BBS UK
Conference Report 2019

Bardet-Biedl Syndrome UK

Registered Charity No. 1027384 and SCO41839










CONTENTS

This Conference Report contains edited excerpts of the talks and presentations from BBS UK Conference 2019. Many of the presentations are available to listen to in full: go to www.youtube.com and enter 'BBS UK' in the channel search box.

Foreword

Welcome to the BBS UK Conference Report 2019, which encapsulates and highlights the successful Family Conference Weekend. This report neatly summarises the conference programme, providing a timely recap for those who enjoyed the weekend and to inform and inspire those who were unable to attend this year.

Regular attendees anticipate this fantastic event with great joy and the Board of Trustees are rightly very proud of this special weekend. The theme, which was again 'Health and Well-being', was planned to reflect a key aim of the Charity, which is to improve knowledge of BBS and to maximise the lives of those affected by the Syndrome.

British Blind Sport 'taster sessions' were available across the weekend to provide a flavour and experience of sport for those who are visually impaired. We were delighted to debut the first BBS Zumba Class, facilitated by Maria Dowswell. There was an informative presentation by Slimming World and inspiring personal perspectives from Tom Oates and Rachael Foley.

Professor Phil Beales once again brought us up to date with research news and we welcomed Dr Murray Stewart from Rhythm Pharmaceuticals who delivered a presentation about a promising new weight management drug.

The Committee would like to thank all the professionals who gave up their valuable weekend time and continue to support BBS UK and explore our complex syndrome with such passion. Edited extracts of all the presentations can be found in the pages of this report. Full audio recordings can be accessed via the BBS UK YouTube channel.

Planning has already begun for Conference 2020 which will take place across the weekend of 17th-19th April – we hope to see you there.

Stefan Crocker
Chairman, BBS UK

Annual General Meeting

The Hilton Hotel, Northampton
27th April 2019

Welcome

BBS UK Chairman, Richard Zimbler, introduced himself and welcomed delegates to the 2019 Annual General Meeting for Bardet-Biedl Syndrome UK (BBS UK). Richard invited the Trustees to introduce themselves, they were:

Abbie Geeson, Secretary
Laura Dowswell, Treasurer
Dianne Hand, Trustee
Emma Oates, Trustee
Rachael Foley, Trustee
Christian Bolton-Edenborough, Trustee
Judith Aylott, Observer
Stefan Crocker, Vice-Chair

Apologies

There were no apologies received from the Board of Trustees.

Minutes of the 2018 AGM

The Minutes of the last AGM of BBS UK, held on 21st April 2018, had previously been circulated and were confirmed to be a true record of proceedings. The approval of the Minutes was proposed by Stefan Crocker and seconded by Dianne Hand.

Matters Arising

Voting procedure was explained: families with one or more children with BBS are entitled to two votes, adults with BBS are entitled to one vote, professionals/ representatives from another organisation and non-member delegates are not entitled to vote.

Reports and Accounts

Chairman's Report: Richard Zimbler

The Chairman's report was read out by Dianne Hand on behalf of the Chairman, Richard Zimbler.

"The Charity has had another busy year planning, organising, and supporting our various projects and events and in raising awareness of our Syndrome. The Charity continues to develop, and the Trustees are very proud of its progress in strengthening its governance, finance, and fundraising activities.

I would like to thank all the volunteers who gave up their time to give support at last year's Conference, the speakers for giving up their time to come and deliver some very interesting and valuable talks to our members, the delegates who attended and made the 2018 conference a very enjoyable

time and experience for everyone, and the staff at the Hilton Hotel who ensured that everyone was well supported, welcomed, and looked after while staying at the hotel. The 2018 conference was deemed a great success with new and old members attending with a wealth of information and advice available and good times had by all.

To the new families who attended last year, I hope that you enjoyed and got a lot out of your first conference, and I hope it was the first of many for your family. I would like to thank the Staff, Honorary Officers, Trustees, and Observers on the Committee for all their input, hard work, and contributions to the Charity over the last year. At this year's AGM, Laura Dowswell is stepping down from her role as Treasurer, and Abbie Geeson is stepping down as Secretary. I would like to thank them both for all the hard work they have done whilst in these roles for BBS UK. I, Richard Zimbler, am stepping down as BBS UK Chairman, and also as a BBS UK Trustee. I've been on the Board of Trustees for 10 years and feel it is time to leave and let someone else come on to the Board to help take our Charity further into the future.

The Charity had its third social weekend in 2018. This was attended by 30 people, who included BBS adults and support staff. We are already planning to organise our fourth social weekend for later this year. The family activity weekend was a great success and was attended by 25 people. This year there will be another activity weekend and this will again be held at Whitemoor Lakes Activity Centre. I would like to take this opportunity to thank the volunteers for giving their support at our events and for their help throughout the year as, without them, it wouldn't be possible for these events to be as accessible as they are to our members. We continue to be active in raising awareness of Bardet-Biedl Syndrome and this has entailed attending various different events such as the rare disease days in London and Birmingham, and Sight Village in Leeds. The clinics are being well attended with numbers continuing to grow and networks between patients being formed. I would like to thank the clinics teams at all the hospitals for the continued smooth running of the multi-disciplinary clinics.

I would also like to thank Tonia, Angela, and Amy for their hard work, dedication, support, and smooth running of all the clinics and support services, and for all their dedicated support to the Board of Trustees and Charity projects. Our financial position continues to strengthen, we have made good progress with the CIO conversion, and the Board of Trustees has recently recruited a new Development and Administration Officer, Rebecca Perfect, who will support the Charity with its governance and development. The Board of Trustees has also recruited a Fundraising Officer, Christine Saxon, who will further develop our fundraising initiatives.

I would like to finish by thanking all the members, their friends, families, and the public for making such a big effort with all the fundraising that they do throughout the year and for the donations, as without your support many of the services that we provide wouldn't be able to carry on in the way they do. Thank you."

Treasurer's Report: Laura Dowswell

"Income during 2018 was £97,945 compared to £51,217 in 2017. 2018 saw an unprecedented year of fundraising in which donations of £72,875 were received compared to £20,808 in the previous year. Expenditure during 2018 was £63,298 compared to £40,238 in 2017. The increase in expenditure was due to several reasons: we did more social and family weekend activities, we have employed an admin officer and there was also the cost of a new website. Core expenditure remains well controlled, although the cost of the

annual conference is the largest item of expenditure: last year, the cost was £13,785 and naturally this rises each year. At the end of 2018, the charity had total funds of £116,748. Of this, £7,466 is restricted and relates to specific grants which will be spent during 2019.

A realistic and prudent budget has been set for 2019, which will enable the core activities and objectives of the Charity to be carried out but, as ever, trustees 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. The need remains and is one of the priorities. Apart from the grants which are restricted for a specific project, 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 that freely give up their time to fundraise.

We do have a reserves policy: Charity funds at 31st of December 2018 were £116,748, with restricted funds of £7,466. The designated fund, which is for our committed expenditure in 2019, includes just over £15,000 for this annual family weekend, £8,600 for social activities, £8,700 for children's activities, £12,000 for staff costs and £35,000 for the research fund, which leaves general funds of £29,689, being free reserves. In early 2018, Trustees of the Charity reviewed the reserves policy and decided that free reserves should be sufficient to cover six months ongoing costs. In 2019, these costs are budgeted at £102,778 less the £87,059 designated fund, which leaves £29,689 for the full year. The reserves at the end of 2018, available as general funds, would represent more than 12 months expenditure and therefore the Trustees consider the current level of reserves to be adequate."

Approval of the 2018 Accounts
The vote to approve the 2018 accounts was undertaken, everyone was in favour and no one opposed the accounts.

Appointment of the Independent Examiner
BBS UK proposed Michael Bannister, of Fryza Bannister Financials Ltd, to act as the Independent Examiner for the Charity for the coming financial year. A vote was undertaken, everyone was in favour and no one apposed the Independent Examiner appointment.
BBS UK proposed a review of the independent examiner service during 2019 and requested approval to change the provider if appropriate. A vote was undertaken, everyone was in favour and no one opposed the proposal.

CIO Conversion: Abbie Geeson, Secretary
"We've worked very hard throughout the year on converting the charity structure and in December 2018 the Trustees were delighted to receive approval from the Charity Commission for the new CIO. Bardet-Biedl Syndrome UK will be registered in England and Wales with a new charity number. The CIO will become active in 2019 and Trustees will continue to set up the finance facilities and renew the membership of the new organisation.

We're not able to transfer membership from our current Charity to our new Charity, so we obviously want to make sure that everyone

who is a current member remains a member, and any new members are more than welcome too; any professionals, anyone from outside organisations and anyone who is linked to BBS, who isn't a current member, please do sign up.

Election of Committee
One half of the membership shall retire annually but shall be eligible for re-election, the members so to retire being those who have been longest in office since the last election but not reckoning ex officio members; Emma Oates and Dianne Hand are therefore retiring during this AGM.

Of the current committee members:

- Christian Bolton-Edenborough has a further year to serve of his current term.
- Dianne Hand is eligible and has agreed to stand for re-election.
- Emma Oates is eligible and has agreed to stand for re-election.
- Judith Aylott has been an observer over the last year and has been nominated and is eligible to stand for election.

A show of hands confirmed that everyone was in favour of the election and re-election of the Trustees. No one was against.

Of the current Officers:

- Richard Zimbler, Chairman, retired from the Board. The Board of Trustees thanked Richard for his support and dedication to the Charity over the years.
- Stefan Crocker, Vice-Chairman, was nominated and eligible to stand as Chairman.
- Laura Dowswell retired as Treasurer and was nominated and eligible to stand as Vice-Chair.
- Abbie Geeson retired as Secretary and was nominated and eligible to stand as Treasurer.
- Rachel Foley, Trustee, was nominated and eligible to stand as Secretary.

No other nominations had been received for these positions and a show of hands indicated that everyone was in favour and no one against the nominations, elections and re-elections of the Honorary Officers. A gift of thanks was presented to Richard Zimbler who, in turn, thanked the Board for their years of support and friendship.

Observers
The Board advised that there were three potential new committee members observing on the Board for the coming year. Members who wished to join the Board of Trustees should contact BBS UK Secretary, Rachael Foley, rachael.foley@bbsuk.org.uk.

Any other business
In the absence of any further business the AGM was closed, and members were thanked for their attendance. The date of the next AGM is Saturday 18th April 2020.

Update on Research and Study of BBS

Professor Beales
Great Ormond Street Institute of Child Health, Guy's and St. Thomas' NHS Trust

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 the Syndrome. This includes the notion that abnormally functioning cilia (small finger-like appendages on cells) lie at the heart of BBS. The challenges that lie ahead involve understanding how dysfunctioning cilia contribute to various syndrome aspects. These discoveries have brought closer the goal of designing treatments to prevent further visual deterioration or weight gain.

Professor Beales has been medical advisor to Bardet-Biedl Syndrome UK (BBS UK) since 1996, was made President of the Charity in 2005 and is a founder member of the recently formed BBS UK Scientific Advisory Board. 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 extract of Professor Beales' update on the research and study of BBS. The presentation is available in full on the BBS UK YouTube channel.

.....

Professor Beales welcomed everyone to the conference and began his presentation with an update of the BBS clinics service, which has now been running for nearly 10 years, a third of the lifetime of BBS UK; he showed a slide of what he called 'the wheel of fortune', showing all the partner organisations involved in the clinics. There are four centres: Birmingham Children's Hospital and University Hospital, Birmingham and in London, Great Ormond Street Hospital and Guy's and St Thomas' Hospital, all supported by BBS UK.

Professor Beales recalled that, when the clinics were originally set up, the world's first genetic testing centre was developed for Bardet-Biedl Syndrome (BBS). The testing centre has been extremely beneficial with almost 800 genetic tests completed to date. He continued by saying that 21 BBS genes have been identified, with a couple more that the team know about. It's only necessary to have two 'spelling errors' in any one of those genes to cause problems associated with BBS. Of the tests to date, about 60% are for those with BBS and about 40% are for relatives, some of whom wanted to have carrier testing. In addition, there have been over 20 'predictive testings' that test in the womb or on newborn babies to determine in advance whether or not they're going to be affected.

Professor Beales said he was really amazed that testing had confirmed a BBS diagnosis in over 80% of those thought to have the Syndrome. It's really important for a number of reasons that we go back over the results to try and understand whether symptoms differ according to different gene mutations which may have a bearing on future treatments.

There are still a few genes that need to be discovered and Professor Beales really wanted to thank Beth Hoskins who had been with his team for over 20 years. Beth did her PhD in his lab many years ago and really is the one who has driven the genetic testing, developed the tests and made them happen.

Professor Beales moved on to talk about research into BBS that has been happening

around the world and shared a graph detailing the number of papers published on the Syndrome since 1949. That year there was one paper, then the number waxed and waned with not many papers published until about 1995 when the number started to increase as people became really interested. The number of papers published shot up in the '90s and around 2000 which coincided with Professor Beales' team finding the first BBS gene and again in around 2005 when the involvement of cilia in BBS was identified. 2013 was probably the high point in terms of numbers of papers, with almost 70 published that year. This year there have been 27 to date, only a third of the way through the year, and the number should reach about 60, indicating that there is still a strong interest in BBS. He said: 'it's up to us to maintain that awareness and interest in the medical community and the scientific community going forward'.

Professor Beales continued: 'one of this year's papers, by Dr Robert Haws from the Marshfield Clinic in the US, might be of interest to BBS UK members. He's published a paper about skin problems associated with BBS that some members might recognise. His paper, published in the *International Journal of Dermatology* was entitled 'Cutaneous Findings in Bardet-Biedl Syndrome'. He essentially determined that there are a couple of skin conditions common in the general population but much more common among folks who have BBS'.

Professor Beales explained that one of them is called Seborrheic Dermatitis and the other is Keratosis Pilaris. Seborrheic Dermatitis is very common, sometimes looks like psoriasis and leads to scaly and itchy skin, particularly in the scalp. It's a bit like a severe form of dandruff and is quite difficult to treat. Keratosis Pilaris, which 80% of those with BBS seem to have, is a mild, itchy rash. It sometimes looks like eczema, but is not. It's due to a buildup of keratin, a protein that sits on the surface of our skin to help us be waterproof. The research suggests that those with BBS produce too much keratin, which clogs the pores, leading to a rash. It is sometimes

described as chicken skin or multiple pimples; it is treatable, but can be troublesome.

Professor Beales then moved on to revisit some of the research presented in previous years, for example that of Dr Victor Hernandez, who has been 'leading the charge' in terms of gene therapy. We've reached the point now where we've successfully completed the first stage of the gene therapy studies in the preclinical phase. Dr Hernandez has developed two gene therapies, one of which is for the eye. The treatment involves a sub-retinal injection of BBS1, replacing the faulty BBS1 gene in the back of the eye, and the study has shown that this actually prevents blindness in the mice that are treated. The next step is to take this forward into clinical trials.

The second therapy, which is a more systemic gene therapy, has been developed in partnership with a funding agency called Apollo Therapeutics. Professor Beales explained that, with this research, BBS1 is given intravenously into the mouse, particularly into the ventricles of the brain, and has been shown to prevent obesity; it is the first gene therapy that has been shown to prevent obesity, which Professor Beales believes could potentially have a huge impact. He said: 'we're at the point now where we think we're fairly close to getting some funding to take this to clinical trials' and hoped he could come back next year to report on whether the funding had been

obtained, what the structure of the particular study might look like and the time it would take to develop.

Professor Beales moved on to discuss the obesity focused research of Dr Sonja Christou-Savina and Naila Haq, then a PhD student. They have discovered that the brain operates as a complete unit with our gut - with the brain, gut and fat stores 'speaking' to each other and with the gut at the centre. Our fat stores send out a signal, called leptin, to the appetite centre in the brain, called the hypothalamus, and what is now beginning to be understood is that the BBS proteins really affect the part in the brain where that signal is received. Signals also come from the stomach in the form of a hormone called ghrelin which is produced when we're hungry. After we eat or during eating, ghrelin goes down in the stomach and leptin goes up. It's early work but grants have been sought to develop further research in this particular area. Professor Beales concluded with a slide telling the Conference about a new study by Rhythm Pharmaceuticals, the Setmelanotide Study, which is also concerned with these feedback mechanisms.

Question: 'The focus seems to be on BBS1. What about the others like BBS 10?'
Professor Beales: 'We did start to develop a BBS 10 study as well but for various reasons it didn't work. It doesn't mean we're not going to do it. We're focusing on BBS 1 because the challenge now isn't actually developing the study - it's getting it funded. The first clinical trials cost about 3 to 4 million pounds and that's just for BBS1, but you're quite right. We do want to do BBS10 because if you do both BBS1 and BBS10, that covers almost 65% of the BBS population.'

How your wee can help us understand BBS!

Dr Helen May-Simera
Johannes Gutenberg University, Mainz

Dr Helen May-Simera is a research scientist who has been trying to understand the molecular mechanisms underlying ciliary dysfunction in BBS since 2003; in simpler terms, she is trying to figure out why people affected with BBS have problems with their health. Helen was born in Great Britain in 1981 and studied biochemistry at the University of Bath. After completing her Master's degree in 2003, Helen was a doctoral student at University College London, where she obtained her doctorate in 2008. Helen then moved to the USA to conduct research as a postdoc at the National Institute of Health in Bethesda, initially at the National Institute on Deafness and Communication Disorders, and then later at the National Eye Institute. In 2014 Helen was awarded the prestigious Sofia Kovalevskaja award, allowing her to continue her research at the University of Mainz, Germany, as a young group leader. This year Helen attended the Conference with two members of her research team (Lena and Ann-Kathrin) who work in the laboratory with Helen, trying to understand what causes BBS so that researchers can start trying to fix the symptoms. Following is an edited extract of Dr May-Simera's presentation. The full presentation can be found on the BBS UK YouTube channel.

Dr Helen May-Simera introduced herself and began her presentation by reminding everyone what cilia are and explained why her team needed help:

Our body - all our different organs and tissues - is made up of cells which come in different shapes, sizes and functions. On many of the cells, there's a tiny appendage called a cilium, or more technically the primary cilium. Dr May-Simera explained that the cilium sticks out from the cell and looks like a hair. For a

long time people ignored it and thought it wasn't important. It was Professor Beales' work around the turn of the millennium which showed that actually this little hair is very important. Previously it was thought that the cilium was just like a radio antenna - simply receiving signals - but we now know that the cilium also submits and sends out signals via little 'packages'. This discovery is a 'game changer', something very new and something the team are trying to understand better.

Dr May-Simera explained that the 'packages' submitted by the cilia have a 'content' on the inside and 'a postal address' on the outside; the team know that the packages exist, but have no idea of the content inside them which is 'read' by other cells. Kidneys contain lots of cells with cilia, which explains why many people who have problems with their cilia, like in BBS, sometimes also have kidney problems. The 'packages' submitted by the cilia in the kidney end up in our urine and it was explained that conference delegates could support the team's research by providing urine samples.

Dr May-Simera reported that, in a pilot study, six samples of these 'packages' from six people with BBS were found to have different amounts of a given protein. This contrasted with control studies with the team's own urine which constantly contained the same amount of the protein. These differences in BBS samples is just one component and the team also need to look at other components. Dr May-Simera concluded by thanking everyone who was happy to help, and all the people who had already supported their research, and said: "maybe by next year, if we're really lucky and work really hard, we might have some results to share with you".

Setmalenotide, a treatment for obesity in rare genetic disorders: results and next steps

Dr Murray Stewart
Chief Medical Officer, Rhythm Pharmaceuticals

Dr Stewart joined Rhythm Pharmaceuticals as Chief Medical Officer in October 2018 and is currently responsible for clinical development, safety, regulatory and medical affairs. Rhythm Pharmaceuticals is a small biotech company exploring genetic causes of obesity. They currently have a treatment in phase 3 called Setmelanotide. Dr Stewart previously served as Head of Research and Design for Novelion Therapeutics, where he oversaw global medical affairs for Juxtapid® and Myalept®, two marketed products for rare metabolic diseases. Prior to that, Dr Stewart was Chief Medical Officer at GlaxoSmithKline (GSK), with global responsibility for patient well-being across the vaccine, pharmaceutical, and consumer business units. Prior to his time with GSK, he worked as a consultant physician and honorary senior lecturer at the Diabetes Center in Newcastle upon Tyne; his research was in lipid metabolism in type 2 diabetes. Dr Stewart began his career as a physician and surgeon. He holds an M.D. and Bachelor of Medicine from Southampton Medical School and is a Fellow of the Royal College of Physicians. Following is an edited extract of Dr Stewart's presentation at Conference; the full recording can be heard on the BBS UK YouTube channel.

Dr Stewart started by describing how he now lived in Boston, U.S.A., but had done all his medical training in the UK at Southampton. He intended to talk about Rhythm Pharmaceuticals, a small biotech company where he is Chief Medical Officer. He was previously Chief Medical Officer of GlaxoSmithKline, where approval was obtained for gene therapy for a rare condition called ADA-SCID, the 'boy in the bubble'

disease. Dr Stewart acknowledged that gene therapy can transform people's lives and he is sure that it will make a difference with BBS, but it won't be an easy road and may take a while. His presentation would give an idea of what it takes to get approval for drugs and he would talk about a product that's now in clinical trials and may be of specific interest to BBS UK.

Rhythm Pharmaceuticals, based in Boston, was founded in 2008 and has 60 employees. The aim of the company is to transform the lives of people with rare genetic disorders of obesity. The list includes POMC Deficiency, Leptin Receptor Deficiency, Alstrom Syndrome, Prader Willi Syndrome and Bardet-Biedl Syndrome.

Dr Stewart explained that their work relates to a pathway in the brain called the melanocortin 4 receptor (MC4R) pathway – the final pathway in the brain which regulates weight by increasing energy expenditure and reducing appetite. He said: 'the MC4R pathway is probably the light switch that says you're either hungry or you're not hungry and alters your metabolism accordingly. Some people with BBS feel hungry all the time because the MC4R pathway is affected and the signal that their appetite has been satisfied is disrupted. Rhythm Pharmaceuticals are developing a drug which targets the receptor at the end of the signalling pathway. The Company is aiming to correct the signalling to try to stop the hunger and make it easier to follow a diet and lose weight.'

Dr Stewart turned to the reasons why it takes a while for drugs to be approved and said that 'a lot of the work is done in labs, looking at mice and then saying, well, if we can do that in mice, can we do it in people? Once you've got a compound that you know is working within a pathway and making a difference, the exciting thing is going into clinical trials. For me, and I've done this a few times now, one of the toughest decisions for a doctor involved in trials is when to transfer the trials from animals to humans.'

Dr Stewart explained that the transfer to humans is carried out very, very slowly, starting with tiny doses which are really not going to have much of an effect. Then the dose is gradually increased under careful medical supervision until there is confidence in the dose level and its safety. Phase two can then follow to establish whether it works in the intended population. If all goes well, then phase three, the exciting phase, involves larger studies where they've shown the drug is safe, shown that it works, and can now see if it really makes a difference to patients' lives.

Dr Stewart explained that regulatory authorities across the world check that the drug being produced is safe and is going to work. In the USA, it's the Federal Drug Administration (FDA) and in the UK it's a group called the Medicines and Healthcare Products Regulatory Agency (MHRA). They look at all the data produced, all the raw data including the animal and lab data, and scrutinize it to check that everything has been done correctly and that the drug is safe and effective before it is approved and put on the market. The process is slow because it's all about ensuring patient safety.

In addition, in the UK, the National Institute for Health and Clinical Excellence (NICE) checks that drug pricing is appropriate and challenges companies to say that their drugs work and are suitable. It takes 10 to 15 years from the beginning right through to getting a drug to market. Dr Stewart said: 'it is a robust process but, when a drug does get to the market, you don't need to worry whether it has been through the necessary process.'

Dr Stewart moved on to talk about Setmelanotide which is in phase three clinical trials for POMC Deficiency, Leptin Receptor Deficiency, and, just starting, for Bardet-Biedl Syndrome and Alstrom Syndrome. He explained that it's a small compound which replaces a natural hormone that stimulates the MC4 receptor. The idea is that Setmelanotide attaches to the receptor and causes the 'switch' to tell you that you're not so hungry. It should make people who were hungry all the time feel less hungry and switch

off their hunger, resulting in weight loss. Dr Stewart explained that it's very important to understand that Setmelanotide is not a substitute for diet; it's really to go along with diet and help people comply with a diet. The drug is a peptide and is given subcutaneously as an injection under the skin regularly once a day.

Dr Stewart went on to discuss the results. In phase one, nearly 100 'healthy' volunteers were given the drug, starting with very small doses of not quite one milligram, but now up to three milligrams. It was shown to be safe in the healthy volunteers which led into the phase two BBS study. Gradually, longer data has shown it's safe over time. The phase two study looked at the effect of Setmelanotide on body weight, to see if the drug affected hunger and would do so in a way that would help people lose weight. The nine participants had a variety of the genetic variants of BBS, their ages ranged from 4 to 61, with the majority being adolescents, and the study looked at treatment over a year.

Six people lost 15% to 20% of their body weight in a year, between two to six stone, which Dr Stewart said is outstanding. Associated with the weight loss was reduced hunger, with hunger scores dropping by 20% to 80%. Participants said they didn't feel as hungry as before which helped them lose weight. Dr Stewart explained that some people with BBS manage fine with their diet and don't feel hungry all the time but, for others who may feel permanently hungry and struggle with their weight, Setmalenotide could help. However, he went on to say that it would be wrong to say the therapy works on everyone: 'with the other three people in the study, two people didn't respond and one person didn't lose much weight, but interestingly showed reduced hunger and an improvement in their diabetic condition, which can be a feature of BBS.'

Dr Stewart confirmed that there were no serious adverse events in the phase 2 study, although he said it should be remembered that there were only nine people in the study. A previous compound, which had not been trialled in humans, had also looked at the MC4 receptor and had shown high heart rate and blood pressure in animal models, so blood pressure and heart rate need to be monitored to make sure that there are no similar adverse effects with Setmelanotide. The injections caused some itching and a small localized rash but no one had any systemic problems. Dr Stewart went on to discuss a notable side effect of Setmelanotide, which is increased skin pigmentation. Setmalenotide affects the melanocortin pathway, which in turn affects the melanin in the skin and causes an increase in pigmentation, resulting in people starting to tan. Some said they were OK with tanning, others in different cultures don't like their skin going dark, and the effect needs to be monitored, in particular to make sure that any change in the skin is healthy melanin and does not cause any damage.

Dr Stewart went on to say that phase three had now started in conjunction with Dr Bob Haws and his team in the Marshfield Clinic in the States. This is going to be a global study and Dr Stewart has already recruited seven or eight patients in the UK, France, Netherlands and Spain.

Dr Stewart explained that during the first three months of the study there will be

some people getting the drug and some getting a placebo, to ensure that weight loss is connected to the drug and not simply enthusiasm and lifestyle changes. He said that 'it would be unfair not to allow all those with BBS to get the drug, so after three months people will be switched from active drug to placebo and vice versa, with the object being to see if the phase two results are confirmed, in other words to see if the drug causes more than 10% weight loss and reduced hunger over a year.'

Dr Stewart concluded by thanking all those who have taken part in the clinical trials and said: 'our research is about developing medicines that can change people's lives and there's an opportunity here with Setmelanotide to help people who've got insatiable hunger, help them with their weight. In addition, there's the opportunity to start making others aware that there are people who have obesity and related conditions where the obesity is not their fault. They may have problems with their genes that mean their signalling is not right and their drive for hunger is very difficult to control without therapy.' Dr Stewart said he wants others to be aware of conditions like BBS, he wants to support BBS UK and to contribute to developing therapeutics which can change lives.

BBS Clinics Telemedicine Service

Dr Shehla Mohammed MD, FRCP
Kath Sparks (CNS)
Guy's and St. Thomas' NHS Trust

Dr Mohammed obtained her degree in Medicine from Pakistan and has subsequently worked in the NHS for 34 years, training in paediatrics before specialising in Clinical Genetics. She was an ICRF Research Fellow in Cancer Genetics prior to taking up a Consultant post at Guy's.

Dr Mohammed has a longstanding interest and experience predominantly in rare genetic disorders and in the care of children and families with life-limiting disorders. She has been involved in setting up and running the National BBS clinic for adults with Professor Phil Beales since its inception in 2010 and oversaw the move of the service to its new home in the Rare Disease Centre (RDC) at St Thomas' in 2018. Dr Mohammed is also involved in the running of other highly specialist services at the RDC.

Until 2017, Dr Mohammed had been Head of Service of the Guy's Regional Genetics Service for 20 years. She represents the genetics speciality on local, regional and national committees (member of highly specialist committee of NICE) working on national policy developments and research. Other interests include travelling, photography, gardening, being a mature student of the cello and a recent convert to kick boxing! Dr Mohammed, supported by Kath Sparks, CNS for the London BBS clinics service, gave a presentation at the BBS UK Conference about the new telemedicine service. Following is an edited extract; the full recording can be heard on the BBS UK YouTube channel.

Dr Mohammed began her presentation by explaining the importance of patient feedback to the service which, she said, was a helpful and useful resource because, over time, there is a need to change how the service manages and helps patients. There has been an increasing number of referrals, which Dr Mohammed said is probably a combination of greater awareness of BBS amongst many medical specialists, as well as greater awareness about the availability of the clinics service which has been established and running now for nearly ten years. Technology advancements have enabled the service

to confirm the diagnosis in about a third of patients before there are obvious clinical symptoms, and patients are being diagnosed with BBS from the age of three months up to their late 60s.

Consequently, the number of people attending all the clinics has been increasing and there is a need to accommodate new referrals, but also to make sure that follow-up appointments run as planned. Dr Mohammed said the team are very aware that the length of the follow-up time has begun to increase from a year to 17 months and in some cases 23 months, which is something that they felt very uncomfortable with.

The service is funded by NHS England and there is only a finite pot of money, so there is huge pressure and constraints on the service. Dr Mohammed explained that extra clinics are just not possible because of space and staffing commitments elsewhere, so the service has introduced telemedicine clinics. The service leads asked for additional funding for all four centres from NHS England and last year the funding was agreed and this initiative has been running since January.

Dr Mohammed explained that the process for patients being seen in the telemedicine clinic is simple, straightforward and secure; the IT equipment used is state of the art and it works. A summary letter is sent out to the patient after their visit in the same way as with a regular clinic and follow-up tests and appointments are also arranged as needed. The team also wanted to use some of the established systems in place such as Certus, the patient management system. For the paediatric clinics, the team contact the child's school nurse and GP to obtain height, weight and blood pressure ahead of the telemedicine appointment.

Kath Sparks outlined the process for signing in to a telemedicine clinic, which for the adult service at St Thomas' uses Skype for Business. The process for what needs to happen pre-clinic depends on whether the patient has a laptop with a camera and a microphone or an iPad. If the patient has a laptop with a

camera and microphone, the team sends a link to their email address. The patient simply clicks on the link, and they are taken to a web page which allows them to join the meeting. If the patient is using an iPad or iPhone, they will need to download the Skype for Business 'app', and then follow the link from the email. Kath reported that there have been some technical difficulties, but they've managed to troubleshoot quite a few. A slightly different process is followed at the Queen Elizabeth Hospital, Birmingham, which uses the hospital's online patient portal. This system currently requires patients to use a laptop with camera and microphone.

Dr Mohammed reported that they have had some initial feedback which has been largely positive; patients felt that their appointment went very well and felt that they had time to ask questions. Some people found there was enough time, whilst others felt that the appointment was too quick. There were some really helpful suggestions for future service users, such as writing down questions beforehand so you could make sure your concerns were addressed.

Dr Mohammed finished her presentation by saying that the team are learning from the process and will fine-tune it as they go along. The telemedicine appointment also gives the team an opportunity to signpost people if they really do need to be seen in person and it is hoped that, in the future, this additional service will mean that nobody has to wait longer than they should for an appointment.

The Importance of Whole Family Support

Sally Flatteau Taylor
CEO, The Maypole Project

Sally Flatteau Taylor is the founding Chief Executive of The Maypole Project and has over 25 years' experience of providing holistic counselling and support including to families facing the diagnosis of a child with a complex condition. Her work also includes training and supervision for counsellors and other professionals. Sally's consultancy work has included ongoing input into strategic development of specialised children's counselling provision in many settings across the UK. Following is an edited extract of Sally's presentation at Conference; the full recording can be found on the BBS UK YouTube channel.

Sally began by explaining that her background was in counselling with a special interest in family support for all ages. She said: 'when a child or children in a family have complex medical needs, the whole family life can be affected and there are quite a lot of gaps in support services.' Sally is a practicing counsellor and supervisor-trainer.

Sally digressed slightly, asking if anyone watched 'Line of Duty'. She had heard a key line in the script, which went: 'at AC-12, we have a UCO in the OCG and we need an ARV at the ready.' She said, 'this may make sense to people who watch the programme and who know the culture of the police. We have to get into the language and to an extent the culture within each area of our community where there are different cultures and different languages and this can be applied to what happens in families where everyone's experiences are so different that communication can become disjointed.' Sally continued: 'it can be a really tough job to get your head around the different names, the medicalized terminology and sometimes the acronyms that are used, like BBS.' People outside the Charity wouldn't necessarily understand if you said my adult child has BBS, which can create separation for all of us living with a child with complex needs.

Sally explained that communication is a really important area of a world that some will feel turned completely upside down, realizing that a child, young adult, or adult has so many different things going on, with different professionals looking at different aspects and maybe not bringing it all together and recognizing Bardet-Biedl Syndrome as a whole. She continued by saying that there is the new language of communication with professionals, with new people who come into your lives, and even within the family, because different people get different bits of communication at different times. New feelings come in too, different feelings for different members of the family at different times. Some will immediately feel quite angry about everything, will go into campaigning, will Google search. Many of us do that and then we can be very much on our own, going: 'Oh, I've Googled it and I've found all this out and now I've got to make sense of it.'

Sally explained further that it's like the effect of a snow globe, you turn it upside down and all the bits of snow or sand start forming a different pattern. That's how life can go, with everything in our lives starting to form a different pattern and people feeling the rug has been pulled out from under their feet. The snow in the globe doesn't fall in the same place. It can be a comfortable place, it can be familiar, but it can also be very unfamiliar. Everything changes, and the way that each family member reacts may be different. Things don't happen in a linear way, they're all over the place. It would be nice if we could say: 'this happens and you feel angry, then you feel this, then you feel that'; but it isn't like that and every member of the family may feel differently. There's a wonderful myth that families all pull together when things happen, and that may be the case, but sometimes the changes can cause

disconnect in families and then the whole family should get support, separately though in a way that can also bring them back together.

Sally continued: 'communication can be the root of disconnection and often one member of the family will become the main carer and as such will get most of the information. This information then needs to be passed on to other family members which can be quite a tough thing to do. The other family members might feel disconnected from the information, disempowered, out of control, even excluded sometimes. That's how serious the problem can be, and why it's important to look at how a whole service can be created to help bring families together.'

Sally explained that at the Maypole Project in Bromley they often get referrals of families to professionals at critical times, at diagnosis or at change of treatment. They found that at the time of diagnosis, at times of change, it's all really busy for the family; they're getting their heads around something new that's happened to them, something new that they're learning or some new treatments and it isn't a time when they'll necessarily engage in family support. They'll maybe want to know if someone's there to help, but it's often in the quietness afterwards when there's no practical work being done that feelings come rushing in.

Sally noticed there was no support in her locality for families with children who had complex needs and the Maypole project was set up to work with families across all different diagnoses. It can be really important to sit down as a family and talk to someone else because in that way you'll hear each other's different perspectives. One of the families that worked with the Maypole Project talked about the Project as being 'the glue that holds our family together.'

Sally thought that the BBS UK Conferences were 'absolutely marvellous' for everyone to get together, to have the time and space to talk in different settings, with the children going out on activities. BBS UK has also, she said, very creatively filled another gap with the clinics themselves, bridging the gap between the medical world and the personal world, helping

families take and digest the information that they're getting at clinic.

Sally finished her presentation by briefly looking at the effect a complex illness or diagnosis can have on children. She said: 'sometimes they know, sometimes they don't – and this applies to their brothers and sisters. The Maypole project does a lot of work with brothers and sisters because again they can sometimes feel excluded from information. They can also be really scared and protective of their sibling, or scared for themselves because they don't understand all of the impacts and everything that's going on.' Sally suggested that the thing to look for in our communities is an organization that can provide family support and maybe offer counselling, but one that is not prescriptive. For example, a prescription of six sessions may not be what you need; you may need something you and your family can dip into, an organization that helps you be in control of your own support, to tap into when you want it and build a relationship going forward, as *you* go forward as a family. The Maypole Project, born in Bromley, works across South East London but does have a national telephone helpline: 01689 889 889.

For more information go to www.themaypoleproject.co.uk or call 01689 851596.

Weekend Round-Up

Breaking Down Barriers

Kerry Leeson-Beevers

Kerry Leeson-Beevers is the National Development Manager for Alstrom Syndrome UK, which holds multidisciplinary clinics at Birmingham Children's Hospital and Queen Elizabeth Hospital for adults. Kerry joined ASUK in 2002 and manages the family support service and oversees contracts with NHS England and with pharmaceutical companies. Kerry's son, Kion, has Alstrom Syndrome (AS). He is 18, has no vision at all and has got severe hearing loss but Kerry says he lives life to the full. Alstrom Syndrome UK is closely linked to Bardet-Biedl Syndrome UK and this has given us the opportunity to work together on the Breaking Down Barriers Project. Following is an edited extract of Kerry's presentation about the work of Breaking Down Barriers; the full presentation can be heard on the BBS UK YouTube Channel.

……..

Kerry explained that the Breaking Down Barriers (BDB) project developed from ASUK's Asian Mentoring Scheme which was set up to reach out to ethnic minority families living with AS; at that time, only three of their families were from an ethnic minority community. They targeted lots of different visual impairment teams and ophthalmology services throughout the UK, and worked with the professionals to link with families, find out what their needs were and bring them into the Charity. The main funder was the Sylvia Adams Charitable Trust and, as a result of this successful project, ASUK now have over 40 members from an ethnic minority background; they are rightfully proud to have become a diverse and culturally representative organisation.

Kerry said, "I presumed, if I employed somebody that could speak Urdu, that would solve all our problems but actually we were successful because they saw me as a mother rather than a professional and that helped gain their trust and brought them into the clinical service. We were able to recruit an equality and diversity officer, which taught us a lot about what changes we could make. One of the key learning points for us was making sure that both parents were supported and given the information that they needed at a time that was right for them."

The Sylvia Adams Trust were very impressed with ASUK's original project and after a couple of years approached Kerry with funds to develop the idea further - together they came up with Breaking Down Barriers, which is about helping patient organisations, community groups, support groups and community networks to develop supportive and inclusive services, making sure that all families, whatever their ethnicity and background, have access to accessible information to be able to make informed choices about their future and better manage their condition. Kerry explained that BDB members work with Jewish communities, French and Portuguese speaking communities and travelling communities; however, their main focus area is in South Asian communities.

BBS UK have used their funding to produce information resources which will be available in Easy Read, Urdu and Turkish. They have also held coffee mornings to engage with families to find out their needs. Kerry was pleased to report that attendance of ethnic minority families at the BBS UK conference has increased as a result of their involvement in the project. She also congratulated BBS UK on their excellent project evaluation and monitoring.

The BDB project is now in a position to fund an additional 15 organisations and received many more new applications than they had expected. BBS UK are extremely grateful to Kerry, the BDB Advisory Board and the Sylvia Adams Trust for their invaluable support throughout the project and look forward to working with them over the coming year.

RNIB: celebrating 150 years

Ellie Southwood: Chair, RNIB

Ellie Southwood is Chair of RNIB and has been a Trustee of RNIB since 2010. Over the past year, she has steered development of RNIB Connect, a growing community of blind and partially sighted people who support, inspire and share information with one another. Ellie is a champion for employment opportunities for people of working age and for involvement of blind and partially sighted people in civic and public life. In 2014, Ellie was elected as a Councillor in the London Borough of Brent, where she is now a cabinet member responsible for housing and welfare reform.

Ellie's career spans the public, private and non-profit sectors. She specialises in supporting organisations to develop strategies and implement cultural and operational change. Ellie's early career was in public policy and includes research and stakeholder management for a local government think tank, a communications company and the Confederation of British Industry (CBI). Ellie has a Masters in Organisational Behaviour (2013) and a first degree in Philosophy, Politics and Economics from Oxford University (2003). Following is an edited extract of Ellie's presentation at the BBS UK Conference 2019; the full presentation can be heard on the BBS UK YouTube channel.

"One of the fantastic things about RNIB is having 150 years of amazing heritage, but that can also be one of the difficult things because we can perhaps look a little old-fashioned. When I was reflecting on what RNIB had ever done for me, I thought about the tactile watch that I learned to tell the time on, the lorry loads of braille books that arrived throughout school and university so that I could study alongside my peers, and tactile paving, that means I can, in most cases, cross a road in safety! But there was so much more that could be done and when I put those views out there, the then Chair said, "well, there's only one way of changing that, come on board and be part of our future."

As part of our 150th celebrations last year, we did some research on what blind and partially sighted people felt were the greatest barriers. Only one in four of us is in work; and if you're totally blind, that's one in ten. There are some very outdated attitudes too which are not intended to be rude or insulting but are born of ignorance, shyness, or awkwardness. When questioned, the general public didn't think blind and partially sighted people could enjoy TV and films, but audio descriptions

have been around for a long time and I've got through a number of box sets recently! They didn't think that we could enjoy reading but I think we'd all disagree with that. Also, lots of people weren't sure blind or partially sighted people could be bosses, teachers, lawyers, or MPs. I haven't plucked up courage to tell David Blunkett about that yet!

We have been working on bringing together and harnessing the voices of children and families and we've been looking at how to make sure our services are UK-wide. We're focusing on improving and protecting services which are, after all, statutory. Blind and partially sighted children have the same right to education and inclusion in the UK as sighted children, but we know that's not a reality for many of those 34,000 children and young people.

We know that families are struggling to get the support they need, so about 18 months ago we coordinated a group called the Young Vision Alliance to take a look at the situation. Depressingly, we found that one in three local authorities have reduced spending on young people with visual impairments, and that meant 700 children were receiving less support than they had previously. A third of local authorities had seen a decrease in the number of Qualified Teachers of the Visually Impaired so they were experiencing huge caseloads.

Only a quarter of young people said they didn't have a problem accessing exams. Rightly or wrongly, our society puts a great deal of store on exams and tests, and how well you've done, so if you can't access it it's not a level playing field. We are working with Ofqual to make the exam system fair. I've got some pretty painful memories of things that happened in my exams. I'd worked for three years on my degree and the only thing I was going to be assessed on were the exams in a two-week period, and they were messed up with wrong braille papers and crashing computers. It took me a long time to come to terms with the fact that I felt almost cheated out of how well I'd done because I wasn't given a fair chance.

Of young people who are older than 16, 38% are NEET, so Not in Education, Employment or Training. That's double the average for the general public. Our recommendations focused on proper support and plans, so that every young person has a plan and a commitment for what will be done to support them. This isn't about getting by. This isn't about running to catch up. This is about thriving in a society where we still face barriers and attitudinal issues. We've been campaigning on those issues, and that's one element of how I think RNIB can help families. Our sight loss advice service, which is available across the UK, takes 500 calls a month from all sorts of people including families and teachers. Our website has some downloads for parents on issues around sight loss including the extra financial support you might need. It also has advice for young people, including the difficult transition from school to college.

This year, we are running 22 shape and share events across the UK, which will be shaped locally depending on what local families want. Engaging with people and hearing their stories helps us to understand what people are experiencing. Our Parents' Facebook group currently has 600 members, but we'd love loads more so that people can share

experiences and then we can amplify those experiences when we talk to the government because I don't want us to be getting the messages wrong. Our library service has 56,000 users of Talking Books, and our Bookshare service, which is aimed at schools and colleges, gives young people access to curriculum material. We also have counselling, transcription and music advisory services.

RNIB has come a long way in 150 years. The founder, Dr. Thomas Armitage, was convinced that access to education and employment was the key for equality for blind and partially sighted people and I'm not sure I'd say anything different today, but the way we respond to that has to change and it has to evolve. I want to see RNIB working much more closely with organisations like BBS UK, because we all add value in our different ways, and we have a responsibility to really improve access and services for young people so that we can thrive."

One of the delegates commented that they had used the RNIB legal rights advice service on a couple of occasions and had found it excellent for getting the help and the advice they needed. Ellie said it was helpful to know what's really useful for when they're making decisions about where to invest resources.

Let's Get Moving!

Maria Dowswell: Zumba Instructor

Maria Dowswell is a Zumba instructor and facilitated several Zumba taster classes across the conference weekend. Surprising the delegates half way through the formal conference programme, Maria had everyone on their feet for a short taster session, which certainly woke everyone up! Before she started, Maria told us a little about how she became an instructor:

"Since I was diagnosed with BBS at age 13, I've been very determined to keep active; however, at school, PE lessons were difficult. Although teachers tried to encourage me to take part, I never found a sport I enjoyed or got into. Since leaving education and going to work, I found a local Zumba class and I decided to give it a go one night. I didn't know what to expect, but it was fun and I wanted to do it all over again, right there and then. I went to classes for a couple of years and made some really good lifelong friends, who I now socialize with outside of Zumba.

My own teacher was very supportive when I wanted to become a Zumba instructor myself and would let me teach a song each week. At first I was nervous with all the eyes staring at me, but I quickly found it natural. It only takes one day to train to become an instructor, but once you become

an instructor you are monitored and assessed on what you're doing. You have to log that you actually teach and do classes, so a lot goes on behind the scenes. Setting up and teaching, well now that's the easy bit. At home, practicing the choreography and getting it right and organizing all your classes and marketing, that's the hard bit. I was very happy and pleased when I passed. I now teach Zumba classes twice a week as well as covering classes for other instructors."

Slimming World

Amanda Avery, PhD, RD

Amanda worked as a community dietitian in Derbyshire for over 20 years, with both a public health remit and as a clinician in primary care. She moved to the University of Nottingham in 2009 as a lecturer in Nutrition and Dietetics, leading on the postgraduate courses, contributing to the undergraduate teaching programme and supervising masters research projects. Amanda initially worked with Slimming World after project managing the Slimming World on Referral feasibility study. Amanda's consultancy remit at Slimming World now focuses on research to evaluate the role of commercial slimming organisations in providing weight management services to diverse population groups. Following is an edited extract of Amanda's conference presentation; the full recording can be heard on the BBS UK YouTube channel.

Amanda opened her talk by commenting on what a privilege it was to be at the Conference and said she was overwhelmed by the sense of community in the room. Amanda introduced herself as a dietitian but one with a varied role and explained that her main job is as a University Academic.

Amanda stressed that Slimming World supports people from different backgrounds, of all shapes and sizes, and said that there are many reasons for people wanting to lose weight. She described Slimming World as a national organisation that has been running for 50 years and was proud that the group had adapted rapidly over the years and now has 19,000 weekly groups running across the UK. Amanda went on to explain that most groups are community-based and run from local venues that are easily accessible. She reiterated that Slimming World is all about group support and people encouraging each other within these groups, which she said was 'incredibly important and valuable'.

Each week people in groups share different ideas and experiences and Amanda stressed that there is no form of judgement and that everybody in the groups respects one another. Members genuinely know how hard it is to lose weight and the groups themselves are led by members that have had to manage their weight. Amanda explained that their aim is to make people feel better about themselves and she listed some of the additional support available, including Slimming World Magazine and a recipe book.

Amanda emphasised that there is a lot of praise and recognition within groups no matter what small steps and progress have been made. This is aimed at all stages to ensure people feel good about their weight loss journey, because it is the long term changes that are important; quick fixes generally don't work. Amanda stressed that Slimming World is not a short-term programme and that it is open and accessible to people for as long as they need the support. The group support helps people think about how they will cope with different situations, they may be thinking, 'I know I'm not going to cope with this' or 'my weight loss journey is going to go haywire' and so

on. She said there is genuine support for these challenging situations.

Amanda explained that Slimming World's philosophy is that they want people to enjoy their food. They encourage everyday, normal family food that's tasty and they introduced the concept of 'Food Optimising Principles'.

There are three main concepts to Food Optimising: 'Free Foods', 'Healthy Extras' and 'Syns'. 'Free Foods' should make up at least 80% of food intake. Lean meat (or meat alternatives), fish, poultry, eggs, pasta, rice and of course vegetables, fruits and pulses are all considered free foods that we should be having plenty of. These foods tend to have less calories than other foods on a weight to weight basis but are more satiating, i.e. help us to feel fuller for longer.

The second group of foods are referred to as Healthy Extras. These are foods that people are encouraged to have in measured amounts. They contribute to the overall nutritional content of the diet and help to ensure sufficient intake of dietary fibre and calcium.

Amanda then introduced the third food group called 'Syns'. These foods have a higher energy density and tend to be high in either fats or sugars. 'Syns' include foods such as chocolate, crisps and confectionery and they all often have low overall nutritional quality. Amanda emphasised that these are foods that we should be choosing last and limiting within our diets but not necessarily going without entirely.

Amanda explained that Slimming World provide simple and easy recipes that don't consist of a long list of complicated ingredients but, more likely, things that members will already have in their cupboards. The recipes also try to cater to people on low incomes who may normally find it more difficult to access healthier eating due to the cost.

Something important to Amanda is that the food and the eating plan principles really improve the nutritional quality of the overall diet and members are encouraged to make wiser food choices using the Food Optimising Principles. As well as making wise food choices, Amanda went on to explain the importance of members becoming more active not just for weight loss management but also for motivation and overall health. This can begin with just 5 or 10 minutes walking each day and gradually increase over time.

Slimming World offer a tailored program for young people aged 11-15 years and they can attend their local meetings free of charge if their parent or guardian is a member. Slimming World on Referral is a 12-week programme that can be extended in blocks of additional 12 weeks, prescribed in certain areas by GPs and healthcare professionals for patients struggling with their weight. In areas supporting this scheme, members receive the same support as any other Slimming World member and will have access to all the same benefits. Amanda finished by emphasising again that Slimming World want to work closely with health care practitioners to forge partnerships with different people from different backgrounds, to enable them to get to where they would like to be, in terms of their weight and their sense of wellbeing.

My Parkrun Journey

Tom Oates

My name is Tom, I'm 22 and I'm going to talk to you about my journey on Parkrun. I decided to start running because there was no option available for me at high school to compete in sport. This made me unfit and I put on weight and I didn't want people to think I was going to be unfit and a lazy teenager.

My mum and dad go running and they persuaded me to try a local Parkrun. The Parkrun is a 3.1 mile/ 5 kilometer run. Parkruns happen all over the world in five continents and 23 countries. The first time I went, I could only manage to walk half of the loop, because I was so unfit. It took me several weeks to build up to doing the whole Parkrun.

On my third run, I managed to clock a time of 44 minutes; this happened in January 2015. The main obstacle for me has been my eyesight - because I'm blind, I have to run with a guide. This can be my mum, my dad, my personal trainer, Stewart, or a friend who I ask to help me, or someone who wants to run with me.

The Parkrun is very popular and I can find the number of people at Parkrun quite challenging, because everyone crowds together at the start line, but I know I've got to listen to whoever is running with me on the day of the run. Doing the Parkrun has made me feel proud and happy, so I was determined to keep going. To start with, I ran with Mum and Dad and my mum's friend, Pam, but now they just can't keep up with me!

When I first started Parkrun I hovered around 40 minutes, until I met my coach Stewart through a community link in October 2015. Since I met him we have set goals which are my weight, my diet and overall my body composition. He is very polite, patient and amazing to work with and very encouraging to run with. I have built a close relationship with Stewart; when I first met him he was just a link, but now he is important to me, a good personal trainer. The prices for personal training are quite expensive for me, but I enjoy doing it. I run every week with him. Through that, I have been able to get the time down to 26:50 which is my personal best.

I have been across the country doing Parkruns including at Perth, Brighton and Colwick. I am now trying to challenge my mum's time of 23:15 which is her personal best and I have completed several 10k runs as well. My next goal is to run a 10-mile run and do it for charity.

My message to you is I really like running and if I can do it anyone can; you've just got to keep working at it.

Living with BBS

Rachael Foley

Following is an edited extract of Rachael's personal perspective of living with BBS. The full recording can be heard on the BBS UK YouTube channel.

"This is me and my life with BBS. I'm Rachael, I'm 38 and I come from Stockport. I went to mainstream nursery, primary and secondary school and it wasn't until about the age of 15 that I started to struggle with night blindness. I started falling over things and my parents thought I was being a clumsy teenager. I went to the optician who referred me to Stepping Hill Hospital where I was diagnosed with retinitis pigmentosa. There was no cure and I was going to go blind. At the age of 16 it was a bit of a shock, as I had always wanted to travel the world and undertake international tourism.

Following the diagnosis, I decided to go to college to study leisure and tourism and during the course I obtained student of the year and got a distinction. I then went on and did the two-year advanced course and got a distinction in that as well. As a student in college, I worked in supermarkets, I had a good social life and the only problem I seemed to have was my night blindness; everything else was good. Following college, I went to work at 'Going Places', a travel agency.

After a year of doing that, I decided it wasn't for me and in 2001 I started work at Stockport Council Bureau. In July 2002, I got a permanent position as a clerical assistant. It was brilliant, it was a new unit in the community safety team, helping raise awareness of crime, disorder, anti-social behaviour, and domestic violence. In 2007 I became a domestic abuse coordinator, helping survivors of domestic abuse, supporting them through court, raising awareness, organising conferences and working in the police station.

Unfortunately, in 2008, someone in my life passed away that was very, very dear; that was my Nan. She had a brain tumor which was diagnosed in February and on the 28th of October she passed away. She was a massive person in my life and she was very determined; she had the gift of the gab, so I think that's where I get it from!

In 2009, I carried on, living life, working at the domestic abuse unit, and I lived with my partner in our own home. In 2010, I was diagnosed as severely sight impaired. My sight had changed, but it was still okay as the only problem I seemed to have was the night blindness. I was able to go out and about with friends and family, and it didn't really stop me doing anything.

In 2011, after several episodes of what I thought was fainting, I was diagnosed with epilepsy; I was given medication and it seemed to help stabilize it. 2011 was also the year I got married. In 2012 I was diagnosed with BBS1 and yet again it was: 'Well, what does that mean? What is BBS?'

I didn't have a clue, but when Manchester Eye Hospital explained everything, things really did click into place. The obesity, tick; polycystic ovary syndrome, tick; wide feet, tick; extra teeth and other things as well all just clicked into place.

2013 was one of the worst years of my life as our unit was restructured and I didn't get the job I wanted. When I went for the position, they gave me the test in a size 11 font; as you can imagine, not being able to see, I had a panic attack and the interview that followed, all I can say is, it went very wrong. I didn't get that position, but I interviewed for another post which I got; it was a generic community safety support officer.

Also in 2013, unfortunately, my marriage didn't work and we went our separate ways. In the October, I had the worse seizure of my life; I was at my mum and dad's, and that's all I know, except that I was cradled in my dad's arms, going blue.

In 2014, I returned to being Miss Foley again - Mrs Walker went out of the window. I got back to being the single girl in her own home and I was trying to find people of my own age who had a visual impairment. I couldn't find anybody in Stockport so I looked further afield, and I went to an RP Fighting Blindness Patient Information Day in Birmingham. Well, for my sins, I met the famous Liam Dempsey and I also met a lady called Ruth Walmsley, who founded Manchester Blind Football Club. I exchanged numbers with Ruth, and all I can say is it changed my life forever. At the end of 2014, I went to my first Manchester Blind Football Club event and made lots of friends.

In 2015, I got more involved with Greater Manchester VI Bees, which is a sports and social club. The club paid for me to take part in the level one tennis coaching course. I was the only blind person on the course; there were ten of us and, yes, I was like the alien that landed in the room. We were paired up and everyone else got paired up first and I thought: 'hello, I'm here!' Eventually, I got paired with a young lad who was 19. He had a brother who has autism and I like to think I raised his awareness of disability; he now plays sound tennis with his brother. I qualified as a level one tennis coach in July 2015 and went on to help assist with and play sound tennis. I also set up and help run goalball within the Manchester area, and I play and facilitate in the blind football sessions and tournaments.

In 2015, I attended my first BBS clinic in Birmingham, where I met two people who now are life-long friends; I see them all the time, and we socialize a lot. I went on my first activity holiday with Galloways for the Blind and tried windsurfing, kayaking, canoeing, high tree climbing and jumping off a 60' tree and abseiling back down.

In 2016, the unit where I was working was going to be disbanded and I was thinking: 'What am I going to do?' They sent me to occupational health who showed me a job description and there was nothing

on there that I could do; I was put on the redeployment list. I went off for Christmas in 2016, and in January 2017 they were telling me that I could be on the redeployment list for more than two years. I was going into work Monday, Tuesday, Wednesday and just doing what I wanted. Everybody around me was stressed out of their heads doing three jobs and I was there doing what I wanted, going on YouTube, Facebook, and helping research charities. In the meantime, I became an observer with BBS UK and I started volunteering with the Royal National Institute for the Blind.

It was suggested that I should go down the route of ill health and three months later, after battles, and meetings with managers, trade unions, you name it, I got the answer I wanted; I was able to leave on ill health grounds and at the age of 36 I became a pensioner. After 16 years working in the community safety unit, I did think: 'What do I do now?' I'm only 36 and part of my contract is I can only do volunteering work, so I decided that I would do more work with the RNIB. I became a member of the Northwest Network Committee; I am now a committee member and the vice-chair. I then decided I would get more involved with BBS UK and became a Trustee; as of today, I've become the Secretary.

In August 2017, I went away to Majorca with three of my best friends who also have BBS. We didn't have any sighted friends with us which scared our parents to death. We had all the support we needed and we were treated like kings and queens. There was one funny moment when we heard a couple talking about us, saying: "These two with the white canes…" "Oh no, there's not, there's four of them with the white canes." We had an amazing holiday.

In 2018, I had a new partner in my life, Richard Zimbler, and in July last year we went on our own to visit Richard's dad in Australia; we had assistance from the airport staff, which was brilliant. We got to hold a koala and feed kangaroos. We went to Sydney on a road trip and went to the Opera House and to the zoo; it was an amazing month.

In 2018, I also got more involved with Greater Manchester VI Bees, going on activities, days out, field trips, gigs; you name it, I did it. I also went to Egypt with my three best friends and we had an amazing time. I would say, to anybody newly diagnosed with BBS, or family members of people with BBS, just because you have been given a diagnosis of BBS, it doesn't mean your life's over. My life has changed, and most importantly it has grown immensely.

I am busier now than I ever was whilst working. I do more volunteering and live life to the full. One thing I would say is focus on the abilities you do have and not your disability. With the ability you have, you can do anything you want in life; you may have to persevere, you may have to do things in a different way and it may take you a lot longer, but I am proof, myself, and other people out in the BBS world, that you can do it, you just have to put your mind to it and be determined. Once I put my mind to something, I don't let it lie and I make sure I do it.

One last thing I'd like to say to two people in the audience, as without them I wouldn't be able to do what I've done, I want to thank my mum and dad as without you both, I wouldn't be the person I am today."

Wilkie Warriors Fundraising Year

Liam and Ashley Wilkie

Ashley and Liam Wilkie gave a joint personal perspective about their year of fundraising for BBS UK following the diagnosis of their daughter, Caitlin; their hope is that by sharing their story they may inspire others to tackle a fundraising event. Following is an edited extract of their presentation; the full recording can be found on the BBS UK YouTube channel.

"We have two gorgeous little girls, Caitlin who is four and her little sister Niamh, who is 18 months, and live in a small town called Kirriemuir which is in Angus, up in Scotland. Caitlin was first given her diagnosis when she was two and a half. I was lucky; I sailed pregnancy. The first thing that we noticed about Caitlin when she came out was that she had six fingers and six toes. As she grew Caitlin, wasn't meeting her milestones. Her peers were crawling and babbling and we had this little pudding that sat there; all she wanted was milk and to sleep. She was the most amazing little baby. She gave us no problems whatsoever; as long she was fed, she was happy; but that was all that she was doing. We got our health professionals involved when she was about 18 months and she was just shy of two and a half years when we got the diagnosis of BBS. Shortly after that, we fell into the arms of BBS; we got in touch with Tonia Hymers of BBS UK, who was a bit of a lifeline for us.

Tonia told us that the BBS UK Conference was about to happen just the following month and so we came down for our very first conference. We came away inspired by some of the talks that we heard and back home we chatted quite a bit about the work that Professor Beales and his team were doing with gene therapy. We felt that if we got a breakthrough in that field that would make such a difference to all those with BBS. We couldn't take the Syndrome away from our little girl but we decided to dedicate 2018 to doing a little bit of fundraising, not only to help BBS UK, but to also help the wonderful research that's been going on at GOSH and other places as well.

Charity wristbands came first! We had to buy a batch of about 200 and we were quite apprehensive because we weren't sure if they were going to sell; within two or three days, the first 200 had gone. We sold a lot to family and friends and had a lot of interest from local shops who wanted them on their counters. To date, they're still selling, and

we've ordered more than 1000.

We were quite lucky that we got some media awareness because of the wrist bands quite early on. Our local paper wanted to do an article on Caitlin. That was in October of 2017 and on the back of this article we started to get people interested in BBS and having people approach us about wanting to do some fundraising. It was, certainly, the start of something very big for us and our little town.

The local Co-op got in touch with us and offered to carry out a Valentine's Day raffle. We were delighted with their offer but even more delighted when we saw the effort they put in and the total amount made. They have continued to support us throughout the year. In total, they've raised over £1,000 for the Charity, which is amazing. The community spirit we have in our local town has really spurred us on.

We decided, as a family, that we would organize a race night. The race night was pencilled in for the 3rd March 2018. We put our heart and souls into it; we got a lot of race sponsors, collected lots for a raffle and had some amazing prizes for an auction. We decided to run our own bar because we knew a lot of our friends enjoy a tipple and in all we raised a super £8,550.

Next up was a local feel-good festival and the opportunity to discuss and explain to our local community about BBS; to say we were nervous is a complete understatement. We worked through our speech and were delighted to see so much interest in the condition. It was an emotional speech for us both as it was the first time we had properly spoken about BBS in public. Despite a few tears, we got through it and the confirmation that we had spread a little further awareness for all the locals who are going to see Caitlin grow up was all we could ever have wanted.

Our three very close friends, Karen, Emma and Karen decided that they wanted to do their little bit for Caitlin and signed

themselves up to do a half marathon, which was a massive challenge for all three of them. We got them running tops, which had 'BBS UK' printed on the front and 'Team Caitlin Wilkie' on the back. This kicked off the first of many running challenges that went on up in Scotland last year for our little warrior.

Next up, was the Willkie Warriors West Coast Challenge. We had a few meetings and decided on a route up the west coast of Scotland, going over to the island of Mull and around the coast before heading back across to the west coast, to Oban, to finish where we started. We won't lie; there was a lot more organisation to this than we had initially thought, organising the routes, hotels, ferry, support vans, support crew, spares and repairs and nutrition. We managed to set off at the end of April and completed the 400 kilometre route in three days. We didn't check the elevations when finalising the route, which was a schoolboy error. It was enjoyable, fulfilling, tiring, emotional above all and completely worth it.

Culloden to Clova was a 100-mile walk by a group of 15 people from our hometown. Before Culloden to Clova took place, we didn't know these 15 people terribly well, but they got in touch to say they had a fundraising idea up their sleeve. A lot of

fundraising went on before they actually did the walk including various quiz nights and bucket shaking. We travelled up to Inverness to see them off and it was extremely sweet that Caitlin stood on the start line and gave every single one of them a hug; they were all in floods of tears before they even started their challenge. They did it admirably and they brought in over £5,000 with their efforts for us.

We were extremely touched by a pensioner who had heard about Caitlin's story through her friend and got in touch. Every year she does this 26-mile walk around Loch Leven. She wanted to do it for Caitlin and for BBS because Caitlin's story had touched her so much. They raised £1,000. One of Caitlin's nursery friend's mum, Emily McDonald, decided she was going to do the Edinburgh half marathon, not just for the BBS but also for Down Syndrome on behalf of her sister. Hugely touched by this, we offered her any support we could, and she ended up completing a half marathon in a fantastic time. She got about £300 for each charity, which was an amazing effort. Again, we were really touched by her efforts.

Our local car club have a fundraiser every year and it's usually in the form of a race night; they made £1,000 which they decided to dedicate to BBS. A local tractor run (a group of farmers who own vintage tractors and go for a drive donating a few pounds in the process) raised £500 from entries. The owner of the farm where it started and finished decided to double this, taking the total up to £1,000. A local fire station had a cheese and wine night and again, just with Caitlin's name being out there and the town talking about BBS, they got in touch to donate their £600 proceeds. One of Ashley's bosses, Gavin Durston competes in multi-terrain running. He signed himself up for the Cape Wrath Ultra, 400 Kilometres in total and he raised over £1,400. The local young farmer's club held a race night and raised £1,000; they also had a ball with charity auction which raised £4,000 for BBS UK. A stall at a local music festival raised £1,000; the list is endless, all evolving from great community spirit and people just hearing our story and getting in touch.

Over the year, there have been numerous friends and family and colleagues who've taken part in runs, ranging from 5K to half marathons. We tried to support everyone by getting them BBS UK t-shirts and ensuring they got publicity for their runs. Over the course of the year, these runs have raised a huge amount of money and just as importantly raised a huge amount of awareness in all the towns around us.

This brings us to the Dundee Kiltwalk which offers three distances for people to walk: 6 miles, 11 miles, or the full 25. We decided to advertise this around friends and family and see if anyone was interested as it literally is just a walk, which all abilities can do. We were staggered when we ended up getting 72 people to sign up for our team. Liam managed to get good sponsorship from a client at work who bought the t-shirts and printed them all. We had everybody kitted out in all colours of BBS UK t-shirts, which was amazing. The walk itself was tough as we chose the 25 miler, however seeing so many BBS UK t-shirts walk around was great. The event itself raised a huge £18,000 in total for us, between the 72 people in our team. The best fundraiser of the year without a doubt and a brilliant event and definitely one we'll be aiming to try and do again this year or next year.

The Great North Run was in September last year. The run means quite a lot to me and Ash as we did it a few years earlier for the Scottish Association for Mental Health after we lost our stepdad to suicide in 2012. We managed to get a place for 2018 and got a lot of publicity for this from the local papers. This helped spread awareness and spread the story, all of which encouraged more people to donate to the cause. The event itself was hard as it was extremely warm on the day but we got around in just over two hours and beat Ollie Murs!

We were extremely lucky at Christmas time that we had donations from people who decided not to send Christmas cards, but instead donate to BBS UK what they would normally spend on Christmas cards. We were completely humbled again when a pensioner that lives in our town who doesn't have any family of her own got in contact; every year at Christmas, she donates £500 to charity. We were quite blown away by this because, again, it was a pensioner that didn't really know us, didn't know Caitlin, but having read our story decided that she was going to donate to our cause. We thought that was a really lovely thing to do.

Conference Sunday brings our fundraising to a bit of an end. We don't think we'll ever stop fundraising for BBS UK, but we had to bring these 18 months of 'big' fundraising to an end, as we have got two little girls who need mummy and daddy around. Tomorrow, my good friend Emma Mitchell and I (Ashley), will run the London Marathon. It's a bit of a bucket challenge tick for us both and we both hit 40 this month. BBC Sport have been in touch, so hopefully we will get some coverage, which, if it happens and they do their montage, will raise amazing awareness for BBS UK.

We always wanted to sign off in style so we decided on an end of year ball, a chance to dress up, have some food, a few drinks and dance to celebrate what's been a tough but fantastic year. Even though it wasn't a fundraiser, we managed to do a small raffle and auction which brought in about £4,000 on the night, which was immense. It was a fitting finale to the year and one that will stay with us for a long time.

In summary, we, obviously, have had a fantastic year of fundraising. We set out to raise £5,000 for BBS UK and we are currently sitting, with everyone's kindness, at around about £79,000 that has come in to date. We've enjoyed every minute of our fundraising and we hope that we've maybe inspired some of you to perhaps think about having some little fundraisers of your own,

because it's just amazing what a tiny little fundraiser can escalate into, as we have learned over the past year; and we will be delighted to offer any help, assistance or advice if needed."

BBS UK Conference Weekend Evaluation Summary 2019

Thank you to everyone who completed an evaluation form during and after the weekend. If you would like to submit comments and haven't already done so, please email rebecca.perfect@bbsuk.org.uk.

Percentage of guests who were 'happy' or 'very happy':	
Booking Process:	100%
The AGM:	96.7%
The Saturday morning programme:	100%
Workshops:	100%
Evening Entertainment:	97%
Breakfast/Accommodation:	90%

Percentage of our younger guests who were 'happy' or 'very happy':	
Friday Evening Activities:	100%
Drayton Manor:	100%
Food at the Park:	100%
Food at the Hotel:	100%
Saturday Evening Activities:	100%
The Quiz and Raffle:	93.4%

First conference - really felt welcome and looking forward to next year already

Too many workshops run at the same time

Some of the speeches went on too long

Keep doing what you are doing

Dr Beales – informative, interesting, could listen to him speak all day…

So lovely to hear so many personal perspectives

Great to hear how much BBS has come along

I love everyone being together and it's one of the only times of the year I don't feel myself or my daughter are judged

I hear nothing but good feedback from delegates and a professional told me it was the best well organised conference they had been to stating they attend quite a few

RNIB – What an inspiration, very motivating

CONTACT DETAILS

BBS UK General Contact
Rebecca Perfect
07543 165804
rebecca.perfect@bbsuk.org.uk
info@bbsuk.org.uk

BBS UK Clinics Family Support Officer: London Service
Angela Scudder
07591 206788
angela.scudder@bbsuk.org.uk

BBS UK Clinics Service Manager
Tonia Hymers
07591 206680
tonia.hymers@bbsuk.org.uk
info@bbsuk.org.uk

BBS UK New Families Contact
Claire Anstee
After 5pm, Monday - Friday
01604 492916
claire.anstee@bbsuk.org.uk

BBS UK Fundraising Officer
Christine Saxon
07512 198484
christine.saxon@bbsuk.org.uk
fundraising@bbsuk.org.uk

BBS UK Clinics Family Support Officer: Birmingham Service
Amy Clapp
07591 206787
amy.clapp@bbsuk.org.uk

BBS UK Assistant Support Officer
Liddy Vincent
07568 601973
liddy.vincent@bbsuk.org.uk

BBS UK Adults Contact
Stefan Crocker
stefan.crocker@bbsuk.org.uk

BBS UK Adults Contact
Rachael Foley
rachael.foley@bbsuk.org.uk

BBS UK

www.bbsuk.org.uk


BS UK