the main Raffle with its fantastic prizes, all of which were kindly donated. We are grateful to every one of you who supported these fantastic fundraising opportunities over the course of the weekend because your generosity and involvement really does make a difference. Thank you.

Dianne Hand

A Young Persons Perspective: Juliana Sweeney Baird

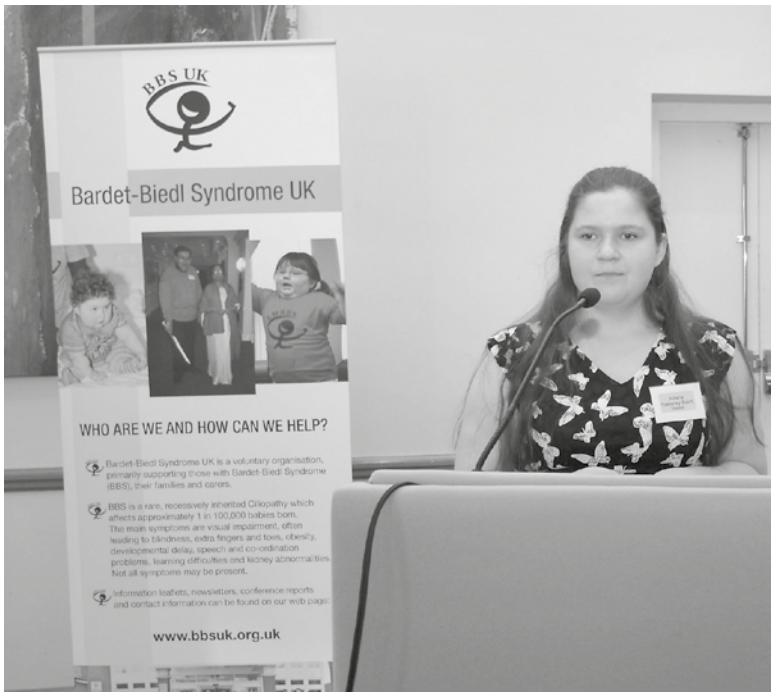


The following is an edited extract from Juliana's talk at the BBS UK Family Conference 2017.

'I am 14 and I was only diagnosed 18 months ago. Before that, when I went to appointments, I was told I was fine and even when I was at my primary school they didn't think there was anything wrong with me. That in itself was emotionally difficult for me. One of my best friends is dyslexic and so I watched her get a lot of help for a long period of time which was upsetting and frustrating for me because I was struggling too. I didn't really know how to tell anyone because I was always told that I had nothing wrong with me. Then, as I was moving on to secondary school, I was registered partially sighted and there was the whole process of getting diagnosed. I started to get a lot more help, but in a way that was hard to process because it was such a change. It was emotional because you're thinking, is there something wrong with me then? Is everything going to be fine? It's a process that was hard to deal with and in a way, I'm still kind of going through it since my diagnosis is still recent.

When I was told the news that I was diagnosed, my mum was ecstatic because before, she had been told that I had something else. Now, she knew that I was going to live and that she was going to have me around for a long time, so in her world she was happy. I was very different in my reaction. I didn't say anything and I was shocked really because I'd been told my whole life that I had nothing wrong with me. I had tears in my eyes, but I didn't cry because I was still kind of very shocked and then that was really when my life started to change. Since I went into my teenage years, my sight has gradually gotten worse and that in itself is quite a hard process to deal with because you don't know what's going to happen. Not knowing is difficult for me to deal with every day, but I've now started to get more and more help.

Because of the way my sight is now, I get out of my classes early to go from class to class because I can't cope with the amount of people in the corridors anymore. It may seem a small thing but it's made such a change to my life. My mum and I have noticed that I'm much less stressed and I'm much calmer. One of the things when I am stressed is I become very grumpy or tired.



I have been into sports since I could walk and my mum would have me doing things all the time, even when I was only a couple of months old. I would be on one of those mats and she would use any toys that were around to get me to do exercise, so I've been on an exercise programme for a long time. My mum was a skater but after my brother and sister didn't turn out to be one, she kind of lost hope. Then, when I was seven, my mum signed me up for skating sessions and I went once a week. My mum was very nervous but I got my balance and then it just kind of took off from there. I did my first skating programme when I was eight. This is my seventh year

of skating and I have won competitions for disability sport. Competing has helped me a lot and one of the things that me and my mum say is nice, is that even if I do lose my sight or my sight gets worse, I'll still be able to skate for a long time.

Another sport I play is Bridge. I have always loved cards and a new maths teacher came to my school and he's a good Bridge teacher. I've been playing that for a couple of years now and last year I went to the Scottish School of Bridge and with my team I was third there.'

Q: You must have had a few injuries skating, what's the worst injury you've had?

This may surprise you but I have never had any injury although you do get quite sore after skating, muscles wise.

Q: Does skating help you take your mind off things?

I would say in a way it does. If I've had a bad day at school, it's kind of nice just to go and do a sport that I love.

Q: Would you hope to continue skating in the future?

I'd like to take my skating career further but we'll have to wait and see where it goes.

Q: Do you get lots of coaching?

Yes, I've had the same coach for six years now and because I can't skate on my own anymore, my mum guides me on the ice and she also does a lot of coaching with me.

Understanding the mechanisms of insulin resistance and adipocyte function in subjects with metabolic syndrome

Dr Shanat Baig

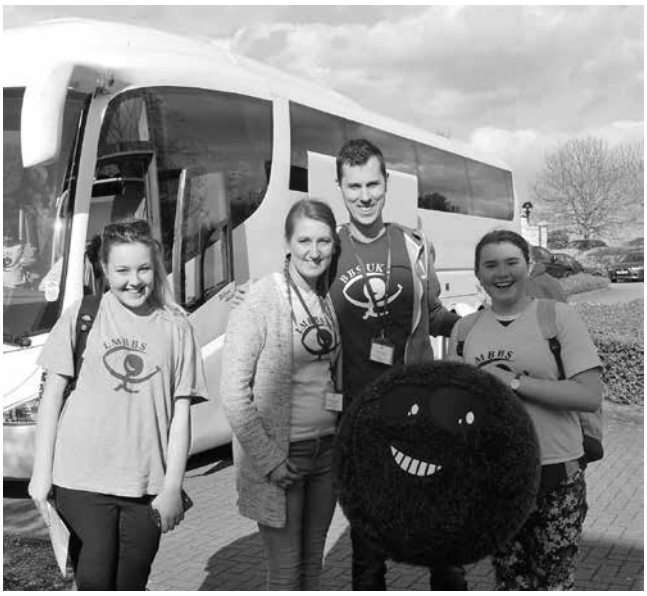
Dr Shanat Baig, a research doctor from the University Hospital, Birmingham, spoke briefly at Conference about a research study he is involved with which aims to understand more about the mechanisms of insulin resistance and obesity related complications such as diabetes, fatty liver, high blood pressure and high cholesterol. Insulin resistance is a pathological condition in which cells fail to respond normally to the hormone insulin. The body produces insulin when glucose starts to be released into the bloodstream from the digestion of carbohydrates in the diet. The level of glucose in the blood is kept within the normal range with the help of insulin mobilising glucose into liver and muscle cells. In obese people, the cells are resistant to insulin and are unable to use it effectively. Obesity is one of the key risk factors in the development of insulin resistance and Type 2 Diabetes as well as heart diseases and cancer. Another problem for people who are overweight is that their fat cells don't store fat as efficiently as they should. Despite intense research over the last two decades, the reasons for this are poorly understood. Dr Baig hopes that the results from this study, which aims to include twelve people with BBS, will lead to a better understanding of how the body handles sugar and fat. Once this information is published in journals, it is hoped that it will be used by various pharmaceutical companies to develop future treatment and management strategies.

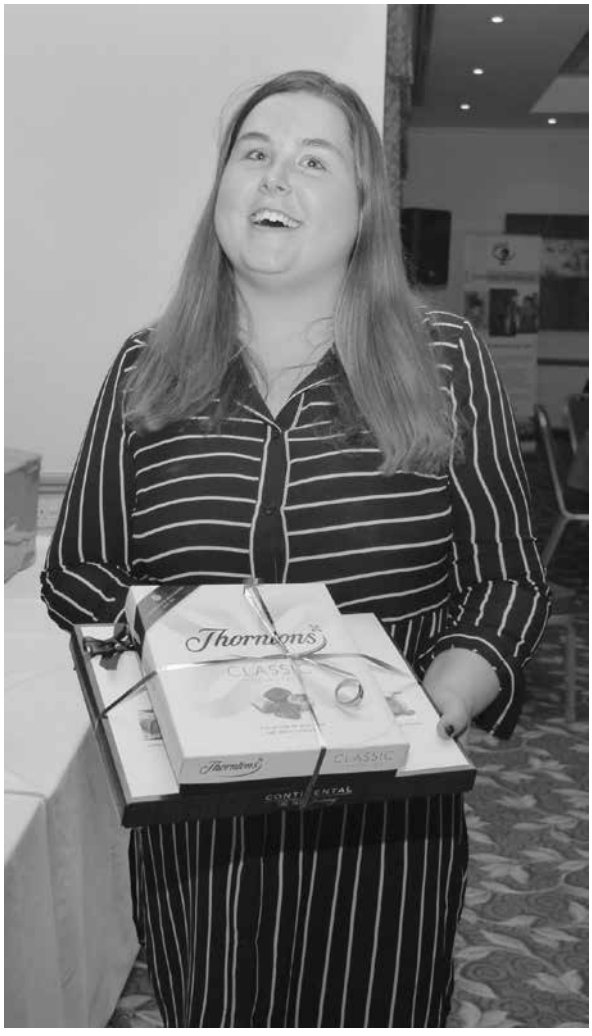
Mental Health and Well-Being in BBS

Dr Jane Waite

Dr Jane Waite, from the Cerebra Centre for Neurodevelopmental Disorders in Birmingham, spoke briefly at Conference about a collaboration with the Queen Elizabeth Hospital in Birmingham. Dr Waite is part of a psychology team that is interested in looking at

behaviour, focusing on cognition and emotion which is recognised as an under-researched area. Dr Waite was keen to use her time at the BBS UK Conference to understand what is important to those with BBS, their carers and families. The key issues that Dr Waite identified from talking to delegates were anxiety, low mood, emotional outbursts, finding uncertainty difficult, social skills and sleep. This feedback will enable Dr Waite and the research team to develop a patient focused research proposal for funding to conduct a project looking at personal characteristics, mental health and well-being in BBS.





Closing Remarks

BBS UK Secretary, Abbie Geeson, closed the morning conference programme with some words of thanks and a special 'farewell'.

'The trustees would like to say a special thank you to a few very important people. Firstly, to Chris and Phil Humphreys who are unable to attend the conference this year. Their dedication to the charity along with all those who laid the foundations and structure back in 1987 is as important to the Charity today, as it was then. Our BBS history means a lot to the medical professionals, researchers, members, patients and families alike. Without it we wouldn't have the knowledge, support and understanding that we have today and we would like to send our love and thanks to those here and those who cannot be with us for their part in shaping Bardet-Biedl Syndrome UK.

The Trustees would also like to say thank you to Julie, Tonia and Angela for their continual support to the Charity and clinics. They really do work so hard every single day and we really do appreciate them. To the team of volunteers who also support us in so many ways, you all make our projects and services what they are and are an integral part of the organisation and again we really appreciate everything you do.

We now have a very special thank you to give to another very special person. Sadly, after much thought, Julie has made the decision to leave the clinic service and Charity in September this year, to devote more time to her family, her health and hopefully some exciting new ventures; this will be Julie's last Conference as part of the team.

Julie attended her first Conference as a new delegate in 1998 and joined the Charity in 1999 where she took on various positions until retiring in 2015. Since 2010 Julie has also been the BBS UK Clinics Ltd Adult Service Manager for the specialised clinics at Guys Hospital, London, and Queen Elizabeth Hospital, Birmingham and is proud to have been part of the clinics team during its inception and growth.

I know that everyone will join the Trustees in thanking Julie for not only guiding us with her knowledge and understanding of BBS but also for the support she has given us with her love, friendship and care for the last 18 years. Julie really will be missed and we wish her all the best and thank her from the bottom of our hearts for her dedication and kindness that has impacted on our lives in the best possible way.'

On behalf of the BBS UK Board of Trustees and Members, Abbie presented Julie with a leaving gift and presented Julie, Tonia and Angela with flowers. She finished by thanking everyone for attending the 25th BBS UK Annual Family Conference.

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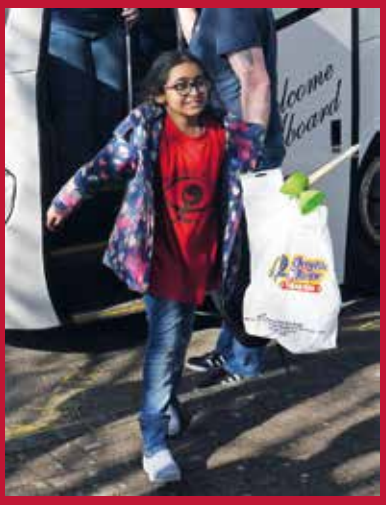


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