



BBS UK

Conference Report 2016

Bardet-Biedl Syndrome UK

Registered Charity No. 1027384 and SCO41839



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This Conference Report contains edited excerpts of the talks and presentations from BBS UK Conference 2016. Many of the presentations are available to listen to in full on the BBS UK YouTube channel. Go to www.youtube.com and enter 'BBS UK' in the channel search box.



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Foreword

As usual, conference time came around ever faster in the Burge family calendar. This year was my first as chairman and I was very proud to be working with the passionate and dedicated team involved in orchestrating this year's conference; with Julie and Tonia at the helm, and with the support of the Trustees, the weekend is always a success and I am very grateful for all of their hard work during the year and in the run up to this important and valued event.

The Hilton staff make every effort to ensure that our guests feel welcome and many of them have gotten to know us and our members over the years. We work closely with them throughout the year to try and ensure that our conference runs smoothly and our guests are well looked after. Some of you will have been aware that there were a few issues this year, especially with the booking arrangements, which we will do our best to ensure does not happen again. The Hilton have offered a voucher towards next year's event for those affected and we are working closely to ensure next year's booking process is trouble free.

A special thank you must go to the fantastic speakers who gave up their time to talk to us about BBS research and developments, coping strategies and personal experience. This year's conference saw a wide range of delegates covering a varied range of topics, including access technology, puberty, sexual health and emotional/relationship issues, chronic pain and fatigue and of course the BBS UK Family Conference wouldn't be the same without an update on research from Professor Phil Beales.

Another special aspect of the weekend is the fact that we have a dietitian, BBS clinical nurse specialists, a benefits advisor, members of the BBS UK Family Support Team and other BBS clinicians available throughout the weekend for informal chat, information and support. We really are very grateful for their time and continued commitment to our charity.

Our most grateful thanks must also go to our fantastic childcare team and sighted guides, who enable us to offer trips to Drayton Manor and Bowling. Special thanks go to Sandra, who retired this year after running the crèche for 19 years, you are all stars!



On a personal note, as this was my first conference as Chairman, I would like to pay tribute to Phil Humphreys, and I look forward to following on from his hard work over the past years as Chairman. The current BBS UK Board of Trustees is a dedicated team of parents and adults affected by BBS and I know that we are all committed to providing a high standard of conferences for the foreseeable future. We will once again spend time looking through the evaluation forms to ensure the event remains focused on our members wishes, because it is your weekend and it is only the success it is because of you. As written in a previous conference report, an aspect of the weekend that proves invaluable is the informal peer mentoring and family support that is provided amongst our members. The BBS membership has become an amazingly supportive community; if a VI member seems lost, someone quickly steps in to help out. If someone is upset, a friendly arm is soon around them and if someone is sitting alone, it won't be for long. So thank you to all of you, you are very special people.

I look forward to welcoming you all to our next family conference in 2017.

Steve Burge
Chairman

Update on Research and Study of BBS

Professor Philip Beales

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) 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 and was made President of the Charity in 2005. In 2010, he established, with the help of the Charity, National Multi-disciplinary Clinics, with a comprehensive genetic testing platform for all persons with BBS in England and Scotland. Following, is an edited account of his presentation at the 2016 BBS UK Family Conference:



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Professor Beales began with an update on the new database system which is slowly being introduced at BBS clinics. The database will be a clinical support tool and will store all patient, appointment and consultation information and will allow clinicians easy access to relevant and appropriate information as needed. The system is accessed during clinics via a tablet and is currently being rolled out in the London service; it will eventually be used across all four centres. Patients will eventually be able to access their information, which will prove invaluable if they need care when away from home. Professor Beales moved on to discuss the genetics of BBS and the developing research and potential treatments.

“We found the first affected gene in the year 2000 and now, in 2016, mutations in 19 BBS genes have been identified. BBS1 is the most commonly affected gene in the UK and North America, closely followed by BBS10, BBS12 and then BBS2. Individuals have to have two mutations in any one of those genes to have BBS. Through these discoveries, we have been able to develop genetic testing, the first genetic testing of its kind, certainly in the NHS and I think probably the first in the world. This has been enabled by Dr Beth Hoskins

at the Institute of Child Health (ICH), Great Ormond Street, who has been instrumental in delivering these particular tests.

Of 376 patients tested, 82% have the mutation confirmed in one of the 12 genes that we test for; 164 relatives have now had a carrier test. We have also been able to offer 18 prenatal tests to determine whether the child is going to be affected or not.

By working together we have been able to raise awareness of the condition and reducing the age of diagnosis has been a really successful outcome of the BBS Service. The average age at diagnosis used to be around 14 years. It came down a few years ago to about 9 years and through testing and through the service and patient engagement, the age at diagnosis is getting lower and lower.

We are now starting to look for medicines that might alleviate some of the problems experienced by those living with Bardet-Biedl Syndrome, using, for want of a better word, gene medicines and gene therapy.

Dr Victor Hernandez-Hernandez (ICH), has

been leading the charge on developing gene therapy using BBS1, with some pretty astounding early results. The process involves taking a human BBS gene and putting it into a mouse, injecting it into the back of the eye.

The back of the eye contains these very important cells called photoreceptors, rods and cones, which receive light and turn it into an electrical signal which goes to the optic nerve and into the back of the brain, to the occipital lobe. In BBS we clearly have a problem with the rods and the cones where they're not working properly. They start off working very nicely at birth and then they deteriorate for whatever reason. What has been shown in other conditions is that it is possible to replace some of the genes and restore or retain a certain amount of function in those photoreceptor cells, which is what Victor has been trying to do.

The process involves injecting one eye of the mouse whilst leaving the other eye untreated, so you have an internal control for that particular mouse. We then monitor the eyes over a six month period using an ERG. Victor has been able to show that the eye that has been treated with the gene therapy will retain its function compared to the untreated eye which continues to degenerate. We have completed a first round using 13 mice over a

six month period.

The second round is in its early stages, just beyond the one month period and is looking even better. There looks to be a very significant amount of rescue or improvement in the treated eye compared to the untreated eye. We've got a lot of work to do yet, so we're not making any promises because there's a lot of regulatory framework to get through and as usual, it's about funding, we've got to get some funding to be able to go to human trials.

The next piece of research is based on the Nobel Prize that was presented in 2012 to Sir John Gurdon and Dr Shinya Yamanaka. Sir John Gurdon first described stem cells back in the 1960s, however Dr Yamanaka created a real revolution in the last few years, when he determined that you can take any cell in your body and turn it back into a stem cell by adding a chemical mix.

Stem cells are cells that have gone right back to the status of a very early embryo. They are 'pluripotent' and can become anything, nerve cells, liver cells, heart cells and so on. So what is the value of this? Well, there are many things that we can't understand or can't test in the lab, because we can't get hold of retinal tissue for example, or bits of brain or nerves. This process allows us to 'grow our own'. We have started to use this system in our lab and PhD student, Rosie Davies, has been working and reformulating the various 'recipes' that are out there to take skin cells and turn them into photoreceptor cells and so on.

56 BBS patients gave a small skin biopsy, which we then took to the Sanger Institute in Cambridge. They turned them into stem cells and we then turned those



stem cells into photoreceptors or retinas. By doing this, we can better understand what's happening in the eye and are able to add medications in order to see whether we can slow down the degenerative process.

We are also interested in the cells that are floating around in urine, because they've come from the kidneys. We take a fresh sample and have to process this within four hours in the lab. We then grow the cells in the medium in the incubator, at a nice warm 37 degrees, for four weeks. Those cells proliferate and cover the bottom of the dish, we can then use them in a couple of ways. We can compare the cells with those of someone who doesn't have BBS, which might give us an idea as to what's going on as far as the kidney disease is concerned.

We can also use them to create a 3D culture, by moulding the cells into hollow spheres or spheroids with cilia on the inside. All the spheroids developed so far from BBS patients have been abnormal, being much larger in size.

This is the first time we've been able to see what is going on, without having to stick needles into the kidneys for biopsies and this now gives us the means to test a number of drugs that seem to prevent kidney cysts from forming. In the next few weeks we're going to add those drugs to these particular cells to see whether we can prevent them from becoming large and swollen. So hopefully next year we'll be able to give you the results of that.

Lastly I will talk about Dr Sonia Christou-Savina's work. Sonia has been studying BBS mice and she's made a pretty astounding discovery in the brain of the BBS mice. The number of neurone branches and synapses (connections) of the brain cells are much fewer in the BBS mice and Sonia has been able to show that a very simple intervention significantly improves these, and that's exercise. When we put a wheel into the cage to increase the exercise levels of the mice, we found that after just a few weeks when we had a look at their brains again, we saw

a complete recovery; the number of neurone branches and synapses had increased significantly.

Through the summer of 2016, we will be conducting a pilot study involving a handful of children to look at exercise and its effect on the brain. We will recruit about 20 children and will first use an MRI scanner to look at the brain and in particular the hippocampus. They will then be put on a six month exercise programme, which we've worked out with a physiotherapist, and will be done at local gyms. At the end of the six months, we will do a second scan to see whether there has been any improvement. This has been done before but not in BBS. A study from the University of Illinois showed activity in the brain is significantly increased in the learning parts of the brain after a 20 minute walk, compared to just sitting around."

Professor Beales took questions from the audience:

Delegate: "The software mentioned earlier on, would it be possible to link up with GPs' IT systems in the future?"

Professor Beales: "I think it's unlikely they would. Where I see interaction with the GPs happening better is in the next version of the database. What we're doing is developing a system where you can look at your own information, so if you happen to be on holiday and you have an accident and you have to go to the hospital, you can pull up your own information with your protected password. You can access results, hopefully in the future see some of the scans, your latest blood tests and those kind of things. And of course you can do the same if you go and see your GP."

Delegate: "That student who has grown the eye, could she grow a complete eye, so take the old eye out, and put the new eye in, connect it etc?"

Professor Beales: "Not in my lifetime, however one day it probably will happen, but there are just too many interdependencies."



Annual General Meeting

The Hilton Hotel, Northampton
23rd April 2016

Apologies

Trustee apologies were received from Dianne Hand, Graham Longly and Rob Hymers.

Minutes of 2015 AGM

The minutes of the last AGM of Bardet-Biedl Syndrome UK having previously been circulated were confirmed to be a true record of proceedings.

Election of Honorary Officers

Of the current Officers, Steve Burge, Chairman; Richard Zimble, Vice-Chairman; Laura Dowswell, Treasurer; Emma Oates, Fundraising Co-ordinator were all eligible and agreed to stand for re-election. Abbie Geeson, having covered the role of Secretary for the past year, was eligible for election to this position. No other nominations had been received for these positions and the Honorary officers were all duly elected unopposed.

Election of Committee

Of the current trustees, Allan Clark, Stefan Crocker, Dianne Hand, Rob Hymers and Graham Longly, all had a further year to run in their present roles. Allan Clark opted to retire this year and was thanked for his valuable contribution. The committee therefore had two vacancies and one nomination in hand; Nominations were received for Margarita Sweeney-Baird for this post who was duly elected unopposed.

Chairman's Report

The Chairman's report was read out on behalf of the Chairman, Steve Burge, by Tonia Hymers.

There have been many changes to the board over the past 12 months starting at the AGM last year where we paid tribute to the long and legendary careers of Phil and Chris Humphries who have given so much of their time and worked tirelessly for the charity. Special thanks



go to Chris for the support she gave to so many families and for the many fantastic conferences she organised over the years, she is very much missed. Phil and Chris are a hard act to follow, however the new BBS UK board have stepped up to the mark and I am very proud to be Chairman of such a dedicated team of people; I look forward to working with you all to develop a strong charity that will ensure that we continue to develop well into the future.

Kevin Sales has retired as a trustee but continues to work as the Finance Officer for BBS UK and BBS UK Clinics Ltd and is working closely with the new Treasurer, Laura Dowswell. Julie Sales and Tonia Hymers also retired as trustees, but will continue to deliver the four amazing BBS clinics that have been running since 2010. Our thanks go to the hard work of Kevin, Julie, Tonia and the rest of the dedicated team ensuring the successful delivery of the clinics; Kevin, Julie and Tonia are still valued members of the BBS UK management team and continue to support the new board of trustees and have played a central role in helping all of the trustees shape the future of the charity.

BBS UK Clinics are, as always, growing from strength to strength. Additional support continues to be provided by Angela Scudder, the Family Support Worker who was employed

after a successful grant was awarded from the charity Jeans for Genes, enabling Angela to develop working with families and ensuring that they get the on-going support that they need to deal with the challenges of having children affected by BBS. I am pleased to welcome a new member of staff to the team, Kate Zaczek, who is the new Adult Support Worker. Kate joined the team in January 2016 and she will be working with all of the staff to ensure that the adults are gaining access to the support that they need outside of the clinics.

Over the past 12 months the whole team have been busy working on several projects to ensure that the charity continues to move forward and support our members, giving them information using all possible means of getting the message out there, so the awareness of BBS grows. At the AGM in 2015 it was agreed by our members to change the name of the charity from LMBBS to BBS UK. This decision was taken due to the fact that in the medical profession BBS is more commonly used to describe the condition and as a board of trustees we felt that it would be prudent to make this name change to bring the name in line with its medical terminology.

At the same time, we voted as members to change the logo of the charity and this was changed to the new BBS UK logo which will be displayed on all publications and literature.

The trustees have been working hard in collaboration with Robert and Nicola Hueting to redesign and build a new website that will be up-to-date and will be a main resource and an exciting place to visit to learn all about what is going on within BBS UK. I would like to personally thank both Rob and Nicola for their outstanding work on the website and also for all of the technical support that Rob has given to myself and the rest of the trustees ensuring that we are able to go forward into the virtual world of the future. The trustees now have new email addresses and these will be posted on the new website once they are active.

I am pleased to announce that after a lot of hard work from Tonia Hymers, we now have a brilliant new medical booklet and Tonia has spent some considerable time ensuring that all of the content is current and endorsed by all of our medical practitioners.

Graham Longly along with Stefan Crocker,

Richard Zimble and myself have set up an adult focus group and this was a result of the two successful adult receptions which have been held for the past two years at our annual conference on the Friday evening, which have been an opportunity for adults with BBS to meet and relax and talk freely in a comfortable and friendly environment. The group has met twice now utilising Skype so that we can meet online and we are working closely with the rest of the trustees to ensure that the adults have a voice for the future. If you would like to join the focus group then please contact either Graham or myself and we will be only too happy to invite you to a meeting in the future.

I want to finish by thanking all of our valued members, friends of BBS UK and fundraisers for all of the hard work that they put in over the year to ensure that BBS UK has the finances, resources and manpower to continue to provide the standard of service that as trustees we pride ourselves on providing year after year. However, it is important to realise in these times of austerity that we need to remain vigilant in our efforts to continue to raise funds to ensure that the charity is financially viable for the long-term future.

The charity would not be able to operate without the generous hard work by all of our fundraisers who, as always, work tirelessly coming up with new ideas to continue to raise desperately needed funds for the charity's continued survival and the board of trustees would like to thank every one of you.

Treasurer's Report

The Treasurer's report was read out on behalf of the Treasurer Laura Dowswell by Abbie Geeson.

For the financial year, 1st January 2015 to 31st December 2015, the charity received an income of £59,796 which is an increase of around £19,477 compared to 2014. The charity's expenses for the same period have increased to £61,311 an increase of £22,891 compared with 2014.

However, during 2015 the charity received a grant from Jeans for Genes of £11,740 for a specific project. If it was not for this grant and other restricted income, the charity's income from donations, fundraising etc. would have been £42,047. Similarly, of the expenses totalling £61,311, £18,544 of this was directly

related to the Jeans for Genes project and other related project costs. Once this is excluded the actual expenditure was £42,767. The funds at year end 31st December 2015 are £37,995.

As you will note the charity has made a loss for four of the last five years and this is unsustainable for the charity's long term survival.

| Year | Income | Expenditure | Surplus/Loss |
|------|---------|-------------|--------------|
| 2015 | £59,796 | £61,311 | -£1,515 |
| 2014 | £40,319 | £38,420 | £1,899 |
| 2013 | £41,741 | £42,824 | -£1,083 |
| 2012 | £48,801 | £57,005 | -£8,204 |
| 2011 | £58,364 | £39,785 | -£18,579 |
| 2010 | £54,223 | £39,785 | £15,223 |

It is important to note that during 2015 the charity received a one-off donation as well as income from a couple of one-off large fundraising events which generated income of just over £11,000. If it had not been for this, the closing position at year end would have been £48,796 with a loss of £12,515. This is unsustainable long term.

If there is not a large influx of funds/grants, it's unlikely we will be able to hold a 2017 weekend conference.

Much work has been carried out by the committee - closely monitoring the finances and putting a new financial policy in place as well as making some difficult financial decisions including reducing expenditure on this year's conference and cost cutting in general. To secure the future of the charity financially, it is imperative that we develop some reserves and we can only do this with the ongoing committed support of our members.

In financially challenging times, every charity relies on its regular donors and regular income to enable its core work to continue. We do have a 'Friends of BBS UK' scheme, with many of our members making regular donations or holding fundraising events. Around £4,700 has been paid into the charity account during the year via standing order by our 'Friends scheme'. Ideally this amount needs to increase and we ask that as many of you as possible sign up to support the charity in this way.

An example of how even a modest amount of just £10 per month per person can really make a difference: If 10 people gave £10 a month the charity would receive £1,200 over the year,

and if 20 people gave £20 a month the charity would receive £4,800 which would really make a difference and help fund future conferences, weekend activities to name just a few and to help support our families.

The committee would like to offer huge thanks and extend our appreciation for those who take time and effort to fundraise for the charity.

Appointment of Auditor

The Trustees proposed that the charity continue to appoint Michael Bannister, of Fryza Bannister Financials Ltd, for the coming financial year and this was duly agreed.

Any Other Business

At last year's AGM, members voted to change the charity's name and following this vote the trustees have proceeded in taking this forward. To complete this name change in full, the Charities Commission required the charity to propose and vote to change its name from Laurence-Moon-Bardet-Biedl Society to Bardet-Biedl Syndrome UK. Voting was by a show of hands and the motion was passed unanimously.

The charity is currently an Unincorporated Association. The trustees voted at its February 2016 meeting for the structure of BBS UK to change and therefore proposed at the AGM that the charity changes its legal structure from a Charitable Unincorporated Association to a Charitable Incorporated Organisation (CIO). Voting was by a show of hands and the motion was passed unanimously.

The trustees also proposed that the constitution be adapted to a CIO model constitution to reflect this new legal change and required members to vote as per clause 13 in the constitution for these alterations to be made. Voting was by a show of hands and the motion was passed unanimously.

In the absence of any further business the meeting was closed.

The full BBS UK Trustee report for 2015 including accounts is available to download from www.bbsuk.org.uk or from the Charity Commission website.

BBS UK Welcomes New Trustees

On behalf of everyone at BBS UK, we warmly welcome Abbie Geeson and Margarita Sweeney-Baird on to the Board of Trustees, they introduce themselves below:

Abbie Geeson

I am mum to Jessica who is 16 years old. She was diagnosed with BBS7 four years ago and ever since then everything has made more sense to us. This is our third year at conference and each time I learn something new which only supports us in our BBS Journey. We have been welcomed into the BBS family and have truly embraced it.



Two years ago I was diagnosed with multiple sclerosis and I did and still am evaluating my path. I have realised that I have a lot more to give in many different ways and I would like to support BBS UK in any way I can for as long as I can, as it is such a vital support network to myself and Jessica and to us all.

I have worked in the charity and voluntary sector for ten years and have had various roles in management, finance, fundraising, project coordination, administration and have previously been a secretary for a management committee. I recently went back to college and have an AAT level 4 qualification in Accountancy and for the last two years I have worked as a Freelance Bookkeeper.

I have gained a wide range of experience and knowledge from the roles I have had and from the different types of charities I have worked for which include the arts, environment, community and children services. I have very

much valued and learnt from the diversity of these different types of organisations over the years.

I was asked last year to be an observer on the Board of Trustees and in June 2015 volunteered to become acting secretary to BBS UK. I have valued learning more about the charity and I am looking forward to the opportunity of working alongside a great team of trustees and employees for an invaluable charity. I feel I understand the difficulties charities can face and the vulnerability of this sector and think I can help support the charity in many ways to aid in its sustainability, growth and development now and in the future.

Margarita Sweeney-Baird

I have three children, Christina who is 23, Kenny who is 16 and Juliana who has just turned 13; Juliana has been diagnosed with BBS 1. As a mum I always knew that the behaviour of



a child who would go rigid with anger when denied food was not normal. She was born at a normal weight and was breast fed and yet was obese by 6 weeks old. She was also very unstable as a young child in every way. 18 hospital admissions between the age of 1 and 30 months old because she would desaturate overnight caused huge problems. She would not regulate temperature and was emotionally very labile.

So started the roller coaster of the patient odyssey of trying to obtain a diagnosis for Juliana. Now that we are firmly established by the genetic tests as being part of the BBS community you now have a captive supporter. Quite simply no matter how busy I am, I will do as much as I can to help BBS UK and the community. I truly understand how difficult life is for the BBS patient and their families and

joining the BBS community I finally feel that we belong somewhere.

My skills are legal and sporting. I was Scottish Ladies Figure Skating champion and a top Law Student at Glasgow University. I was awarded a John F. Kennedy Scholarship to study at Harvard where I obtained my Masters in Law. The Kennedy Scholarship introduced me to the world of disability sport. The dearly beloved late Senator Ted Kennedy himself told me about Special Olympics at a Kennedy Reception. Special Olympics was well established in Great Britain but there was no figure skating program and ice skating was not in the Paralympics. So I set to work. With the confidence of a Kennedy scholar I thought - if Eunice Shriver Kennedy could do it - so could I.

Inclusive Skating is the result, a charity that I set up to run Inclusive Skating events for skaters with any form of disability. Inclusive

Skating has created events for all disabilities on a fair and cost effective basis. The rules and procedures for the skaters are designed and adjusted to allow skaters with all forms of disability to compete. Skaters with cystic fibrosis, missing limbs, heart conditions, Downs Syndrome, Bardet-Biedl Syndrome and Goldenhar Syndrome (to name just a few) all compete together at the same event. We have successfully held our 5th Anniversary Event at Glasgow 2016.

Juliana is the new British Inclusive Skating Novice Ladies Champion and the first British Blind Sport B5 Novice Ladies Champion; I am very proud of Juliana. BBS patients need sporting opportunities. So do children and adults with any form of disability. I hope that my skills at building new events and opportunities for skaters and athletes with disability can also be used to help the BBS charity.

Cilia and the Eye

Dr Helen May-Simera

Dr Helen May-Simera was born in the UK in 1981 and studied biochemistry at the University of Bath. After completing her Master's degree in 2003 she was a doctoral student at University College London, where she obtained her doctorate in 2008. She 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 she was awarded the prestigious Sofia Kovalevskaja award, allowing her to continue her research at the University of Mainz, Germany as a young group leader. Having missed last year's conference, Helen said she was excited to be back and to talk about the work she and her team are focusing on in her lab in Mainz. Following, is an extract from Dr May-Simera's presentation at the BBS UK Family Conference.



For the few people in the audience that are new to this field, I will begin with a basic introduction to cilia. All animals and plants are made out of cells and there are many different types of cells, for example, skin cells, bone cells and hair cells and they all come in different sizes and shapes. Some of these cells have tiny little hairs sticking out from their surface, called 'cilia'. There are two different

types of cilia, 'motile cilia' and 'primary cilia'. Motile cilia all work together, like a sweeping brush to move dirt or fluid along whereas primary cilia function in a similar way to a radio antenna. For many years, people didn't think the primary cilia were important and it was through Professor Beales' work back in the late 90s that they found out that this signalling antenna, the primary cilium, is really very important as it helps cells communicate

with each other by sending and receiving signals.

The eye has many different types of cells that have cilia or are comprised of cilia. The retina is found at the back of the eye and is very sensitive to light; it's very thin and almost transparent. When you look at the retina itself and break that down, you again have different layers and some of these layers have a very special cell called the photoreceptor cell. What is so special about these photoreceptor cells is that the whole outer section of these cells are comprised of a primary cilium. There are two types of photoreceptor cells, those that look like rods and those that look like cones. Both of these have a modified primary cilia. If these cilia aren't working optimally, then the cell starts to break down and the retina gets steadily thinner, which is when you start to have problems with your eyesight.

BBS genes tell the cells how to make proteins, the building blocks needed to make and build the cilium. If there is a mutation in the proteins, then the building blocks will not come together properly and the cilium can't function optimally; it's still there, but unable to do its job. So that is generally what we've always been interested in and what we've been looking at.

Most people in the scientific community are working on these photoreceptor cells and focusing on the rods and the cones. However, there are lots of other cell types in the eye and one very important cell type is the RPE, which is a cell layer behind the retina, behind the photoreceptor cells. The RPE cells are the caretakers of the photoreceptor cells, their job is to generate, maintain, and build new photoreceptor cells.

No one is focusing on the fact that these RPE cells also have cilia and that these cells are important for nurturing and looking after the photoreceptor cells, which is where I come in. I believe we also need to look at these other cells because they're not looking very happy, we need to make them happy. If those cells are happy, they can then support the sensitive photoreceptors. This is our aim, trying to improve and help the growth of the RPE cells which will then support the photoreceptor cells.

I've been in Germany for about a year now and last summer I was invited to go to the German Patient Society which meets every two years, with only about 20 families. They were keen to learn about the BBS specialist clinics in the UK, because they really want to implement them as well, however not having the NHS means that it is so much more difficult to coordinate. I realised just how fortunate we are to have the NHS which has allowed the team to develop this service. Finally, I would like to thank the BBS Community, because it is you who give us the motivation, drive and the reason to get out of bed in the morning to do what we do.



Christian Bolton-Edenborough: A Personal Perspective

I was diagnosed with BBS1 in October 2014. I am a Masters Law student at Bangor University and I live in Hertfordshire. I go to the Guy's BBS clinic every year. I know this might sound strange but I was quite thankful of being diagnosed with BBS because it explained a lot of things that happened in my childhood. I'd always struggled to see long distance. I'd been given various glasses which didn't help and I could not understand why. I went to university, not knowing why I couldn't see in the dark. I've got a big passion for car racing, so I always wanted to drive but my local optician thought I might have a problem and sent me to Moorfields where I went through lots of tests and one of the consultants thought I might have BBS so sent me to see Professor Beales.

My eyesight is still quite good so I can do a lot of things in daylight, I am only blind in the dark. I am appreciative of the sight I've got left and I want to try and make a difference by helping people with BBS who maybe can't exercise as well or be as mobile. When I went to my first clinic I was told I was obese and needed to lose weight. A real motivation for me was to try and raise funds for BBS UK. Nigel Hills was starting up a Cycle Across Holland event so I thought that sounded good motivation. I come from a family of cyclists so it is something I can do to help raise funds. Through that I managed to hit my target of losing the weight so there was a dual benefit to it. Stefan Crocker and myself are doing a 10 mile run on October 23rd. I'm his guide runner, so that's going to be interesting. You've got me with bad peripheral vision and spatial awareness and you've got Stefan with limited central vision... I like to think of us as the Chuckle Brothers of BBS so we'll see how that goes!

I hope I can make a small difference with Stefan. We're trying to raise £2,000 for the charity. I'm hoping to give an interview on Radio Wales to try and raise awareness because not much is known about BBS in Wales. I want to try and put information out there about the cilia and research that is going on and how we need the funds to make that research happen.



I want to tell you a funny story from before I was diagnosed with BBS. As a student you go out and have a good time and have a drink. Getting slightly tipsy and being blind in the dark is probably not the best recipe for success! I was the social secretary of my university's tennis society and heading home late one night, I suddenly realised it was very dark, all the street lights were off and I was in a bit of a pickle! I got to what I thought was my door, turned the key and thought - hang on a minute, why has the landlord changed the lock? I knocked on the door furiously asking for someone to let me in. I suppose I should have been wise to the fact that three Chinese people were standing at the door with spatulas looking at me very aggressively... and since when have we had a door knocker? And then there's my flatmate standing two doors to the left shouting, "Christian, we're here." Unfortunately, I've not been able to live that down since!

Another story is to do with food and being colour blind, which I am. You'd expect Ketchup sachets to be red, so when I picked up what I thought was a normal Ketchup sachet, it turned out to be vinegar, much to my horror after pouring it all over my food. Due to my pride, I tried not to let any of my friends know because they would have taunted me furiously for this. I ate all my pasta with this soaked vinegar on it and tried to cover up my blushes!

I hope to do more things because I've got a real passion for sport. It's my way of coping with the disability, it makes me feel positive about it. By

making a difference it feels like I'm fighting my own syndrome and over the last year I feel like I am able to conquer it. I will continue like that and hope it doesn't stop me going into a career in law because I aim to become a solicitor.

One frustration has been with work where I have had a few setbacks because of BBS. I've had a few jobs where I turned up for work and although I told them I was disabled with eyesight problems, they said, "Well, you're not registered. You're going to have to work at the same pace as every fully sighted member in this factory." Even though I can see in light, I was having to peer at things and unfortunately my supervisor didn't like that and said I was working too slowly. I was threatened with the sack because I couldn't see properly. Things like that have been challenges but I'm very fortunate now because I've managed to get a pub job with a lot of light above me.

My first ever conference has been lovely and I hope to keep coming. I am always happy to chat to anyone and share any experiences to build up a bank of knowledge so I can then go



and present evidence to raise awareness and apply for funding and things like that. I really want to push knowledge of BBS forward.

Special Educational Needs: Support for Special Educational Needs, and Applying for Education, Health and Care Plans

Eleanor Wright, CEO of SOS!SEN

Eleanor Wright is Chief Executive of SOS!SEN, a charity which helps the families of children with special educational needs and disabilities (SEND), particularly in relation to accessing the correct support for special educational needs (SEN). She is a solicitor specialising in education law, and worked in private practice until February 2015. Her work includes leading workshops on various practical and legal aspects of SEN provision, providing advice at the charity's advice clinics and on a 1:1 basis, helping with casework and drafting letters, representing parents at tribunal appeals, and representing the charity on various committees and in lobbying the Department for Education. Following, is an edited extract from Eleanor's talk at the BBS UK Family Conference, April 2016.



The charity SOS!SEN helps families of children with special needs and disabilities to access the right support. Parents and carers who know their rights and know the system, are so much more empowered to stand up for themselves and not be misled about what their children's rights are.

SOS!SEN operate in various ways. They have a helpline on weekdays and Tuesday evenings in term time which is run by volunteers, including parents who have been through the system and have children with difficulties, and ex-teachers. They also run a number of drop-in centres, mostly in the south but they do have one outpost in Manchester. They also help by

going to meetings, drafting letters and helping prepare for appeals to a tribunal. They do full or half day workshops, usually with up to ten people attending. More information is available from the SOS!SEN website and Facebook page.

New legislation is contained in the Children and Families Act 2014 and various regulations under that Act, however the most useful document from a practical point of view is the Code of Practice (2015 version, mainly chapters 6-9). If you are at the stage of getting an EHC Plan, chapter 9 is particularly helpful. Every local authority (LA) has to have on its website a local offer which details all the provision available to children and young people in their areas with disabilities and SEN, including things like play schemes, schools, social services support, health support and things that can be accessed from outside the area; plus information on how to apply for an Education, Health and Care Plan (EHC Plan).

Legislation requires LAs to have regard to the need to support the child or the young person to facilitate their development and help them achieve the best possible education and other outcomes. A "young person" is defined as someone over the statutory school leaving age (i.e. past the last Friday in June in the academic year that they are 16). Once a child becomes a young person, all the relevant rights pass to them. Provided that they have the necessary capacity to understand what is involved, young people have the right to ask for an EHC Plan or be consulted about its contents, or about what school or college they want to go to, and it is they who have a right of appeal in respect of special educational provision. The expectation is that parents will still be heavily involved and of course if the young person doesn't want to do it or doesn't feel able to do it, they can delegate. If the young person lacks the necessary capacity in legal terms to make decisions, then the normal expectation is that the parent will act as their representative.

In the new SEN system (Special Educational Needs), SEN Support has replaced School Action and School Action Plus, and the Statement of Special Educational Needs has been replaced by an Education, Health and Care Plan. This is meant to be a holistic document recognising that, for children with SEN and disabilities, there are often medical and social difficulties as well

as educational ones and it is artificial and wholly unhelpful to view them separately.

Parents should be looking ahead to every stage in the child's future and think about where they want them to be in the short, medium and long term. The older they get the more everyone concerned should be looking towards adulthood and preparing for independence and self-care and getting into work, or simply leading a fulfilling and happy life. When making decisions, LAs and schools must listen to children, parents, carers and young people and pay attention to their views, wishes and feelings.

With or without an EHC Plan, it is the duty of nurseries, schools and colleges to use their best endeavours to meet the needs of children and young people with special educational needs and disabilities. This also includes adjustments to try to avoid problems happening. The SEND Code of Practice covers children from 0-25 years, and there is a lot of emphasis on identifying problems and supporting children as early as possible. Early Years providers, health visitors and schools need to be alert to identifying problems, meeting them and reviewing progress. They must not discriminate against someone by reason of disability and schools cannot refuse to admit a child by reason of the fact that that child has learning difficulties.

A baseline should be established when a child goes into school or a college so that they can be monitored to make sure that they are making the progress that is required. 'Less than expected progress' is defined in the SEND Code of Practice as progress that is significantly slower than that of the child's peers starting from the same baseline. This means the gap between them and their peers fails to close, or widens. At the very least, progress needs to be consistent. When we talk about progress it's not just academic progress, it includes behaviour, social skills, communication and sensory issues.

If a child isn't making adequate progress, the first step is usually to put them onto SEN support. This should give the child access to high quality teaching which is differentiated for individual pupils. SEN support is meant to be a cycle of assess, plan, review, i.e. you assess what the problem is, plan how to meet those needs, do it and then you review it to

see how it's working, then maybe go back to the beginning of the cycle with appropriate adjustments. The SENCo (Special Educational Needs Co-ordinator) needs to be involved and parents ought to be kept informed.

Parents must be told when a child is put on SEN support. The SEND Code of Practice requires that schools meet with parents three times a year to set targets and review what is going on. Schools have to keep good records of children's progress and need to show if they are putting interventions in place, such as emotional literacy groups, speech and language groups and one-to-one sessions. All schools should have a SENCo, SEN policy and a governor who is responsible for SEN.

If a child does not make adequate progress when on SEN support, consideration should be given to applying for an EHC needs assessment which may lead to an EHC Plan. The application can be made either by a school or the parents (or the young person if over 16). The criterion here is simply whether the child may have SEN and whether they may need help under an EHC Plan. That in turn depends on whether their needs can be met without an EHC Plan, i.e. whether they can be met within the resources normally available in school. Sometimes a child will make good progress because the school is going well beyond the course of duty. This is great, but it is not guaranteed because if their funding is cut, that support could disappear. There may also be a situation where they are making progress because the parents are putting in a lot of support and they may be getting specialist tuition or therapies and so on. These important factors must be taken into account by the LA and must not be used by the LA as an excuse not to do anything because progress is being made.

School funding works on the basis that a notional sum of £4000 a year is available for all children in maintained mainstream schools, and a further £6000 is delegated for those with SEN whether they have a statement or EHC Plan or not. Some LAs automatically refuse assessment on the basis that they allege that the cost of meeting the child's needs won't exceed that figure. That is incorrect. Until the child is properly assessed, they do not know what the costs of meeting needs will be; and there is considerable case law where the tribunal has

found that it is not good enough for an LA to say that support is theoretically available when in practice the child isn't receiving it.

If an assessment is needed, the request can be made either by the school or by parents. Once an assessment is requested, the LA has six weeks to decide whether to do it. If they say 'no', parents can appeal. If they say yes, they have approximately ten weeks to carry out the assessment: it must include obtaining advice from the school, any relevant medical advice, and an educational psychology report. It should also include other appropriate advice, or advice which the parents reasonably request, for example from speech and language therapists or occupational therapists. Having carried out the assessment, they must inform the parents whether they intend to issue an EHC Plan, and if they decide against, the parents again have a right to appeal. If they decide to issue a plan, they should complete the whole process within twenty weeks from the date that the LA received the original request, and parents should make a fuss if they are going beyond those timetables.

The advantage of an EHC Plan is that it means there is a document which, if it complies with the law, summarises the child's difficulties and sets out in detail what support they should receive so that school staff know precisely how they should be helping; that it brings extra funding if necessary; and that support set out in section F is the child's right which can be enforced by law if necessary, with the help of legal aid in the child's name.

For further support and information, go to: www.sossen.org.uk



Management of Pain and Fatigue in Chronic Conditions

Malena Hallahan, Counselling Psychologist

Malena Hallahan is a BPS Chartered and HCPC registered Counselling Psychologist with an expertise in Chronic Pain and Physical Health conditions as well as Addictions. Drawing upon years of experience working in various NHS settings as well as in private practice she delivers evidence based, cutting edge psychological treatment such as Cognitive Behavioural Therapy (CBT) and Acceptance and Commitment Therapy (ACT).



Since qualifying as a Psychologist in 2004, Malena has been working with various mental health conditions and Physical Health Psychology settings within the NHS in both London and Nottinghamshire. She has extensive experience working with those suffering emotionally as a result of chronic or acute illness. Malena has recently joined the BBS Multi-Disciplinary team at Guys Hospital, London. Following, is an edited extract from Malena's presentation at the BBS UK Family Conference:

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"My talk is about the psychology of pain and why psychologists are involved with chronic pain, especially as people generally perceive pain as only having a physical element. I will talk about the issues and difficulties that people experience with pain so that those afflicted can recognise how the issues apply to them and what they may need to attend to.

To begin, take a couple of seconds to think about how you understand or would describe your pain. How would you explain it? It sounds easy but it is quite complex to describe. Pain is an unpleasant sensory experience which in a physical sense, functions to alert the sufferer to actual or potential tissue damage. But as well as being physical, pain has a biopsychosocial impact on the sufferer which is often overlooked. Pain is always subjective. Each person will experience pain in a different way. So it is important to understand each person's experience of their pain.

An explanation of pain is that it is the result of the brain's assessment of a threat. For example, if you put your hand on a hotplate, signals quickly go to your brain. The role of the brain is to assess what is happening

and whether the heat is at a level that will cause danger or damage. If the evidence of danger outweighs the evidence of safety the brain's reaction is to protect the injured area with pain. In this way pain both protects us and prevents further injury. It also promotes healing by drawing our attention to the injury so that we seek help and look after ourselves which enables the injury to heal.

It is important to differentiate between acute pain and chronic pain. Acute pain is a useful warning and tells us something needs attention. It warns us that there is a danger of potential tissue damage or a possible fracture. Unlike chronic pain, acute pain is normally quite responsive to medical treatment and it lasts for a shorter period.

Chronic pain is ongoing pain that persists beyond three months. It is not necessarily a warning of ongoing tissue injury and is unfortunately less responsive to medical treatment. Chronic pain is persistent and has far reaching social, emotional and physical impact on everyday activities and the sufferer's long term way of life.

Persistent pain can impact on us emotionally and lead to a focusing or over focusing on

pain. Imagine having high levels of chronic pain, on a rating of 1-10 maybe at a score of 8 or 9, on a daily basis, even constantly for 24 hours, seven days a week, every day, every hour, every minute, every second and never having a break from it. Such pain can lead to gradual withdrawal from the world.

Whilst difficult thoughts and feelings around persistent pain are understandable such thoughts are unhelpful and increase inactivity. Catastrophizing thoughts are where we overstress what is happening to us and they set up a pattern of thinking and feeling that can lead to intense difficult and overwhelming feelings that all is "doom and gloom" whilst continuously thinking "I cannot cope", "this pain is terrible and will never stop", "my pain will always be there". This unhelpful thinking pattern may lead to the person suffering with chronic pain starting to experience anxiety and/or depression.

This fixation on pain can lead to people experiencing high levels of anxiety, fear, stress, guilt, anger, mood swings, poor concentration and reduced quality of life. Can you imagine having chronic pain and believe you can't cope with it and that you can't trust your own body? You might want to walk a certain amount but fear that you might not be able to get back. This can impact on self-confidence and the sense of who you feel you are. Maybe you've always seen yourself as sporty or active, or very sociable. Over time you may feel that the chronic pain has changed how you see yourself and how you are and behave with other people. You may gradually withdraw from the world and disengage from life, stopping activities and hobbies. The fun goes out of your life and you feel isolated.

If you suffer with chronic pain you might want to consider attending a pain management programme and I will now tell you a little about this approach mainly from a psychological point of view. Pain management programmes aim to moderate the emotional impact of chronic pain. They help individuals understand and change their experience and

their relationship with their pain. If pain is impacting physically, mentally and socially, helping yourself with the emotional aspect can lead to a better sense of coping and improve your quality of life. Awareness of the psychological aspects of chronic pain can reduce the behaviours associated with having persistent pain and reduce physical tension and help you manage fatigue better.

What I'm suggesting is that if pain is subjective and dependent on our thoughts and feelings then that's quite an optimistic thing. Because if we understand what's going on for us with our pain and recognise our thoughts and feelings about it, we can cope better with pain and improve our daily activities.

The biopsychosocial model used incorporates all aspects impacted on by pain. Increasing disability can lead to social, family and financial disadvantages as not coping with the pain leads to having to adjust work tasks, going part-time or giving up work. Psychologically pain affects our mood, beliefs and how we perceive the world and can impact on physical health and lead to disability. Cognitive Behavioural Therapy (CBT) is well recognised to be of benefit with this huge emotional impact and is ideally incorporated in the treatment for self-management of pain.

Other difficult thoughts and feelings are around a person's sadness and loss about their situation. They may be angry and frustrated with their pain which they haven't chosen to have but have to live with. Also the loss of independence and having to rely on others can lead to feelings of hopelessness and helplessness.

The effects of depression, low mood or people losing motivation or interest in things can affect our appetite and sleep patterns which can leave us feeling fatigued, guilty and worthless. We may change what we eat or the way we eat or we may stop eating, or eat too much.

Another common problem with having chronic pain is the effect on sleep pattern such

as sleep deprivation, insomnia, disturbed sleep and oversleeping. People can find it difficult to fall asleep because of their pain, or maybe to stay asleep because of difficult thoughts and feelings, or struggle to get back to sleep because of pain waking them up or keeping them awake. Even if a person does have a full night's sleep they may still not feel refreshed.

With symptoms of anxiety associated with chronic pain, people can feel muscle tension, restless, irritable and that we are not in control. The body reacts to being in constant pain. Muscles tense up and over time feel achy and tired. Then if the body becomes less active, the resting unused muscles become less flexible and less powerful. This can lead to even less engagement in usual activities like walking. So our behavioural responses to pain can reduce the function and quality of life. So we need to attend to how thoughts and feelings impact on the subjective experience of pain.

In our multi-disciplinary, self-management of pain approach we work with people to enable them to learn new ways of coping with their daily activity by breaking up activities in very specific ways such as learning how to engage in paced activities and to learn various pain management strategies. One such strategy is learning about sleep and sleep hygiene and another is engagement in physiotherapy led by physiotherapists with expertise of working with chronic pain.

Another strategy is reducing physical tension by learning to relax and break the vicious cycle of pain. Learning about deep relaxation, how to deep breathe can help relax the mind and body. Learning about mindfulness can



help a person to live in the present moment rather than worrying about the future or being preoccupied with what's happened in the past. The skills learnt can enable people to live in the present and appreciate what is happening right now. Furthermore, helping people to reconnect with their values, set goals and assess what's working or not working for them can enable them to work towards what they want from life and improve their quality of life.

It is important to learn about pain and to understand your response to pain as this gives you power and enables you to work with the pain rather than fighting it.

My top tips are; Communication, talk to each other. It is important not to bottle up your thoughts and feelings. Aim to build up long term healthy strategies, to learn to live a valued, full and rich life. Remember the holistic approach, look at all aspects, physically, emotionally and socially."

Weekend Round-Up





Making the Most of Modern Life

Robin Mackenzie Spinks, Senior Strategy Manager (Digital) RNIB Business

Robin has worked in the disability and corporate sectors for sixteen years including four years in international development, the private sector and governments in Africa, South Asia and the Caribbean. Robin is also presenter/producer of the popular Access Talk podcast www.accesstalk.co.uk and is Vice Chair of Vision 2020 UK Technology for Life Working Group. He co-authored Real Lives, Personal and Photographic Perspectives on Albinism and he speaks regularly on issues around mobile technology, low vision and personal development internationally.



As a passionate technology enthusiast and a person with low vision, Robin travels extensively promoting the enabling potential of digital technology. His current interests include the accessible smartphone as a life device, voice control and smart wearable devices. He is also strongly focused on using smart technology to improve employability and self-advocacy.

Following, is an edited extract of Robin's presentation at the BBS UK Family Conference.

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"As someone who is attending for the first time, I'd just like to congratulate the organisers of this conference, I think it's been absolutely fantastic.

So my name's Robin Spinks, I do work for RNIB as I was introduced, but what I want to talk to you about in this particular session is enabling possibilities through self-advocacy and through digital accessibility. Often people talk about technology as if it's over there, it's the domain of geeks and it's very high flying and you have to be really capable and outward looking in order to master it. I'd like to blow apart some of the myths around getting started with technology.

One of the things that brought it home to me very recently, that anyone at any stage in life can benefit from using technology, I was giving a talk, a little bit like this to the Macular Society and I was talking about what various apps on mobile phones can do and the various pieces of technology, mainstream and specialist. A lady who was in the audience introduced herself as being 72 years of age, she interrupted and she said, "Can I just ask a question? It's alright for an articulate 40 year old like you to be enthusing about technology, but I'm 72 and it's a lot more difficult when you're 72 to use technology and it's a lot more difficult to get started." So I said,

"That's a fair point, you know, let's talk about that." And just as I said that, another member of the audience got up and said, "excuse me, I'm old enough to be his granny, and I'm old enough to be your mother and I'm 91 and I've just learned how to use a smartphone, so just you get on with it my dear." The 72 year old came and found me at the break and she said to me, "I feel as if I've had a telling off." I said, "Well, I think you have had, but it's all good, let's just talk about it."

So a little bit about me. I qualified as a careers advisor at the end of 1998, I did that for a little while and then I ended up very quickly moving into the world of disability. I've worked in disability for 15 years in the UK, but also in 34 countries around the world, across Africa, South Asia, and the Caribbean, so you might be thinking, hang on a minute, how does a visually impaired guy do that? Around about the age of 30, I got a phone call about a job that involved travelling all over the world, working in the health sector, NGOs and disability NGOs and I thought, I have to apply for that job, so I did, and I got the job. I spent five years just travelling and travelling with 25 weeks a year spent flying around the world supporting projects, doing evaluations and research. Some people that I know and

love very dearly said to me, "Look, you're mad, you're registered severely visually impaired, is it really all that sensible for you to be doing that?" Now, of course this was like a red rag to a bull, I said, "Listen, guys, just watch me, I'm off." They said, "Oh, but what about travelling, what about being on your own and you'll not be able to recognise people. And you might be taken advantage of and you might get kidnapped." I said, "It's true, all of those are possibilities." But you know what, I look at life's challenges as opportunities, I try and think about something as a challenge and it's there to be overcome. I'm a keen cyclist and also a walker and a skydiver. A couple of years ago I decided to chuck myself out of a plane at 13,000 feet, superb fun, I would highly recommend it. Somebody said to me, "Is it not a bit more difficult if you're visually impaired?" I'd never even thought about that, in fact, that thought crossed my mind just as I was about to tumble out of a plane that was doing 190 miles an hour. It's quite a sensation to be in an aircraft at 190 miles an hour when the back door is open. And even more of a sensation when you remember that you're about to be bailed out like a sack of mail.

What I am really passionate about is the use of mobile technology, and particularly smartphones as life devices for people with a disability and I think it gets really interesting when you take it into the realms of mindfulness and meditation. The smartphone starts to become the Swiss Army Knife in your pocket and I'm really interested in its potential for reducing anxiety and facilitating access. One application that I used to get me here today is called **Train Times**, which is a simple little piece of software that runs on your phone, and it enables you to very quickly and very accurately find out where your train is leaving from.

Those big departure boards that you get in train stations, they're not actually designed for humans, they're designed for giraffes. I used to take a little monocular to read it, which used to get the security people really interested. I've had my monocular confiscated on five occasions in different airports around the world. In fact I've actually sat in a little room while it was deconstructed, just to check that I wasn't transporting anything inside it. The wonderful thing about a smartphone is that everyone's

got one, wherever you go, people are glued to their screens; we've become a nation of screen obsessed people from a very early age, so I'm really interested in the potential of that.

There's an application called **Hotel Tonight**. If you've ever found yourself working in a strange location and maybe your day goes on a bit longer than you were expecting, you need to stay an extra day, I've got this application on my phone called Hotel Tonight, and I timed it from making the decision that I need to stay in a place to actually having secured a hotel room and paid for it, I was able to do it in 34 seconds, which I think is pretty quick.

So all of those smartphone helpers that are out there, if you're interested in any of this, then let me tell you, it's not complicated. If you expect something to be difficult and complicated it possibly will be, but if you see it as something that you could overcome then probably you will.

Staying on that theme there's a really cool application for any smartphone that you may have and it's called **Headspace**, which is described as a gym membership for your mind. It is a daily dose of mindfulness to accompany your conflicts or commute; you can switch it on, and have a little 10 minute meditation, no one needs to know about it, you've got your headphones on and you could be listening to anything, you can keep it completely private.

The founder of Headspace, a guy called Andy Puddicombe, has a couple of really good videos on YouTube, he says that we can't change the things that happen to us in life but we can change the way that we experience them. This really struck a chord with me as a visually impaired person because I was actually thinking, this is not targeted at people with a visual impairment but is so relevant and actually the more you think about it, the more you get into it. We are all experiencing things on a daily basis, it's coming at us in the train station, in the cinema, in the doctor's surgery, at work and out and about. We're all kind of ingesting life and experiencing people and places and if we could find out how to do that a little bit more smoothly and a little bit more happily, wouldn't that be really cool, whether or not we've got a visual impairment. I think if you have a visual impairment, it's probably more

applicable because we sometimes experience those things a bit more acutely. We've all had occasions where we think we're on top of the world, we're managing things really well and then something happens and the wheels fall off and you kind of feel like it's a real challenge to do things and actually this is just one little tool that can help you with that.

To finish, there's a little mnemonic that I learned a few years ago at a conference like this, and it really stuck with me and I think it's actually influenced a lot of the things that I've done since. It's a little formula for thinking about life, thinking about technology, thinking about people and situations that you might get yourself into and how you might experience them more positively and a little bit more vibrantly. So when you come across something that feels like it's a challenge or a difficult situation, what you need to do is you need to REACH OUT:

R is about reassurance, we all need that. We need to sometimes give it to ourselves and to people around us, reassure one another that we're all human beings. Sometimes we struggle with stuff and actually that's fine, it's okay, but reassuring ourselves and reassuring one another is really important.

E stands for energy and effort, because you constantly need an endless supply of both of those things. One of the things we can all do is find out what enables us to top up our energy and our effort. Coming to conferences like this, is a great way of topping up your energy and effort, because although you might feel really tired at the end of it, actually it replenishes your reserves, because you spend time with other people who know what it's like to have the condition that you have, which is a hugely important thing to do.

A is for assertiveness and it's really important to separate assertiveness from feeling aggressive and feeling annoyed, as people often do because we're all humans. Assertiveness is about being able to communicate really clearly how you feel and what

you would like to happen. Some of the most assertive people that I've ever met are also some of the nicest people, so there's never a need to be aggressive or to be loud to be assertive.

C is for courage, because you'll need a lot of that. That's about finding your own personal courage to deal with situations and to challenge things.

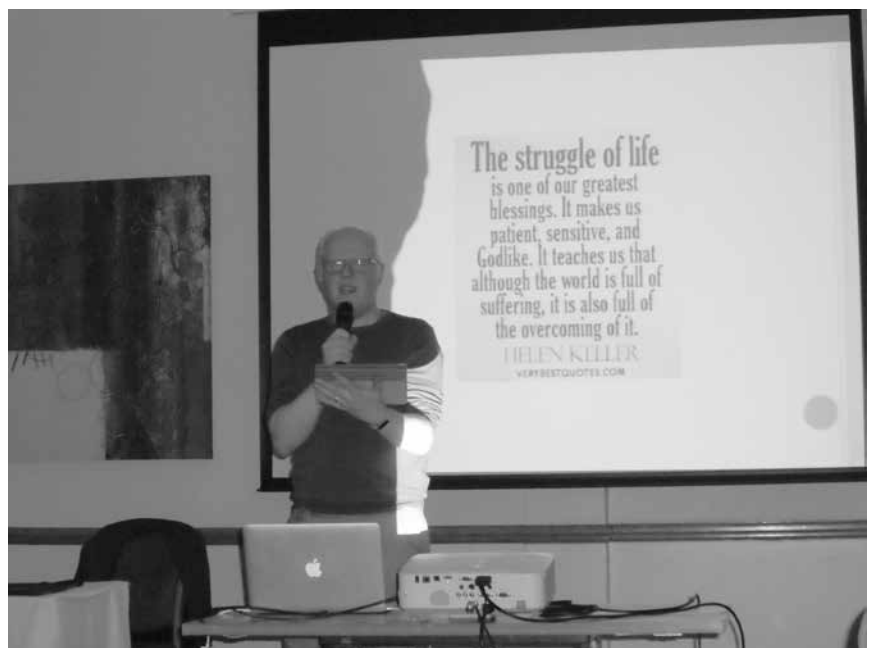
H is for humour because a laugh always helps, it really does. If you get into any kind of disability situation, if you can bring humour into it, and we know it's not always easy to do that, it can be really difficult, but when you can it'll help and it'll make you feel better and it will make other people feel more at ease.

O is for 'over there', that's just remembering that sometimes what we have to do is just distract people and pull their attention away from us if we're feeling like we're being fussed over and things are being too heated.

U is for understanding, because that's what makes the world a better place. The more that we can understand one another, people, places and technology, understanding is absolutely key for everything that we do.

T is for try again, if none of this works you try again.

I'll leave that with you, enjoy the rest of the conference and you know what, above all, reach out and stay positive."



Puberty, Sexual Health and Relationships: support and information for sensory impaired young people

Gillian Leno, PSHE Teacher, QAC College, Birmingham

Gill is a specialist sex and relationships teacher, writer and researcher based at Queen Alexandra College (QAC) in Birmingham, where she works as their full time PSHE (Personal and Sexual Health Education) Specialist. QAC is a national specialist college for young people aged 16-25, with complex learning needs. Gill also works in collaboration with other schools and colleges (and various other professionals and support agencies) to develop and deliver specialist sexual health education and policy. Whilst her work is primarily focused on young people, she also facilitates Sex and Friendship Education (SAFE) workshops for adults with complex needs in the community. Gill's particular interests are around developing accessible and inclusive resources, as well as the importance of sex education in supporting positive behaviour and consent. She also writes for Progress Magazine and can be found on Twitter - @sen_sre. Following is an edited extract from Gill's presentation at the BBS UK Family Conference.



Queen Alexandra College, Birmingham is a specialist further education college for young people of 16 to 25 years, with complex needs; about three-quarters of the students have a visual impairment. The majority of students have got some form of learning disability or are on the autism spectrum as well as having physical conditions which impact on their day-to-day life. I have a very fancy title which is 'PSHE specialist'. Amongst the students, I'm known as the 'Sex Lady', which is not quite so great when being introduced to the parents' forum! That happened recently and there was a long horrified silence, before I explained that I educate about it, not facilitate it and then things went a little bit easier! I teach about online safety, drugs and alcohol because I am teaching teenagers, and they get into all sorts!

I've got nearly 200 reasonably willing students who are enthusiastic about their sex education and making improvements to it. This means I've got a research base with nearly a decade's worth of data to draw on to show that it does work. This is pretty useful when you're out there trying to explain why we should be teaching about sex education and consent to people with learning disabilities, because let

me tell you, it freaks people out.

I do mandatory PSHE sessions which are mostly sex and relationships sessions every week for all of the students, regardless of abilities. We have students who have got very complex needs who may not achieve independence or be able to live without a great deal of support after college. We also have students who have a really diverse range of learning needs, and students who go off to university. All of them have sex and relationships education for the whole time they are there, although for some students that's going to be a lot more in depth and complex than for others. We also do quite a lot of specialist work around gender as well. If, by the time my students leave college, they know that their body is their own, that they have absolute say over who touches it and when and whether they want to be engaged in anything, then I think that's pretty good.

Sex and relationships education is important because we can't achieve independence and go out and be in the community if we can't follow the rules, or know what is appropriate, socially and sexually, when we are in company. If you know what you can and can't do in

public you're going to be a lot safer. If you don't know what something is, or if you don't understand what's happening to you, how are you going to disclose about it?

Sex education is important because it allows people to make informed choices about their sexual health and reduces the risk of sexual abuse, sexually transmitted infections and unplanned pregnancy. One of our basic fundamental human rights is to make a choice and that applies to our sexual health rights as well. If you don't give people information in a way that they can understand and use, they may not be able to make any positive choices for themselves. I am a safeguarding designate so my work is solidly grounded in the fundamental need to make sure that people know what their rights are and that they can exercise them in a positive way. This is important when working with and for people who have additional needs, particularly additional physical needs because if you are somebody who receives a lot of personal or intimate care, you may feel that your body kind of doesn't belong to you anymore, it is somebody else's to care for, somebody else's to sort out, someone else tells you when to go to the loo. I think it's very important to talk about our bodies, it gives a sense of equality for everybody and autonomy that actually this is yours and you get a say over what happens. It also improves social skills and expression.

I've had many disclosures over the last ten years, when someone has said, "actually this happened to me and I wasn't happy about that." Or, "this has happened to me in the past and I didn't know what that was." Or, "this is happening right now and actually I see that this isn't something that I want to continue." This makes sex education an incredibly powerful, protective safeguarding mechanism. It establishes and reinforces appropriate behaviour, which is why it's so important that it's done in classes amongst peers in context. I genuinely believe it's much more powerful doing it that way than in a one-to-one lesson.

Much of the information that's presented for young people and people in general around sex and relationships depicts straight, white,



non-disabled people, all smiling and not experiencing any pain or awkwardness, which is not really how it works. So getting your resources right is very important and they need to be age appropriate and relevant. They also need to be inclusive and tactile in some cases so that everybody can use them, because it's something of a challenge when you're trying to deliver something abstract to people who may not be able to use visual cues. There are not many resources available but this is something that is being worked on and new ones are coming out all the time.

Formal and informal sex education messages need to be matched up so that people get consistent, ongoing information. I'm the formal aspect, the person standing at the front with the lesson plans, the bit they try to forget as they slope out of the room and run away. The informal aspect is what everybody else is saying: family, friends, media, social workers, support workers and so on. Being willing to talk about it is a pretty good start because otherwise things don't stick, do they? The biggest barrier in teaching sex education, is the fear of leaving young people more vulnerable so it's important that everybody is clued up or misinformation can be conveyed and remembered.

Understanding boundaries and appropriate relationships is a concern and unfortunately, unless we start treating it as a mandatory part of the curriculum, there's not going to be enough training for teachers and support workers to rectify that. We have to have a 'no secrets'

culture around talking about bodies, sexuality and relationships. Unfortunately, we know that that is often not the case, particularly talking about it with people who have got additional needs. There has to be an environment where people can talk without being frightened or feeling ashamed of themselves if they have to bring up a problem or discuss it; frank conversations and no glossing over things is important. The reason we know that this works is because students at our college have a much higher rate of self-disclosure; they know when they've been at risk and will talk about it.

It is incredibly important to focus on the positives and pleasures of growing up and relationships, when we work with people with additional needs, whether they are physical, learning or sensory. There is often this language of fear, that sex is something that's done to you, that there's terrible risks involved, that it's not something you're going to enjoy and people are unlikely to speak up if they think that something's not right, that's just how it is. We've found that when we do baseline assessments of students coming in, the words they use to talk about sex are almost universal and negative, which is really worrying. It means that there's a very high rate of young people

saying, "I'm not allowed to do this." Or, "Don't tell my mum that I've got a boyfriend or girlfriend." Why not? I'm sure they'd be quite happy to know actually. There's a lot of secrecy that's brought on by that kind of attitude so it's important to be body positive whenever you do sex and relationships education.

Online safety is incredibly important. If you are a parent of a young person, you know it's a breeding ground for terrible things. You don't have to be on Facebook, but you do at least need to know how it works because if you don't, you can't safeguard your child. It's very important that we are on top of making sure we are as literate as they are, and I think that's something that parents are getting better at.

Useful Links:

Brook: the sexual health charity for young people under 25: www.brook.org.uk

ThinkUKnow: the home of CEOP (Child Exploitation and Online Protection Centre), promoting online safety for young people, with parent and teacher spaces too: www.thinkuknow.co.uk

FPA (Family Planning Association): the sexual health charity: www.fpa.org.uk

Two Sides of the Same Coin: a personal perspective

Julie Sales

Our precious daughters, Danielle and Hollie, both have BBS1 but are experiencing different journeys. Both were born with six fingers and six toes, and this is probably the only aspect of the condition which they share. Their diagnosis in 1997 by Professor Phil Beales brought mixed emotions, relief that we had answers, but dread for the future. Coming to terms with the eye condition was initially the hardest to bear but over time this has changed as both girls have had to deal with many other health issues.

Danielle was a normal delivery at 41 weeks. We were told not to worry as 1 in 100 babies can be born with extra digits and at the time we said it was better to have too many than not enough.

These were removed when Danielle was four months old. We noticed from a very early age that Danielle was not meeting her milestones, but thought we were being overprotective first time parents. Danielle has learning difficulties, and as a child she had behaviour problems and co-ordination/dyspraxia problems, such as not walking until she was nearly two years old, not being able to climb, ride a bike or walk up steps and she also found PE, catching a ball and tying up shoe laces very difficult.

Danielle has always suffered with weight issues and even at the age of one, we were told that we must have been over feeding her, as her weight just wouldn't come down. Poor girl

didn't even have a sweet or a piece of cake until she was a toddler. I remember when she was little that I daren't tell her the time, as if I said 12 o'clock she would say oh good it's lunch! We always try our best to live a healthy life style. Those with BBS can lose weight, but it is a lifetime struggle for them, with many medical implications attached.

From an early age, Danielle has suffered with psoriasis and has had numerous operations for continuous ear problems. At nine years old, she was diagnosed with chronic fatigue and for two years Danielle was in school for mornings only, and rested in the afternoons. To this day both girls still have to be careful not to overdo it, or they become ill. Attendance slowly increased but this impacted on her social skills and confidence and she was quite withdrawn for a few years, but Danielle persevered and worked very hard. Danielle had a Statement from Year 2 and left school with six GCSEs. She successfully studied photography at college and University, graduating with an HND 2:1 grade. Seeing her in her graduation gown was the proudest moment.

Three years ago Danielle was told she had slight damage in her eyes which could lead to visual impairment. This has been a major blow for us all, as we always hoped that Danielle may be one of the lucky ones that missed this. This was probably wishful thinking, but with future research you never know. Danielle was also diagnosed with Polycystic Ovaries, which affects her skin and she suffers badly with infected boils, even to the extent of being hospitalised.

Throughout her life Danielle has felt she is on the autistic/OCD spectrum, experiencing emotional sensitivity and outbursts. It was hard to help her understand that she is beautiful as she is but with continued support we helped her to minimise these tendencies.

On a positive note Danielle is now working part time, has a boyfriend and she is still driving her car. She has such a bubbly personality and enjoys socialising, cooking, bowling, cinema, afternoon teas, clothes shopping and spending time with her family.

Hollie was found to have a heart condition at our 19 week scan which probably meant she



would need an operation at two days old. At 28 weeks, I had my first bleed, was in hospital for two weeks and was told to rest. Quite difficult with a three year old! At 33 weeks I had a major haemorrhage, and was given a transfusion of two pints of blood but as the last drop went in the whole lot bled out and I was rushed into surgery. Three hours later after another five pints of blood, Hollie was born six weeks early, weighing 4lbs; we both nearly didn't make it. It was a very traumatic time for us all.

Hollie didn't need the heart operation but her plumbing works differently and she has a loud murmur which is monitored regularly. Constant chest problems from four months old, including septicaemia, pneumonia, and asthma, resulted in many stays in hospital.

From the age of five, Hollie had severe arthritis in her legs, sometimes resulting in being wheelchair bound. After many years on high dosage painkillers, we finally got the pain under control with regular acupuncture.

Hollie's eyes began to deteriorate at ten years old. She started having problems with night vision, and everything became blurry; she was registered blind in 2007 and had a Statement of Educational Needs at school. Hollie went

to a secondary mainstream school where she coped very well for a couple of years with support. Unfortunately, after she had three sight losses in an 18 month period, she understandably didn't cope very well and she became very upset and confused. Numerous neurological tests all came back clear. She went from using enlarged print to not being able to see at all.

In January 2009 it all became too much for Hollie. She had a nervous breakdown, and was diagnosed with severe ME, which included visual disturbances. Hollie lost her memory and didn't know who we were or how to do anything. She couldn't get dressed, eat, bathe or even get out of bed without instructions. It was like teaching a toddler all over again. She was constantly exhausted, and on doctor's orders was only allowed to do an activity for ten minutes and then rest. She wasn't allowed to go back to school until she could do an activity for more than one and a half hours without feeling ill, so was home tutored for five years in IT and Braille. It was an extremely traumatic time for her and the family, but Hollie has slowly improved over the past seven years and was eventually able to attend a health needs school for two years where she could manage around an hour's study, and pursue other activities.

In October 2011, after many years of having bowel problems, Hollie went through various tests which showed the last 12cm of her bowels didn't work properly. Following a bowel biopsy, whenever she ate, she was in extreme pain lasting 22 hours and she lost three stone in weight. After six months they inserted a nasal gastric tube but she was still not eating, still losing weight and bordering on anorexia, so she was fitted with a PEG and is now fed through her stomach. Not being able to participate in something as normal as a family meal and being in constant pain and unable to eat for four years is understandably very upsetting for Hollie.

After numerous tests, they found that she has a nonrotation of the bowel, her appendix and gall bladder are in the wrong place and she has three spleens. They have finally given us a diagnosis of functional dyspepsia, which is a problem within her stomach causing extreme

nerve pain. They can't find anything medically they can correct and if the strong pain killers she is now on don't work, this could be for life. We are waiting to see further specialists to discuss nerve block treatment or a spinal stimulator.

Hollie's autoimmune nervous system is not functioning properly and she now has fainting fits, and is undergoing heart scans and tilt tests to check for postural tachycardia syndrome. She also has a diagnosis of Polycystic Ovary Syndrome.

Through it all Hollie has tried her hardest to stay positive, and we are proud to say that she started a 'prep for life course' in September 2014 at a local college and now wants to do a catering course there. In the last couple of years, she has become very confident using her cane and now walks from the college to our local train station where I meet her, which is something I never dreamed would happen.

We are very proud of her as she works so hard to stay positive and enjoy other areas of her life, like playing football, going to watch Liverpool with her dad, concerts, bike riding, swimming and like Danielle, cooking for the family, even though she can't eat. As a family we have had to deal with so much illness and heartache and without the support of our special family and friends, I am not sure we would have got through it.

As parents we would all go to the ends of the earth to make it easier for our children without sometimes looking after ourselves. Kevin has always been my rock and I am sure he has found it hard sometimes having to cope with three women, but he has supported me through thick and thin, breast cancer, gynae and bowel operations to name a few, and I couldn't cope without him. No wonder the poor man has no hair left.

We are so very proud of both Danielle and Hollie. Through adversity they have turned into two beautiful, confident and caring girls with great work and family ethics. What more could a parent ask for. As you can see, although they both have BBS1, they are two sides of the same coin, who suffer with this disease so very differently.

My First Time at the BBS Conference

Tanja Chaudhry

Hello to my lovely BBS family. I want to say a few words about our experience at our first BBS UK Family Conference. I came with my husband and my three children. The youngest of them has BBS10. When we first arrived and parked our car, we got very emotional. It was very hard seeing some of you using a cane or even seeing how much support some of you needed, thinking that could be our future as well. But all of those mixed feelings disappeared very quickly. After the first ten minutes I already felt much better. I've never seen so many lovely people in one place.

Everyone made us feel very welcome. Some of you walked up to me and said, "Hello, you are Tanja right?" because you remembered me from the Facebook support group. It felt so nice not to feel like an outsider and made me feel a bit posh at the same time! It didn't take long to get into conversations and the best part was you all knew what I was talking about, I did not have to explain myself over and over again. The conference itself was very meaningful but also emotional at the same time. I learned a lot about my daughter's condition. Knowing that lots of research about BBS is going on has given me a little hope for the future.

My children had a very enjoyable time as well. I did not have to worry about them when they went to Drayton Manor and my youngest was looked after in the crèche. It gave me and my husband a bit of time to ourselves. The activities for the kids were very amusing for them, they joined in the fun while I was putting my baby to bed. It was so nice to be reassured that my kids were looked after by lovely people who kept them busy with different activities. Not only for the kids but there were lots of things going on for us as a family. It was a shame that the weekend was over so quickly because we were enjoying ourselves a lot. I am so glad that we took the decision to attend the BBS Conference because it felt like we had found our long lost family. I'm already looking forward to next year's conference and many more in the years ahead. Thank you to you all for making our experience so special.



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