BBS UK Conference Report 2018

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Foreword

Welcome to the 2018 Conference Report which celebrates and reports on the all-round success of the Bardet-Biedl Syndrome UK (BBS UK) Family Conference. It is a useful summary for those who attended and helps to inform those who were unable to attend. Regular attendees anticipate this fantastic weekend with such joy and this year we were once again blessed with gorgeous sunny weather and high temperatures.

The committee are rightly very proud of the Conference; the aims and themes that were planned to run through the weekend directly reflected a key aim of the Charity, which is to provide further knowledge of BBS and to maximise the lives of those affected by the syndrome.

The theme of the weekend was 'Health and Well-being'. Logan Gray from British Blind Sport provided VI sports taster sessions across Friday and Saturday, Christian Bolton-Edenborough spoke about his experiences as a visually impaired athlete, we learned about the effect of exercise on the brain and we also had a dietary workshop.

We were fortunate that Dr Helen May-Simera brought some of her lab team along who provided hands on DNA extraction workshops which was great fun. Emma Oates and Laura Dowswell spoke about their visit to Helen's lab in Germany in 2017 which provided a lovely human connection for the hardworking scientists and researchers.

The committee would like to thank all of the professionals who give 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 fabulous Conference Report. Full audio recordings of the presentations can be accessed via the BBS UK YouTube channel. All that is left for me to say is bring on Conference 2019 which is to be held across the 26th-28th April. I look forward to catching up with friends old and new.

Stefan Crocker
Vice-Chairman, BBS UK

Bardet-Biedl Syndrome UK
Annual General Meeting Minutes
The Hilton Hotel, Northampton, 21st April 2018, 9.30am

Welcome

BBS UK Chairman, Richard Zimbler, introduced himself and welcomed delegates to the 2018 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, Observer; Margarita Sweeney-Baird, Trustee; Christian Bolton-Edenborough, Observer; Stefan Crocker, Vice-Chair.

Apologies

There were no apologies received from the Board of Trustees.

Minutes of the 2017 AGM

The Minutes of the last AGM of BBS UK, held on 22nd April 2017, 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:
The Chairman's Report was read out on behalf of the Chairman, Richard Zimbler by Dianne Hand.

'The Charity has had another busy year planning, organising and supporting in 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 of 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 valued talks to our members, the delegates who attended and made the 2017 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 whilst staying at their hotel. The 2017 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 contribution to the Charity over the last year. Since last year's AGM, Emma Oates has stepped down from her role as Fundraising Co-ordinator and would like to continue as a trustee in the upcoming year and I would like to thank her for the work she has done. Margarita Sweeney-Baird is retiring from the committee and I would like to thank her for all her hard work for the Charity and wish her well for the future. Since becoming Chairman I have valued my role and feel my confidence has grown step by step. I would like to continue in this role and develop my skills and attend events to raise awareness of BBS.

The Charity had its second Adult Social Weekend, and this was attended by 25 adults and 6 support staff and we are already planning to organise our third Adult Social Weekend for later this year. The Family Activity Weekend has been rearranged for June 2018 and will be 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 BBS and this has entailed attending various events such as the Rare Disease Day in London, Sight Village in Birmingham and Manchester and Tonia represented the team at the Cambridge Rare Disease Summit. The Charity has now formed a Scientific Advisory Board. This is to provide greater support in the research of BBS and will be a useful mechanism to assist and collaborate with others in the national and international communities. We value and welcome the Board's founding members who are Professor Phil Beales, Dr Elizabeth Forsythe and Dr Helen May-Simera and we look forward to working more closely with them.

Our financial position needs to continue to strengthen and there has been a lot of hard work in developing fundraising initiatives. We have made good progress with the CIO conversion and at the end of last year, the Board of Trustees decided to recruit an administration officer to support the Charity with its governance and projects.

The clinics are being well attended and numbers continue to grow. I would like to thank all the volunteers and clinicians at all of the hospitals for the continued smooth running of the multi-disciplinary clinics. I would also like to thank Tonia, Angela and Amy for the smooth re-structure of the support services and for all their dedicated support to the Board of Trustees and charity projects as well.

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

Treasurer's Report

'Income during 2017 was just over £51,000, compared to almost £64,000 in 2016, which included a restricted grant of £5,000. Expenditure during 2017 was just over £40,000 compared to almost £31,000 in 2016. Therefore during 2017, we can see that income has reduced and expenditure has increased.

During 2016, BBS UK received £29,000 in donations and fundraising, however, in 2017, this would fall to just over £20,000. Similarly, in 2016 we had marathon runners who raised almost £6,000 compared to just £50 in 2017. 2017 has seen bike rides raise a total of £6,500.

Income from our Friend's scheme has increased. This is an important income stream for BBS UK as it is the regular monthly amounts that helps the Charity to plan ahead. Our thanks to all those who have signed up to this scheme.

Expenditure remains well controlled although the cost of the annual Conference is the largest item of expenditure at £14,500 and naturally the costs rise each year. Other costs in 2017 include the purchase of new promotional stock merchandise and the costs associated with the CIO conversion. As of the 31st of December 2017, the Charity had total funds of £82,101, of this, £13,648 is restricted and relates to specific grants which will be spent in 2018.

A realistic and prudent budget has been set for 2018 which will enable the core activities and objectives of the Charity to be carried out, but as ever, the 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. We do have a reserves policy in place and this need remains and is one of our priorities. Apart from the grant which is 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 who freely give up their time to fundraise. As mentioned, total funds as at the 31st of December 2017 are £82,101. If we take off our restricted funds and designated funds we are left with just £27,310 at the beginning of this financial year.'

Approval of the 2017 Accounts

The vote to approve the 2017 accounts was undertaken, everyone was in favour and no one apposed 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, and everyone was in favour and no one apposed the Independent Examiner resolution.

CIO Conversion

BBS UK is an unincorporated charity and the Board of Trustees is working towards becoming a charitable incorporated organisation. Information was circulated to all members with the AGM invites to inform them of the process and answer any questions that they might have. The draft CIO constitution was made available to members on the BBS UK website prior to the AGM. No questions were raised by the members.

BBS UK members were asked to vote on the following resolutions:

- To authorise Trustees to dissolve the existing unincorporated charity in accordance with clause 14 of the BBS UK constitution once the registration of the new charitable incorporated organisation has been approved by the Charity Commission. A show of hands informed everyone was in favour and no one against.
- To authorise Trustees to transfer the assets and liabilities of the existing unincorporated charity to the new CIO following approval. A show of hands informed everyone was in favour and no one against.
- To accept the terms and conditions detailed in the draft CIO constitution and to authorise Trustees to apply to the Charity Commission for registration as a CIO on the basis of this new draft constitution. A show of hands informed everyone was in favour and no one against.
- To authorise Trustees to approve any further changes to the draft constitution which may be required by the Charity Commission following submission in order to achieve registration of the CIO. A show of hands informed everyone was in favour and no one against.
- To authorise Trustees who are in office at the time of registration to serve as Trustees of the new charitable incorporated organisation until they are due to retire by rotation in accordance with clause 15 of the new draft CIO constitution. A show of hands informed everyone was in favour and no one against.

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 Margarita Sweeney-Baird therefore retired during the AGM.

Of the current committee members:

- Margaret Sweeney-Baird decided not to stand again for re-election to another term.
- Dianne Hand had a further year to serve of her current term.
- Emma Oates was eligible to stand for re-election to a third term.
- Rachel Foley was eligible to stand for election after observing for a year.
- Christian Bolton-Edenborough was eligible to stand for election after observing for a year.

A show of hands informed everyone was in favour and no one against the election, re-election and retirement of Trustees.

Of the current officers:

- Richard Zimbler was nominated and eligible to be re-elected as Chairman.
- Stefan Crocker was nominated and eligible to be re-elected as Vice-Chairman.
- Laura Dowswell was nominated and eligible to be re-elected as Treasurer.

- Abbie Geeson was nominated and eligible to be re-elected as Secretary.

No other nominations had been received for these positions and a show of hands informed everyone was in favour and no one against the nominations, elections and re-elections of the Honorary Officers.

Observers

BBS UK has three trustee vacancies, members are requested to observe on the Committee for twelve months prior to being nominated to stand for election. If any members are interested in joining the Board of Trustees, and would like further information please contact Abbie Geeson, BBS UK Secretary: abbie.geeson@bbsuk.org.uk.

Any other business

Richard Zimbler welcomed Laura Bennett as the new Administration Officer for BBS UK.

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

Endocrinology Issues in BBS
Professor Jeremy Tomlinson
Professor of Metabolic Endocrinology and Consultant Endocrinologist
University of Oxford

Professor Jeremy Tomlinson is a Professor of Metabolic Endocrinology and Consultant Endocrinologist based in the Oxford Centre for Diabetes, Endocrinology and Metabolism (OCDEM), University of Oxford. His research tries to understand the causes of conditions including insulin resistance, non-alcoholic fatty liver disease and polycystic ovary syndrome focusing in particular on the role of natural steroid hormones and their metabolism. More recently he has begun to explore the specific metabolic changes that occur in patients with Bardet-Biedl Syndrome (BBS).

Alongside his research, Professor Tomlinson works as a consultant endocrinologist within the Oxford Centre for Diabetes, Endocrinology and Metabolism (OCDEM) seeing patients with a broad variety of endocrine conditions including a dedicated non-alcoholic fatty liver clinic that he runs jointly with liver specialists. He has provided metabolic and endocrine input to the Bardet-Biedl Syndrome clinic at the Queen Elizabeth Hospital in Birmingham for many years, since it was first established. In collaboration with colleagues from London, Professor Tomlinson has helped to redefine and provide the most accurate and up-to-date description of the metabolic and endocrine conditions that are associated with BBS. Professor Tomlinson's presentation can be downloaded in full from the BBS UK YouTube channel.

Professor Jeremy Tomlinson began his talk by explaining that an endocrinologist is a doctor who studies hormones. A hormone is a medical term for a substance that gets produced by a gland in the body. They are signals produced by one part of the body that affect another part of the body.

Endocrinologists study those hormones and are interested in the clinical effects of hormones on organs in different parts of the body. Professor Tomlinson took the audience on a whistle-stop tour through some of the endocrine glands.

The pituitary gland is the master gland in the body, sitting at the base of the brain behind the eyes. It controls every other gland in the body and, amongst other things, tells the ovaries and testes how to function. It also controls stress hormones.

One of the commonest glands that endocrinologists focus on is the thyroid gland which sits in the neck like a bow tie; the thyroid gland controls the rate at which we all function.

The pancreas sits deep within the upper part of the tummy and produces insulin amongst other things, which is very important for controlling blood sugar levels; when it doesn't work well, diabetes can develop.

Diabetes is very common, affecting about 3 million people in the United Kingdom, a figure that has increased dramatically over the last ten to fifteen years. Diabetes is a condition whereby individuals can't regulate their blood sugar levels properly. There are two different types of diabetes, type 1 and type 2. The most common type of diabetes in the UK is type 2 which is rapidly becoming one of the UK's biggest health problems.

Professor Tomlinson briefly explained the mechanics of diabetes. When we eat, sugar or glucose from food enters our bloodstream and is delivered to our pancreas which, if it senses a high level of blood sugar will respond by producing the hormone insulin. Insulin produced by the pancreas will signal to all sorts of other tissues that action is required, in particular the muscle. The insulin allows sugar from the blood to get into the muscle which we then use as fuel, a bit like the petrol in a car. Without insulin, the sugar stays in the blood and can't get into the muscle, which means the muscle can't work very well and our blood sugar levels get very high which is essentially diabetes.

Type 1 diabetes is where the pancreas just doesn't produce enough insulin. It is much more common in childhood, but is pretty uncommon in patients with BBS. Type 2 diabetes tends to occur more in adults and for whatever reason, when the insulin from the pancreas tries to signal to the muscle to allow the glucose (sugar) to get into the muscle, it just doesn't work.

Professor Tomlinson stated that the first treatment for patients with diabetes is lifestyle modification. Our bodies struggle with a diet that is very high in sugars and as weight is gained, it becomes harder for the insulin molecule to work. A combination of a healthy balanced diet, exercise and drugs is advocated when dealing with diabetes for the general population and Professor Tomlinson said that the approach is the same in patients with BBS. Approximately 15%-16% of patients with BBS have type 2 diabetes, very few have got type 1 diabetes. Around 25% of the patients who have diabetes are managed with diet and lifestyle alone, about 40% use insulin and about 30% are prescribed the drug, metformin. Metformin is the most common drug used for the treatment of diabetes and it works by helping our own body's natural insulin work better.

Professor Tomlinson continued, "Weight is a problem in the UK and across the world. In London, about 60% of the population are either overweight or obese, whereas in the north of England it may be closer to 70%. A relatively small number of BBS patients are able to maintain a very healthy body

weight and around 80% or 90% are either overweight or obese and there are reasons as to why patients with BBS might accumulate more weight than others. This is an area of active research, and what we know so far is the fat cells in patients with BBS behave very differently from patients that don't have BBS, in the sense that they may be more well-programmed to store fat in comparison with the general population."

Professor Tomlinson explained that patients with BBS appear to be at a slight increased risk of developing metabolic syndrome, a cluster of conditions including cholesterol levels, blood pressure and fat levels in the blood. 54% of Birmingham and London BBS clinic patients showed evidence of metabolic syndrome whereas just 26% of the control-matched group, a group that didn't have BBS, matched for age and weight, showed evidence of having metabolic syndrome.

Professor Tomlinson moved on to talk about other hormone involvement in BBS, starting with thyroid hormone produced by the thyroid gland, mentioned earlier on, which controls amongst other things, the rate at which we function. If the thyroid is overactive and produces too much thyroid hormone, we may be overactive, be unable to sleep, sweat a lot, experience heart pounding and weight loss. If the thyroid is underactive and doesn't produce enough thyroid hormone, we may slow down, sleep a lot and experience weight gain. The vast majority of patients with BBS (77%) have entirely normal thyroid function, about 20% have mildly underactive thyroid function and about 7% of patients have a properly underactive thyroid function; underactive thyroid function can benefit from and be treated with a replacement thyroid hormone.

Professor Tomlinson has a particular interest in polycystic ovary syndrome (PCOS), both clinically and from a research perspective; it is the most common endocrine hormone condition seen in BBS clinics, affecting 15% of female BBS patients; in the general population the figure is 5%-10%. PCOS affects periods and the ability to conceive, it causes an imbalance of hormone levels and little cysts can be seen on the ovaries. Those affected may experience difficulty losing weight, facial hair growth, hair loss on the head and oily skin, which can all have a dramatic effect on mood. Professor Tomlinson stated that it is commonly reported in textbooks (Williams, Oxford etc.) that menstrual irregularities are common in virtually every woman with BBS, as is infertility. He reported that the data established from the specialised BBS clinics across the country proves that to be 'absolutely not true'.

Professor Tomlinson continued, "Okay, let's talk testosterone… women and men produce testosterone and testosterone has lots of important roles, it's not just about bodybuilding and sexual function. Testosterone is important for maintaining bone health, it helps to reduce fat mass and to increase muscle mass, which is why it gets abused by athletes. Not having enough testosterone can cause medical problems however about 80% of patients with BBS have normal testosterone levels. The Williams and Oxford textbooks were saying that low levels of testosterone are pretty much across the board in patients with BBS but we now know that simply isn't true; only 20% have a low testosterone level, and within the patient group that we see across the country with BBS, several have been able to father children. Infertility is not across the board in patients with BBS and I think what's really important is, don't believe all that you read in textbooks and don't believe all that you're reading on the internet. We've now seen several hundred patients across the clinics and we learn from our patients. There are different aspects of the endocrine system that can be affected by

BBS and no one patient's BBS is the same as anyone else's, which is why it's important that as clinicians we make an individual assessment of each patient."

Professor Tomlinson said that a key take home message, is that if an endocrine condition is picked up, there are things that can be done, hormones and treatments that can be used to help with the condition and address the symptoms that patients are suffering with. He continued,' One of the things that we do need to begin to understand is what happens with those hormones over time. I think the beauty of the clinics in London and Birmingham is that we can track the natural history of these hormones, because as we all get older, even outside the context of BBS, hormone levels change, but is there something about BBS that affects how those hormone levels change over time that we just don't know about.'

Professor Tomlinson finished by thanking all those involved in bringing the data together including Dr Elizabeth Forsythe, Professor Phil Beales, Dr. Barbara McGowen, Dr. Bobby Huda as well as the Kings Diabetes research group and Dr Jonathan Hazlehurst who helped collate the Birmingham side of the data. His biggest thanks were to those living with BBS; he said, "At the beginning, 10 years ago, I confess, what I knew about BBS was what I read in textbooks and I think we've moved a long way in terms of what we understand about BBS and hopefully we can, using this data, really make the management, from an endocrine perspective, a little bit more of a 21st century approach to managing hormone issues in Bardet-Biedl Syndrome. "

British Blind Sport
Logan Gray
National Partnership Manager, British Blind Sport

Logan has 10 years' experience of working in the sport sector across a number of sports and this is allied to his experience from coaching, volunteering, competing at international level, and sports commentating. He joined British Blind Sport looking for new challenges and to give something back to sport. Logan's presentation provided an overview of British Blind Sport services, information about the benefits of regular sport and physical activity on people's lives, health and wellbeing as well as information about upcoming events and how to find local accessible and inclusive activities; it can be heard in full on the BBS UK YouTube Channel.

Logan introduced himself and explained that the main aim of British Blind Sport is generally about getting people with visual impairments active, by whatever means. Logan began his presentation with some audience participation; he asked all those in the room who consider themselves to have an active lifestyle to stand up – around half of the delegates rose to their feet. He then asked everyone to stand up according to how much exercise they thought the Chief Medical Officer recommends daily for children aged 5-18 years; the majority were correct at 60 minutes per day. Finally, delegates were asked to think about how many minutes of moderate exercise is recommended for adults per week – most people thought it was 250 minutes when surprisingly it is much less at just 150 minutes per week.

The Chief Medical Officer recommends five 30-minute sessions per week; any activity lasting 10 minutes or more can count towards that goal. Exercise and activity should be about building strength, improving balance, working our heart and lungs and our muscles and bones, which in turn will improve our quality of life. Being active means working hard enough to get a bit warmer and get our heart rate going a little bit faster; It's not about running marathons or running the 100 metres as fast as we can, it can be as simple as walking at a brisk pace.

A BBC headline last year said 'Middle-aged Adults Need to Walk Faster'. According to Public Health England, everyone was just dawdling around and if they just went a little bit faster, then they'd be able to contribute to that 150 minutes they need to maintain a general level of fitness. Not only are there health benefits, both physically and mentally, exercise improves our quality of sleep, maintains a healthy weight, manages stress, increases productivity and generally improves overall quality of life.

British Blind Sport did some research in 2014, looking at barriers to participating in sport and physical activities for people with visual impairments. The main motivations for participating in sports were the health benefits and to meet people and make friends; social isolation is a real issue.

Being active doesn't always have to involve sport, Logan suggested we could join a group fitness class or a dance session for older people; gentle exercise is becoming more and more popular. Or people might enjoy group walks; Logan recommended 'Walking for Health' as a great organisation in this respect.

British Blind Sport's mission is to make a visible difference through sport. They have only got nine members of staff and have about a thousand members. Behind the scenes, British Blind Sport staff advocate for the visually impaired community working with sporting organisations to become more inclusive. They have produced an e-learning module, for coaches and leisure centres, to help them be more inclusive and confident working with visually impaired individuals. British Blind Sport also work with organisations like Bardet-Biedl Syndrome UK to provide experiences and develop opportunities.

Logan explained that he works with about 12 to 15 different sports, which are the ones that the visually impaired community say they especially want to take part in. One of their main focuses, at the moment, is their local 'Have a Go' taster days, which are set up like a roadshow, travelling around the country. British Blind Sport have a database of around 1,400 clubs that they think should be offering a positive opportunity to take part in sport, so those who live in a certain area and are interested in a certain sport should get in touch with them and they will help to make it happen. One of the main areas they are targeting is people who don't already take part in sport or physical activity.

Logan went on to talk about other British Blind Sport projects.

'See My Voice ' is a new project that gained funding last year. It is a three-year project aimed at getting young people, aged 11 to 18 years, into volunteering opportunities to deliver social action and develop leadership qualifications. Although they have already recruited for Year 1, they will be recruiting again towards the end of summer 2018 for Year 2. British Blind Sport want to speak to young people directly, rather than via their parents and teachers, to find out what they want to do

and work out how British Blind Sport can help to facilitate that and give them a better chance of employment when they come to their later teen years. It is a one year term and involves up to 50 hours of volunteering within the year. If the young person does everything that is planned, they should get a leadership qualification at the end. Logan explained that the project will provide the opportunity for young people to shape their own future, because they will feed back their experiences to the sports sector decision makers, to become more inclusive for visually impaired children and young people in the future.

'First Steps' is a project aimed at children aged 3-11 years, to develop their physical literacy at an early age. Participants will receive a box through the post containing a pump, inflatable ball and an activity booklet full of exercises that a family can work through with their children. The aim is that they will gain the confidence to access more mainstream sports clubs and activities. British Blind Sport have completed a pilot in the West Midlands and in Scotland and are trying to access funding that will allow them to roll the project out nationally.

'Find a Guide' is a project aimed at developing a database of sighted guide runners that are trained and licensed. Visually impaired runners enter their postcode and the database will tell them who is nearby and able to provide guide services for a run or a fast walk; it's not about finding a training partner for a marathon, but about getting active and moving in the right direction. The website, www.findaguide.co.uk has recently been relaunched and upgraded.

For more information go to www.britishblindsport.org.uk where you will be able to subscribe to receive newsletters. You can also follow British Blind Sport on Facebook and Twitter.

Adapting and Perseverance: A Personal Perspective
Christian Bolton-Edenborough

Christian is from Potters Bar, Hertfordshire. He was diagnosed with Bardet-Biedl syndrome (BBS) in early 2015 whilst studying for a LLB Law degree in North Wales. He graduated in 2015 achieving a 2:1 and then a distinction in a Law Master's Degree in 2016. Christian is currently studying the Legal Practice Course at BPP University London and aims to become a solicitor in Family and/or Property Law in the future. He competes in the Tennis Foundation's Visually Impaired UK Tennis Championships, and from October will be studying to become an LTA Level 1 tennis coach. The following is an edited extract of Christian's presentation at the BBS UK Family Conference. The full audio recording of his talk can be downloaded from the BBS UK YouTube Channel.

"From birth and even before, I have had to persevere. Doctors told my parents that kidney cysts and tumours showing on their machinery meant that I wouldn't live long. Those findings thankfully disappeared not long before birth and here I am nearly 24 years later.

From a very young age, I have been heavily involved in sports and I want to thank my parents for that. They were really interested in getting me involved in sports from a young age, not only because of the weight aspect but also for the mental benefit, it is greatly important to be active early

on. My mum threw me into football, I went cycling and they had me playing tennis every week. From age 6 to 12, I was doing some sort of sports every week, just remaining very active.

When I was around 11 years old, the doctor said that an issue I had with my foot (due to the removal of an extra toe) would mean that I would be in a wheelchair by the age of 18 and that I wouldn't be able to continue playing sports. He suggested a joint fusing operation but I didn't want that, I didn't believe it was feasible. So I carried on, I persevered, and they turned out to be wrong. Sometimes you have to believe in your own instincts.

As my teenage years progressed, I'm not sure why, but the sport tailed off and I put on a lot weight. I moved schools at 16 and threw myself back into sport. I may not have had the most talent when it came to sport but I had a real interest, I thought, 'If I throw myself into it, hour after hour after hour, let's see what happens'. From age 16 to 18, I put in 10 to 12 hours every weekend. I'd be at the tennis court just learning, picking up what I'd seen on TV, trying to adapt what I had learned from an early age. I started university in 2012 and was selected for the tennis team trials, I'm thinking, 'Oh, this is great, I could play university tennis, this is a big stage to get myself on to'.

Unfortunately, something hindered me at the trials and continued over the next two and a bit years. I was just getting beaten left, right and centre by players who were of a similar ability and I couldn't understand it, to the extent that I was going to give up on the dream of playing competitively. It was a couple of years later, we found out why - I've got BBS and I have perhaps around 30% central vision, putting me at a greater disadvantage to sighted players. I carried on, I tried to adapt my game, but have since found Visually Impaired Tennis where I am able to compete with my perseverance and prior adaptations on an equal level.

The diagnosis in 2015 changed my outlook in terms of my approach to every day by not taking things for granted. It has strengthened my resolve and given me leadership and initiative qualities I did not know I had. This has given me further motivation as I know that through perseverance and hard work, I can attain my career goal of becoming a qualified solicitor and achieve my sporting aspirations. I hope that in achieving these aspirations, I can encourage further BBS members to keep trying to achieve their dreams, whatever they may be.

After my first BBS UK Conference in May 2016, I represented the Wales University Pool team at the Universities Home Nations Championships. I was, and remain, the first visually impaired player to have participated in the tournament. I was able to compete against and beat some of the best fully sighted university players from across the United Kingdom. It was a proud achievement.

I want to be a World Champion of visually impaired tennis, I've got aspirations. I want to go as far as I can and having BBS, as I previously said, has opened the potential for me to do so. This is because before, I'm thinking I was just mediocre at sports, whereas now, I've got the focus to train. I loved exercising in the past but I now have this goal to work towards because of being visually impaired.

I would like to finish with a quote and it's from Nelson Mandela; I think it's quite apt for us:

"I'm the master of my faith. I'm the captain of my soul."

I feel that it is fitting for those of us with BBS because we can do whatever we want, with the right support and if we work hard. You have to work with your limitations and that's what I've done. I'm not trying to create an image that it will be easy, but by initially attempting those activities you will ascertain what your specific limitations are and once you have done that you can then find ways to get around them - with perseverance. It will take a lot of effort, but if you're committed to doing something, if you really want to do something, you will find a way and there are these groups, like British Blind Sport that will help you."

Conference Fundraising

As you all know, fundraising is absolutely vital so that our Charity is able to put on events and provide support for anyone affected by BBS. The more we can raise, the more we can do and the fundraising team would like to say a huge thank you to everyone who contributed to the fundraising efforts over the Conference Weekend.

Heartfelt thanks go to knitting heroes, Sandra Dale and Janet Wakelin who have for the second year running, worked hard all year round knitting beautiful dolls, teddies and other animals for the very special Teddy Raffle. Their efforts have raised hundreds of pounds for BBS UK but more importantly have brought big smiles to so many faces, young and old alike. We understand that knitting is already well underway for Conference 2019 and the new collection includes beautiful elephants, cute cats and adorable dolls – thank you both so very much.

Special thanks also go to Helen Petty for the fabulous main quiz prize and Aneeba Ahmed for the special hand-made bracelets. We are grateful to every one of you who supported these fantastic fundraising opportunities over the course of the weekend because your generosity and involvement really does make a difference. Thank you.

Update on Research and Study of BBS
Professor Philip Beales
Great Ormond Street Institute of Child Health
Guys and St. Thomas' NHS Trust

Professor Beales obtained his degrees in Genetics and Medicine from University College London. He undertook postgraduate training in both general medicine and paediatrics before specialising in Clinical Genetics. In 1999, he published the largest survey of Bardet-Biedl syndrome (BBS) symptoms with the help of members of BBS UK (then LMBBS). This culminated in the identification of a number of new features not previously described in the medical literature and enabled Professor Beales to propose new diagnostic criteria, with the expressed purpose of enabling an earlier diagnosis for many more children.

In 1999, Professor Beales, with colleagues at Baylor College of Medicine in Houston discovered the first gene (BBS6) to cause BBS. Since then, at least 21 genes are now linked to the syndrome.

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 BBS. This includes the notion that abnormally functioning cilia (small finger-like appendages on cells) lies 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 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 summary of Professor Beale's presentation at the BBS UK Family Conference, the full audio recording can be found on the BBS UK YouTube Channel.

Professor Beales began his presentation with an overview of the NHS funded specialised clinics service in Birmingham and London, which he said, is growing enormously:

"The demand is outstripping capacity, and that's been a real theme and issue for us this year. One report, submitted at the end of last year for Great Ormond Street Clinic, identified a sudden jump of about a 20% increase in numbers over the 2016/17, and then the 2017/18 period. If we look at all of the clinics, we see a pretty similar figure. In other words, the whole of the service is outstripping our expectations and there are a number of reasons for that. I think part of it is clearly BBS UK, the Charity and all the good work that they have been doing in terms of disseminating the information and raising awareness amongst the medical profession as well as the general public.

We have reported this information back to NHS England and have given them the reasons why we think we might see more people within our service: greater awareness, new born children, immigration (BBS affects people from all ethnic backgrounds), but principally, greater awareness.

I'm one of those people who doesn't like to complain about things without proposing a solution, so we proposed that we need to redesign how the clinics are being delivered, so that the 'healthy' individuals, the people who need less of our input, could come to clinic less frequently. Although we're supposed to see everybody on an annual basis, by and large it has slipped to at least 18 to 20 months for some people, and that is just not good enough.

We are also going to redesign or restructure the clinics in some way, so we have asked for funding to deliver 'virtual clinics' or 'telemedicine'. Telemedicine has been around for a long time now and is used right across the world and it's something that I believe could work really well here. The good news is that NHS England have agreed to support this model going forward.

So the next thing we need to do is look at the design and make sure that we coordinate very closely with the GPs, so that patients don't miss out on the blood tests that we need to do, for example, or other investigations that might need to be done locally, before we have that particular tele-consultation with you. We will trial it, roll it out and see how it goes, and see whether it makes a difference."

Before introducing the research segment of the day, Professor Beales spoke briefly about the amount of research currently being undertaken and BBS UK's involvement:

"The area of research around BBS, continues unabated. If we look at all of the research papers just up to April 2018, there have been 35 published around the world; Japanese papers, Chinese papers, US papers, and three of the papers we've contributed to have been published this year including Elizabeth's seminal paper (Risk Factors for Severe Renal Disease in Bardet-Biedl Syndrome)."

Professor Beales finished his introduction with a thank you to BBS UK members and BBS patients, he said, "You guys in the past have been kind enough to take part in all sorts of research programmes, one of which was a skin biopsy programme. Cells taken from a little piece of skin were sent to Cambridge, where they were grown into a lot of cells, which were then turned into stem cells. The reason for this is because there is an increasing demand in the research community to have access to samples from people who have various rare conditions; it's an incredibly valuable resource. BBS patients contributed the most samples of all the diseases to the national Human Induced Pluripotent Stem Cells Initiative (HiPSci) study, a £13 million study that was funded by the Wellcome Trust and the Medical Research Council which finished earlier this year. All of those samples are now available, and researchers from all around the world can access a little bit of your material (where permission has been given) for research purposes. So I just wanted to say a big thank you, because BBS is a multi-organ disease that affects virtually every part of the body and it's very important that the research that we undertake is not only driven by you, but is focused on the various areas that affect you most."

Gene Therapy for Bardet-Biedl Syndrome
Dr Victor Hernandez
Senior Research Associate, Institute of Child Health, UCL

Dr Victor Hernandez MSc PhD FHEA is a Senior Research Associate at the Great Ormond Street Institute of Child Health at UCL. He finished his PhD at Barcelona University and came to the UK to start his research in Professor Phil Beales' Ciliary Disorders Lab in 2008. During his postdoctoral years he focused on the molecular mechanisms involved in Bardet-Biedl Syndrome (BBS). He developed and described new BBS mouse and zebrafish models, establishing the link between BBS genes and cytoskeletal function and collaborated with international partners in the discovery of new genes linked to ciliary function.

Recently, Dr Hernandez opened a completely new gene therapy project to treat BBS. The preliminary data obtained has been sufficient to secure funding from Fight for Sight and Apollo Therapeutics to further investigate this treatment potential. This support has allowed him to start a research group with the only focus being the expanding gene therapy research for BBS. The group is now developing different strategies to tackle retinal degeneration, brain derived symptoms and a complete multi-organ approach to target all affected organs, in collaboration with national and international researchers.

Following, is an edited extract of Dr Hernandez' presentation, the full audio recording is available on the BBS UK YouTube Channel.

Dr Hernandez attended his first BBS UK Conference ten years ago when he had just started working with Professor Beales. He said so much had changed during that time, in the way that BBS is understood and in the way that they approach therapies. Dr Hernandez aimed to give an overview of his research and hoped that it wouldn't be too technical.

Dr Hernandez explained that the reason people have Bardet-Biedl Syndrome is because they have a gene that is not working (mutated). If a normal copy of the gene is delivered to the cell, the cell will be able to function optimally again – this is gene therapy. The most common gene is BBS1 and the second most common is BBS10, it is these genes that research is currently focused on.

Dr Hernandez' research is focused on blindness and obesity in BBS, both of which feature in the BBS mouse models used in lab research. The team started their research three and half years ago without knowing anything about gene therapy. They started on a project developing techniques on the mouse models to see if they were able to repair damaged cells and tissues.

Their first focus was to look at photoreceptor degeneration in BBS. Their second focus was obesity, as they knew it was a malfunction of a neural network at the base of the brain called the hypothalamus, responsible for regulating hormone function and appetite. They found that this whole network was not working in BBS, which is possibly one of the reasons why those with BBS sometimes have the urge to constantly eat. Unfortunately, it's not easy to deliver a normal copy of a gene into a specific cell type because the cells have a membrane that protects them, stopping the DNA from entering the cells. The solution is to find a 'vehicle' to carry the DNA and enable it to move into the cells, and that's where the virus comes in. Dr Hernandez and team used a virus called AAV, adeno-associated virus and focused on the most common BBS gene, BBS1.

The team's first aim was to stop or delay the retinal degeneration. The retina is a layer of cells behind the eye that receives light and sends an electrical message to the brain enabling us to 'see'. In BBS, the cells that are dying in the retina are the photoreceptors and it is those cells that the team need to give a normal copy of BBS1.

There are two types of photoreceptor cells, rods, which provide low light vision and peripheral vision and which the team are working on, and cones which provide central and colour vision. Dr Hernandez explained that they need to consider two things, what type of cells to target and when; if they go in really early, they will be able to stop the degeneration of these cells for a longer time.

The first trial involved putting BBS1 into a virus and injecting it in the retina of month old mouse models. A year later the team were able to use an electroretinogram (ERG) to show that the treated mouse models retained 50% of their vision compared with the untreated models. They weren't satisfied with this however so they created another virus and targeted both one month old mouse models and one week old mouse models; so far the treated animals are keeping their vision much better than the untreated ones.

The team's second project is concerned with obesity, responding to a few publications that relate this aspect of BBS with the incapacity of the hypothalamic network and issues with the hormone, Leptin. The team injected mouse models with the virus containing the corrected BBS gene and followed them for a year; after a year, none of the models had gained any weight.

It is incredibly exciting to hear that it is possible to rescue the vision in BBS mouse models, however Dr Hernandez cautioned that they still have quite a way to go before moving to clinical trials. He said, "right now, we cannot cure BBS, but I'm absolutely, totally convinced that we are heading in the right direction, and in the future, not that many years from now, we'll be able to start producing some of these therapies for you."

The Secret to a Bigger, Better Brain

Dr Elizabeth Forsythe
MRC Clinical Research Fellow (Clinical Genetics)
UCL, Great Ormond Street Institute of Child Health

Dr Elizabeth Forsythe is a Medical Research Council fellow and clinical genetics registrar based at the UCL Great Ormond Street Institute of Child Health. She has a background in paediatrics and is a member of the Royal College of Paediatrics and Child Health.

Dr Forsythe has been working in the London Bardet-Biedl syndrome (BBS) clinics since they started in 2010, is completing a PhD in the Beales Cilia Disorders Laboratory and is a board member of the Ciliopathy Alliance.

Following, is an edited extract of Dr Forsythe's presentation at the BBS UK Family Conference; the full audio recording can be downloaded from the BBS UK YouTube Channel.

"My name is Elizabeth Forsythe and I'm a clinical geneticist. I've been involved in the BBS Clinics since they started in 2010 which means I've been really privileged to speak to lots of you and I've seen lots of children grow into young, confident adults; it's been really exciting to speak to so many of you and hear from you, your caregivers and parents about the things that are important to you.

I'm going to talk about what I've learned in the last couple of years about how we can optimize our brain function and also about the study that we completed last year.

Over the last two years, I've been learning a lot about the different structures of the brain and in particular, about how things are slightly different in people with BBS, in particular in relation to a structure called the hippocampus. We've got two hippocampi, one on each side of the brain and

they do an amazing job, storing all of our memories, everything from our drive to work, to the first time we held a baby, and when we store those memories in our hippocampus, the structure changes forever.

Sadly, in people who have the form of dementia called Alzheimer's, their hippocampi shrivel up, they become smaller as they lose their memories. We also know that the hippocampus is involved with our mood, we know that the hippocampi in people who are depressed becomes smaller and with antidepressants, the hippocampi can become bigger again.

So why am I telling you this? We know that the hippocampus is slightly smaller and a little bit unusually shaped in many people with BBS and when you think about the function of the hippocampus that makes sense, because we've heard from BBS patients that they may be struggling with school, to learn new skills or have issues with mood.

Here's the good news. We used to think that the brain is what it is, you've got the brain you've got and there's nothing really you can do about it. Now we know that actually there are two parts of your brain where you can get more neurons, more nerve cells: one of them is the olfactory bulb, which has to do with our sense of smell and the other is the hippocampus.

The average brain has around 100,000 billion neurons and in the hippocampus alone, we make 700 new neurons every day and that may not sound like a lot in the grand scheme of things, but it does mean that by the time you turn 50, you have a new hippocampus. Eating more healthy food, fruits and vegetables, reducing our alcohol intake, learning new skills, exercising more, reducing stress and improving sleep quality are all things that will improve our turnover of cells. This is a list for all of us to follow, not just those who have BBS, because for all of us as we age, the turnover of cells goes down.

I want to tell you a little bit about the study that we started doing in 2016. We felt inspired to look at how we could improve the size and the function of the hippocampus, in particular in people with BBS. Our research is based on the following facts:

- We know that in people with BBS, the hippocampus on average is a little bit smaller and a little bit unusually shaped.
- We know that in people with Alzheimer's, their hippocampi get smaller as they lose their memory, but we also know that you can slow that decline with exercise.
- BBS Mouse Models that exercised on spinning wheels had more dense nerve cells in the hippocampus than those that were less active. (Dr Sonia Christou-Savina)

In 2016, we invited a group of children with BBS into Great Ormond Street Hospital for an MRI scan of the brain and for an assessment to check their memory and fitness. The children were divided into two groups and they went home and followed a programme: half carried on with their normal routine and half worked really, really hard with a personal trainer. We're incredibly grateful to the families who did this because we realize that it's not a small undertaking to have to take your child to see a personal trainer once a week when you've got other children and work, so we're very grateful for that.

The fantastic thing was that when we re-tested, we found big changes in the children who were fitter. Not only did the children who were fitter have better blood flow to their brains, which is a good thing, they had bigger hippocampi and were getting better at the memory game, which is a really hard game by the way. What was particularly exciting for us about this was that although we knew already that you can improve the size of your hippocampus and you can improve your memory and so on through exercise, we didn't know that if you were born with a small hippocampus, that

you could still make a positive difference. We are hoping to do this on a much bigger scale and involve many more BBS patients.

Another positive side effect is that lots of people were feeling more confident, lots of people came back to us and said, 'I feel more physically confident, I feel more confident to take on sport, I feel more confident to go into a gym.' In particular that was true for the people who had embraced it as a family; we know that the biggest effects of lifestyle change, especially involving children, happen if you can embrace it as a family.

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

I want to leave you with one thought which is this: if a drug company was able to distil the effect on your brain that exercise has, they would be laughing, because there is no tablet that can do for you and your brain what exercise can, exercise really is the key to having a bigger, better brain."

Dr Forsythe invited questions from the audience and a delegate had this to say:

"I want to support and confirm what Dr Forsythe said about the sports activity and how much it can change children with BBS… … I have two boys with BBS, my youngest son, he was extremely obese, he was over 115 kilos at the age of 12 and had challenging behaviour. You cannot deal with him, you cannot reason with him and he's constantly eating so much… … He decided to go for a healthy diet, eat fruits and vegetables, he drinks only water and he exercises every single day. We got him some equipment like an exercise bike and he goes for walks. Now, he behaves, he's thinking, he's reasoning more, he's extremely honest and polite to people, he's lost over 15 kilos; it's just like he's a new person.

My message to all parents, new parents that have children with BBS syndrome, they need to start from an early age. I didn't know that, I didn't know the sport and the activity would help so much… …I'm extremely proud, I'm surprised and shocked."

Personal characteristics, mental health and well-being in BBS: A new research project

Dr Jane Waite
Lecturer in Psychology, Aston University, Birmingham

Dr Jane Waite is a Lecturer in Psychology at Aston University and a Clinical Psychologist. Dr Waite completed her PhD at the University of Birmingham, which focused on the behavioural and cognitive phenotype of Rubinstein-Taybi syndrome. Following her PhD, she continued her research into behaviour and cognition in rare genetic syndromes including Williams, Kleefstra, Lowe and Bardet-Biedl syndromes.

Dr Waite's current research focuses on understanding the development of mental health difficulties, and improving the identification of these difficulties, in people with intellectual disabilities, rare genetic syndromes and autism spectrum disorder. She is currently developing clinical assessment

tools for use within the NHS. Dr Waite has also worked extensively on online resources with the aim of improving knowledge exchange between families and professionals.

At last year's Conference, Dr Waite and her colleagues from Aston and Birmingham Universities asked people with Bardet-Biedl syndrome (BBS) and their families about their research priorities in the areas of emotion, behaviour and cognition. Dr Waite returned to Conference this year to provide feedback on the research priorities that were described, and introduce a research project developed to examine personal characteristics, mental health and well-being in people with BBS. Dr Waite's presentation can be downloaded in full from the BBS UK YouTube channel.

Dr Waite began by explaining that there are often very characteristic needs within genetic syndromes and she is interested in whether there might be a specific profile of needs in people with BBS which is different to the profile of needs for people with other rare genetic syndromes.

Dr Waite and her team began with a literature review to see what knowledge and understanding was already published; the review reported a preference for routines, repetitive or obsessional behaviours, social and communication difficulties, mood and anxiety disorders and emotional regulation difficulties.

The team attended the BBS UK Conference in 2017 to talk to those living with the syndrome to find out what their research priorities were and whether there were specific things that patients wanted them to focus on. People with BBS and their families reported a consistent pattern of issues including anxiety and low mood, emotional regulation, struggling with change, being inflexible, as well as sleep issues, poor quality of life and difficulties making friends.

The team has started to think about some of the biological and cognitive mechanisms that might underpin some of the characteristics of individuals with BBS; with a particular focus on mechanisms that might help to understand low mood, anxiety and emotional regulation difficulties. Dr Waite explained that it's certainly not the case that everybody with BBS develops these difficulties, some people are doing really well, but some people are not doing quite so well and they want to try to understand that.

Dr Waite has pulled together a group of very experienced clinicians and researchers and has successfully applied for funding from The Bailey Thomas Foundation, to fund a researcher for two years. They will be focusing purely on BBS, thinking about well-being, emotion, cognition and behaviour.

Dr Waite explained that the research will involve in-depth interviews around emotion, depression and anxiety. For younger children, they will use play based assessments to look at social interaction skills and development, and the implications of sight loss. They will also ask specific questions around anxiety.

At BBS UK, we look forward to supporting Dr Waite and her team with their research over the coming year.

Visit to BBS Research Lab in Germany

Laura Dowswell & Emma Oates

Dr Helen May-Simera is a research scientist and started working on Bardet-Biedl syndrome (BBS) in 2003, as part of Professor Beales' team in London. Helen is now based in Germany and has her own laboratory and research team, trying to understand what causes BBS on a molecular level and how these changes in the cells can cause the different symptoms that people with the syndrome experience on a day to day basis. Helen explained that it is basic research that then funnels into other research. In 2017, Laura and Nick Dowswell and Emma and Steve Oates visited Helen's lab and were treated to a tour of the facilities. They were also given hands on experience of extracting DNA. Following, is an edited extract of Laura and Emma's account of the visit, the full presentation can be found on the BBS UK YouTube Channel.

"When we arrived, to say we had a warm welcome is a complete understatement. We had a chat, did some introductions and got to know each other. The team were very keen to hear our family stories which we shared; it was quite emotional. The team talked about what they were doing and their areas of interest. It was quite overwhelming for us. What came across most to us, apart from the huge amount of knowledge and sheer intelligence, was the passion that all these young guys have for researching the causes of the vision loss in BBS.

One lady explained that she was looking at the Retinal Pigment Epithelium (RPE) which is effectively the support for the retina and is a newer area of research. We were given a tour of the labs, one of them was a sterile environment so we had to wear white coats.

Firstly we looked at RPE; Helen showed us the retinal cells under the microscope. We could all see the difference in the cells where they clump together, which was very interesting. We were shown into the darkroom where we looked at slides that showed the cilia and cells in the retina. We were quite overloaded and quite a bit overwhelmed, it was amazing.

We spent some time with Viola, Helen's lab manager, who talked about and showed us lots of the research that is going on and again, lots of questions were fired at her. We left, feeling really excited about the huge amount of research that is going on but very humbled at how hard this whole team works in the background.

The following day, we were invited back to the lab and this is where it got a bit more hands-on. We were able to conduct an experiment to extract DNA from fruit, which is the same process used to take DNA from blood. We were able to use everyday things to extract the DNA, washing up liquid, salt and rubbing alcohol. We mashed the fruits up, mixed it with the liquids and filtered it through tea and coffee filters into test tubes. Then, after a few minutes, the mashed-up liquid settled to the bottom and the DNA rose to the top and we were able to lift it out of the test tube.

It was very simple and very, very interesting to do and again, we had so many questions for Helen who was very patient in explaining everything. At the end of the day, our heads were full of information

and buzzing with all the new information that we'd learned. We are incredibly grateful to Helen and the team for accommodating us; they were so warm and patient with us, it was an experience that we'll never forget. "

Helen replied that it was so rewarding for the team to get to know the families and to meet the people who are actually affected by the syndrome and to show why the team is doing what they do and the reasons behind their work.

My Daughter, My story: A Personal Perspective
Nick Dowswell

'My name is Nick, I have a daughter with Bardet-Biedl syndrome (BBS). 'So tell us something we don't know', I hear you say. Every year, I've been to the conference, I've heard personal perspectives from mums and people with BBS but not from a dad. Here I am.

Maria was born in 1993 at St. Helier Hospital, Sutton. Like all new fathers, it was the most amazing thing, becoming a dad. This little bundle of flesh and bone, just amazing to witness a new life entering the world. As I stood there holding my daughter for the first time, I remember thinking, 'Wow, this girl has so many adventures to have and opportunities to grab, nothing would stand in her way'. It's true to say at this point that Maria has always been daddy's little girl, even now at the age of 24.

In the early years, Maria seemed like any other little girl, if a little clumsy at times. She had to wear glasses to correct a squint and she had a couple of operations to try and correct it with limited success. However, always in the back of my mind was, 'something isn't right' and after an incident at home involving a tray of tea, we began the journey to where we are today. At first we found ourselves at Moorfields Hospital, being told she might have a brain tumour.

When we did get the clinical diagnosis of BBS, I didn't seem that surprised as I had always thought something was wrong and wrongly assumed something could be done about it. After all, dads fix things don't they? Unfortunately for me, the reality soon kicked in, and the difficulty of what I was facing began to sink in. I would always try to do the right thing by my family, but this was something I had no control of and didn't understand.

In truth, and much to my shame, I have to say I didn't take it in, as I was in my world of disbelief. This can't be, not my daughter. Over the next months and years, I felt distant from my wife and daughter, as I tried to find reasons to blame, instead of supporting those I should have been. The strain in our marriage was sometimes unbearable for me.

I felt a huge amount of guilt about my daughter because through no fault of her own, she'd been touched by the BBS gene curse. To me it was a sense of failure. I, we, had let her down somehow. Instead of a bright future, what future if any, was she to have; so much for the strong dad. Her teenage years were a bit of a blur as I wallowed emotionally in self-pity, but at the age of 16, a decision was made to send her to WESC in Exeter, which is a school for visually impaired students.

While there, she studied Equine Horse Care at a partner college. She'd always loved horses and indeed rode at stables when she was younger. I have to admit, seeing her on a large horse used to scare me, but she seemed to take it all in her stride. Little did I know that the decision to send her to Exeter was once again, to change things forever. Her time at WESC, although sometimes difficult, was to change my thinking, that just maybe, there could be a future for her and that she could achieve things I thought were not possible.

One of my proudest moments was seeing her graduate in her Equine and Horse Care course alongside all the other able bodied students. This for me was something that for many years, I thought would never happen. Her confidence grew and her ability to look after herself, and do things herself, increased. Although it wasn't all plain sailing. Maria to everyone else might seem lovely and calm, but, like a sleeping volcano, she could sometimes erupt with destructive emotional consequences.

Maria is now 24 and living in a flat on her own in Exeter, while holding down a part time job. Things I doubted were ever going to be possible. Far from letting BBS stop her, she has made it work for her, almost pushing her to break barriers and misconceptions about her condition. When I think back to the early years, and how having a daughter made me happy and full of hope for the future, to thinking afterwards, after her diagnosis, that there might be little hope and little future, I feel sadness and regret for the way I felt. Immersing myself in feelings I should never have had. You see, the modern man should be strong and supportive of his family, but instead I just fell apart. As I stand here today, I've never been more positive about the future because my daughter constantly surprises me by what she says and what she does. As I've already said, she's faced challenges but has always dealt with them her way, and as a father I couldn't be prouder.

Even though she has this condition, nothing stops her and I shall finish with this thought. At one time I used to think, 'how am I going to turn her life around for the better?' Well, Maria, thank you. Because of your strength and positivity, you have instead turned mine around.

Specialist Employability Scheme

Tim Broome
Student Recruitment Manager, Royal National College for the Blind

Tim Broome is the student recruitment manager for The Royal National College for the Blind, Hereford. Following, is an edited extract of Tim's presentation at the BBS UK Family Conference. The full audio recording can be found on the BBS UK YouTube Channel.

'The Royal National College for the Blind (RNC) is an 'Outstanding' residential college, which has recently been graded as 'high standard' by Ofsted for the residential care element. The college runs academic courses from pre-entry, GCSEs, A-levels, vocational courses, right through to transition to university preparation. Generally, about a third of students move on to university; in 2017, 100% of students on A-level courses gained offers at their first choice university.

For a lot of people, the ‘soft skills‘ learnt at college, the social skills, the friendships, the opportunities that are available, are invaluable. The college is very much a can-do college, we don't really say 'no' to students. We try to risk assess into things, not risk assess out of things.

The encouragement that students gain from the staff, the students, the peer support and the friendships is fantastic. For some, 16 is a very young age to be leaving home, but at college they become mature young adults. Independence is massive whether it be the independent living skills that are learnt in the classrooms or the independent learning skills that they're learning through their courses, or just being an independent young adult. Adults staying at RNC are treated as adults so they do enjoy the social life of Hereford. They don't have curfew times like the youngsters’.

Tim presented photos and case studies of young people who have BBS who are studying at the college, starting with Jody who accessed the Specialist Employability Scheme.

‘Jody came back to college after originally studying massage and complementary therapies to do an NVQ Level 2 in Business Administration. On top of her academic programme, she has had intensive support with work experience to find new opportunities to gain receptionist skills, focusing mostly on telephone skills, booking systems, and customer contact. Jody also looked at employability skills, focusing on updating her CV, and interview skills. Jody has recently left the college to take a 16-hour post as a receptionist at a local gym, fully funded through the Department of Work and Pension’s Specialist Employability Scheme.’ The scheme is a roll-on, roll-off programme and adults can join at any time throughout the year. We have around 15 to 20 new intakes every month, normally for a six month provision, but again it depends on the individual. If we can support adults back into employment quicker, great, that's exactly what we'll do. For others, it might take a little bit longer, again, no problem. It's a one to one tailored program for their needs and is very much employment-focused. If you, or someone you know would like to join the scheme, you need to visit your local job centre; they are the gatekeeper for the program. They will send a referral form to us, and then we will make contact and start the process.

Tim went on to talk about their Younger Learner Programme, for 16-25 year olds.

‘Lydia joined us in September 2017, she is doing our Aspire program, Level 1 Braille, Level 2 Maths, Level 1 English and OCR Performing Arts. On top of that, she has Independent Living Skills Training, Mobility Training, Transitional Support and Career Skills, Lydia says, “I've got lots of friends at RNC, I've been doing lots of activities, it's a lot better here. People are friendly and understand me better. I'm more confident. I'm really enjoying Aspire. I really like the practical work, I've learned how to handle tools and decorate the products. I'm looking forward to doing the performing arts in schools.”’

‘Nathan is doing an NVQ in Fitness Instructing, Level 1 English, Level 2 Maths, NVQ award in Spanish, Level 2 Braille, Independent Living Skills, Mobility Training and Transitional Support, Nathan says “I did not make as much progress as I would have liked during my time in mainstream school but honestly I feel that I've been improving. I'd like to improve my independence by learning how to cook for myself. I want to continue my sports studies as my aim is to qualify as a personal trainer, which will give me a chance of getting into a job.”’

Tim continued, 'RNC are different from mainstream colleges, our class sizes are a lot smaller, we have generally about 1:4 ratio in the majority of classes, sometimes it drops to 1:1 or 1:2, depending on the programme and depending on the courses. We don't have teaching assistants, we don't have learning supports, our students are learning to learn as independent young adults. When they go off to university, when they go into employment, they don't need someone by their side, supporting them or telling them how to do things or making work accessible for them - we teach them how to do it for themselves.

The courses are all built around the individual. When we first meet a student, they'll have a pre-entry assessment and they will tell us about their needs, their wants, their outcomes, what they hope to achieve in the future and from that, we will build a timetable around them. The skills learnt are transferable and based around work experience and work readiness. Work experience is built into timetables; if you do a fitness instructor course, you're actually working in a gym, if you do a business administration course, you're actually working on reception duties, if you do massage and complementary therapies, you're actually in our clinic where members of the public are paying to come in and use that clinic, everything is work-focused and ready for the future.'

For more information go to www.rnc.ac.uk

My Visit to RNC, Hereford

Emmy Anstee

Following, is a report of Emmy Anstee's recent trip to the RNC:

'I went on a trip to Hereford, with the charity, VICTA from the 11th to 14th April and I stayed at the RNC College. At first I was a bit nervous, so were mum and dad, but they came and saw the room in which I was staying and they felt more relaxed. After they left we played games such as 'Two Truths and One Lie' to start to get to know one another.

Our first activity was a Cathedral and Historic Tour of Hereford and I asked one of our group leaders to guide me because we walking past roads and I was somewhere new and didn't know my surroundings. First we went to Hereford Cathedral where they showed us the clay poppies that had been made due to a special event that was going on at the cathedral and they also explained how they were made. Then we went to the Hereford museum which was opposite the street from the cathedral and above the Hereford library. There were many interesting artefacts in the museum such as a roundhead helmet which I tried on.

Then we went back to the college and had dinner and afterwards we still had three quarters of an hour until we went out on a ghost tour of Hereford so I did some chemistry revision in preparation for my GCSEs. On the ghost walk I had to ask the group leader to guide me again because I am night blind, but it was very interesting. We walked around Hereford and were told ghost stories that are related to the town and because I like history I asked lots of questions. When we got back to the college I was so tired that I rang mum and dad and said good night to them and after that I went straight to bed.

On the second day my alarm woke me up at ten to seven and I spoke to mum and dad and then got into my swimming costume and sports clothes because after breakfast our first activity was the spa rooms where there was a hot tub, steam rooms and sauna. I spent most of our hour there in the hot tub. Then we went back to our rooms, got out of our wet swimwear and back into our sports gear because we had a fitness class where we had to alternate between sprinting, sidestepping, jogging and walking. Then we played goal ball and my team won our match and after that I was running around collecting the balls that had rolled off the pitch.

After lunch we had the option of playing either football or goal ball. I decided to play goal ball and had a great time. Afterwards we had an hour to have a shower and get changed and then we went to The Grove to play golf and bowling. On the way there we did karaoke in the mini bus. Golf was really fun, it took me eight goes to finally get the ball in the hole, but by then I had worked out the proper technique. When bowling I was facing the wrong way on one of my goes and my ball went into the gutter of the third alley to my left! I came fifth in our group and fourth overall. Whilst having dinner I got to know some of the other kids better and made good friends with a girl called Abi. Again that night when we got back, I went straight to bed because I was very tired.

On the third day which was the Friday, we went to Oaker Wood Leisure Centre and our first activity was tree top trekking, I was very nervous and I screamed a lot but I still gave it a go. After lunch we did the zip wire but because I generally don't like heights, I didn't in the end. Our final activity was a monkey climbing course which I climbed to the top of the ladder, this was a big achievement for me. That evening after we had got ready we all got together in the common room and started dancing to Cotton Eyed Joe, Gangnam Style and Cha Cha Slide - it was very funny. We went out for dinner and we had a great time. I exchanged numbers with Abi and the next day I went home.

This trip was a great experience. It took me out of my comfort zone, with people I don't know in a surrounding I'm not used to and doing activities I wouldn't normally even attempt and I had a great time. It has helped to boost my confidence and allowed me to be more independent. I met some great people. I hope to have as great a time on the Spanish trip in August which I have applied for because this trip was so good that I immediately wanted to do another one. So I would like to thank VICTA and the RNC for a great trip and an excellent challenge and because of it I had the courage to push myself to do other similar things like going on the Accelerator ride at Drayton Manor during the BBS UK Conference Weekend.'

Shine Like a Diamond: A Personal Perspective

Aneeba Ahmed

I am Aneeba Ahmed, I'm 27 years old and I live in Birmingham. My family knew that I had Bardet-Biedl Syndrome (BBS) from birth as I had extra fingers and toes, I have BBS 10. I do have a mild learning disability and speech impairment. I also have got mild kidney problems. I am visually impaired and have been registered since 2010. I have always worn glasses since I was eight months old; I am photo-sensitive to bright sun light, and I have also got wobbly eyes. I used to have a weight problem but I have controlled this now due to being on a low carbohydrate diet because of my high blood pressure. I have had four operations on my ears, because I have a bilateral hearing impairment. I have also have got spondylosis.

In my spare time I like to sing and listen to music, I love listening to anything of pop, like One Direction or Little Mix. I also like to listen to audio books; I love listening to Jacquelyn Wilson or J.K. Rowling. I also love to go out on my power walks or on my treadmill and like to socialise with family and friends.

I've had the best educational life; I attended Queen Alexander College in 2010 where I studied Preparation for Life, and BTEC Level 1 IT. I used to volunteer for the fundraising team at RNIB, and I'm hoping to go back to RNIB as a volunteer. I like to give something back to BBS UK, I have written a few articles for the newsletters. I have also done three Fun Runs with Sarah Borrows (Clinical Nurse Specialist, Queen Elizabeth Hospital) to raise money for BBS and for RNIB; this year I will be doing it again with Sarah and with Amy Clapp.

I have also attended the BBS UK Adult Social Weekend in November and I have attended the BBS UK Conference in 2014 with my sister - we thought it was amazing; BBS have become like a special family to me.

Yes, there can be days when I go out with someone, and if I see someone who doesn't have a disability, I sometimes think to myself, I wish I was like them, but it doesn't matter because I am grateful for the support I get from my family and friends and especially you guys (BBS members). I didn't just want to come here today to explain about my own experience of living with BBS, I wanted to say something to those who are newly diagnosed with BBS, that you are not alone, because my dad says, 'Special, is not being special, special is getting the support that you need in life.'

Chris Humphreys: A Tribute

Chris Humphreys, as many of you will know, was the heart and soul of BBS UK for many years and was the first point of contact for families looking for support and information; so many of us benefitted from her compassion and experience. As conference organiser, Chris developed the BBS UK Family Conference Weekend into what it is now and even when she was no longer able to attend, she remained supportive behind the scenes, encouraging and supporting the team to ensure our families had the best weekend possible. Sadly, Chris passed away in 2017 and this year was the first conference the team has organised without Chris' encouragement and support. It is a testament to Chris' passion and vision over the years that so many families, new and old, continue to benefit from this very special weekend and we would like to pay tribute to Chris in recognition of her commitment to BBS and all the families she supported through the years.

Chris was married to Phil and together with their family they attended their first pre-diagnosis day conference at Exhall Grange in Coventry around 24 years ago when their son, James was 12 years old. Chris remembered it as a day of initial euphoria at recognising the features of LMBBS and being welcomed with open arms; friendships were forged and suddenly they were no longer alone dealing with the unknown. Chris wanted all newly diagnosed families to experience this and so joined the Committee during the very early stages of the Charity's development. In later years, Phil would become the Chairman of BBS UK. Chris cherished her role and was to become the Charity's National

Co-ordinator, the first point of contact for newly diagnosed families. A member and Charity beneficiary wrote, 'I still treasure the letter you sent me when my son was first diagnosed and I was in a very bad place. Touching things that you have done like that will stay with people forever and please know that you will always have a special, grateful place in my heart.'

Chris retired as a Trustee in 2015, having dedicated 21 years to the Charity. The well wishes and letters received showed the high regard and affection with which Chris was held by those she supported through the years. Those of us who worked with Chris benefitted so much from her unwavering support and encouragement, professionally and personally, and were inspired daily by her selflessness and commitment to BBS UK and the families she supported.

Chris wanted all newly diagnosed families to be able to experience the euphoria of recognition, understanding and acceptance, to forge friendships with those who understand and to feel supported in the BBS community, as she did all those years ago. The team at BBS UK are committed to ensuring this very special weekend remains focused on those aims and objectives. Chris' vision and passion was woven throughout this year's Conference Weekend, benefitting families new and old as it will be for many years to come, which is quite a legacy.

We hope you have enjoyed listening to this conference report, don't forget, contact details can be found at the beginning of the CD.