



BBS UK

Conference Report 2015

Bardet-Biedl Syndrome UK

Registered Charity No. 1027384 and SCO41839



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Foreword

Conference 2015 will be remembered as a particularly emotional one for several reasons, not least because Phil and I bid farewell after 21 years involvement with the Society. Tonia, Julie and Kevin also retired as Trustees, however will remain closely involved with the day to day running of the charity for the foreseeable future. Steve Burge was elected as Chairman and Richard Zimble as Vice Chairman and we welcomed Laura Dowswell (Treasurer), Graham Longly, Rob Hymers, Dianne Hand and Stefan Crocker onto the committee, joining Allan Clark, Emma Oates and Claire Eccles to complete the LMBBS Board of Trustees for the coming year. Phil and I wish the new team every success. Finally, it was proposed and agreed that the name of the Charity, LMBBS, would be changed to BBS UK. Once again, we had an excellent array of speakers, facilitators, meetings, receptions and activities spread throughout the weekend, ensuring there was something for everyone. The crèche and trips to Drayton Manor and Bowling were once again a success thanks to our dedicated volunteer care team who return year after year. Professor Beales brought delegates up to date with all the latest news on research in the field of BBS. Daniel Evans gave an inspiring personal perspective about living with BBS. We heard an excellent presentation from Professor James Bainbridge on Gene Therapy, followed by Abigail Mance's presentation on communication difficulties in children with autism. Ray Perry gave us an update on Benefits and Tonia Hymers ended the morning with a personal perspective. Following closing remarks, Phil and I were presented with gifts and cards for our retirement. All presentations and reports from the weekend can be found inside. Some of the presentations are also available to listen to in full on the BBS UK YouTube Channel. Following a luxury buffet lunch, the afternoon was dedicated to workshops and resource tables and provided the opportunity to speak with Birmingham Action for the Blind, The



WESC Foundation (Specialist Centre for Visual Impairment), Royal National College for the Blind, Hereford (RNC), a Benefits Advisor, a Transition Co-ordinator and BBS Clinics Dieticians, in addition to the morning's speakers in an informal setting.

The weekend also provides us with an opportunity to raise some funds and our grateful thanks go to everyone who donated or made items and took time out over the weekend to oversee the fundraising stalls or supported them. Sandra Dale and her friends made an excellent selection of homemade cards (I for one have my cards purchased for the months to come), the tombola and raffle were once again well supported and our delightful up and coming young fundraisers, Emmy and Holly Anstee sold handmade items on their own fundraising table. Our grateful thanks to everyone involved.

The success of the BBS UK Conference depends on the continued support of our members and our amazing team of professionals, carers, volunteers and fundraisers. We know how important this weekend is and how much it is appreciated by everyone who attends, especially newly diagnosed families. I have seen many changes over the years, but one thing that will never change is the support and friendship we give one another in what at times can be a challenging world.

Phil and I wish you all every success for the future and we look forward to seeing you at future conferences.

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.



“Prior to 2010 we convinced the Department of Health in England to fund clinics specifically for patients with Bardet-Biedl Syndrome. The argument for this was that BBS comes with so many unique problems that we needed a unique or bespoke clinic to be able to deal with each of those problems. We developed a multi-disciplinary clinic, where we have at least seven specialists at each clinic. In London we have one at Great Ormond Street Hospital, where we only see children up until about 17 years of age. After that, they are transitioned over to the adult clinic, which is based at Guy’s Hospital. In Birmingham we have a similar arrangement, whereby we have the adult clinic at Queen Elizabeth Hospital and the children are seen at Birmingham Children’s Hospital. We have been very fortunate in having the continued support of the Department of Health (now NHS England), who fund this particular clinic, and we are pleased to say that they are going to continue to run it and fund it for the foreseeable future.

I am really pleased with the way the clinics have been going and the feedback that we have been getting. We have got the support of the family representatives from BBS UK and we have multiple volunteers who also contribute to these clinics; without that support these clinics

would not be the success they are. Every year we have an ‘audit day’, where we discuss how the clinics are progressing, the good points, the bad points and how we can improve things. The main objective this year on our agenda has been discussions around developing a patient management system with a company called Certus.

We want to be able to record all clinic activity, so when a patient is due in clinic, we know when they are coming, their latest results, including weight, height and so on, any medication they are on and we want this all to be in one place. We will also be able to help implement what we call the care pathway, which for rare conditions



in the UK is a government-led remit. We will be able to see what is supposed to happen for each patient and where they are on their care pathway. Eventually we hope to be able to include things like x-rays, ultrasounds and other images as well.

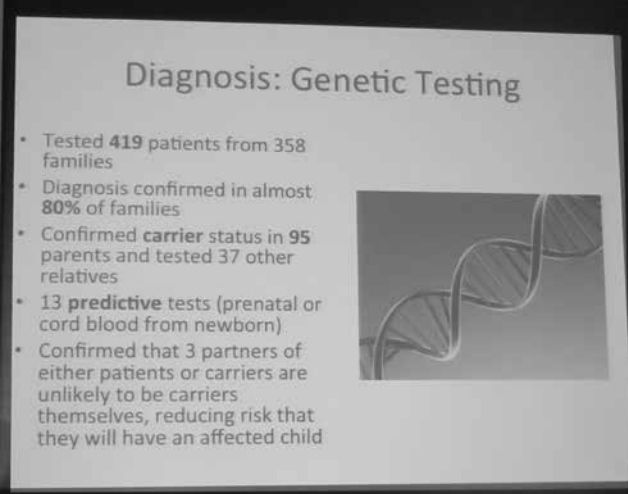
We are going to develop a service 'dashboard', where we will have an overview of the four clinics and all the patients who are attending. There will also be a patient 'dashboard', where we will be able to see the notes from the consultations, where a patient is on the care pathway, what medication they are on, whether the blood pressure has gone up or down, whether the weight is stable and so on.

Certus is developing this system to enable patients or their family members to access their own information, their own appointments, and even communicate with the team. We hope to roll this out for first testing later this summer and it should be up and running by the autumn.

Moving on to another component of our clinic, the genetic testing service, 419 patients have now been tested, from 358 families that attend BBS clinics. That is by far the largest group of patients in one place who come to this type of clinic and have been genetically-tested. Of these, the diagnosis has been genetically confirmed in 80% of the families. We have now identified about 19 genes, which is more or less all of them, which means we can now deliver an incredibly robust diagnostic service. We can test for carrier status in the family members of those who have BBS and we can also use this test for pre-natal testing; we've done this successfully in about 13 or so cases now.


The age of patients who have been tested in the clinic ranges from six months old right the way through to 59 years. The average age of diagnosis has been reduced from around 14 years in 1999 to around 9 in recent years and genetic testing has now reduced that even further. 94 patients have now been diagnosed before the age of 9 and 24 patients have been diagnosed before the age of 3. For very young children, before visual impairment is detectable, it has been very difficult to make that diagnosis, so this has been a major advance in this particular area.

So that brings me onto the activities that we've been undertaking in my research lab in London at University College London (UCL). I am based



Diagnosis: Genetic Testing

- Tested **419** patients from **358** families
- Diagnosis confirmed in almost **80%** of families
- Confirmed **carrier** status in **95** parents and tested **37** other relatives
- **13** **predictive** tests (prenatal or cord blood from newborn)
- Confirmed that **3** partners of either patients or carriers are unlikely to be carriers themselves, reducing risk that they will have an affected child



at UCL, Great Ormond Street Hospital, and also at Guy's Hospital. We have assembled a cilia-disorders lab, and have about fourteen people actively working in various areas. The majority of people within the lab are working on, not only understanding what the causes of Bardet-Biedl Syndrome are and why each of the problems develop, but also towards finding therapies for this condition.

A lot of people are still working on understanding the variation and progression of the disease, our focus is on the potential therapies we could develop. The pharmaceutical industries are really interested now in rare diseases. They couldn't care less about five years ago, they were more interested in the diseases that more commonly affect the majority of people in this country as that is where they saw the money being made. They are now turning their attention to conditions like Bardet-Biedl Syndrome because they see that if they get involved in our condition then they might be able to understand better other, more common conditions, and also provide new drugs for those conditions.

Moving on to gene engineering or gene therapy, the idea of gene therapy is not to replace the faulty gene but to augment it, to insert a normal copy of the normal gene and by doing so hope that the normal copy of that gene, will produce the protein (because that's mainly what genes do) that is missing or is faulty. We are looking at whether it is possible to prevent, slow down or delay the visual loss in Bardet-Biedl Syndrome by inserting copies of a functioning gene. In order to look at this we have made a number of mouse models of disease. The normal gene is injected into the

back of the eye and although it is very early days, the early results suggest that in the treated eye, the activity is retained.

Bardet-Biedl Syndrome doesn't just affect the eye, it is a multi-organ disease and if we are going to do gene therapy, wouldn't it be better to give that therapy via injection, so that it reaches all parts of the body? This is what we are aiming toward and some colleagues at University College London, Doctor Ahad Rahim and Professor Simon Waddington have already developed the proof of concept. Using a virus to carry the gene into the cells, they have been able to inject the virus into mice and show, using a coloured tag, that the virus is getting to all the relevant parts of the body. The clever thing is that with this particular system, and using these particular viruses, we are able to cross the blood/brain barrier. We want to exploit this opportunity to see whether we can deliver the BBS gene to all the different parts of the body.

To finish off, I want to talk about an area which is very new and relatively unpublished. We know that some BBS patients have some learning disabilities and others have some problems with memory and these aspects of the brain have not been studied before in Bardet-Biedl Syndrome. This is an area we really want to advance. In our lab, Dr Sonia Christou-Savina has been working really hard on trying to understand what is going on in the BBS brain and has been looking at the brain cells in mice that have Bardet-Biedl Syndrome. On each of our brain cells are finger-like appendages called dendrites, and on these dendrites there are little buttons called synapses. The synapses form the connections between the brain cells and both the number and function of synapses is important. In the Bardet-Biedl Syndrome mice, several of them have reduced numbers of synapses, so there is less synaptic activity, less connections and therefore less opportunity for communication between those cells to happen. This gives us a starting point to be able to think about how we might be able to modify some of these learning difficulties and Dr Christou-Savina has gone one step further and thought about how you might actually do this in the simplest form. It has been known for a number of years that simple activities, such as increasing exercise on a daily basis, can stave

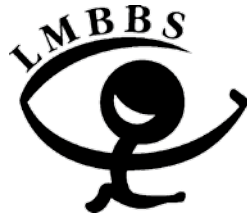


Dr Forsythe spoke about the BBS Memory Study

off dementia and can improve our learning ability. Dr Christou-Savina put the mice onto a treadmill and has been able to show that the number of synapses in the BBS mice has increased back to normal, compared to the ones that are deprived of exercise. We are hoping that this is something that we can also benefit from as humans, all of us, but in particular the Bardet-Biedl Syndrome community."



BBS Research Team: Dr Forsythe, Dr Hernandez-Hernandez and Dr Christou-Savina



Annual General Meeting

The Hilton Hotel, Northampton
18th April 2015

Apologies

Apologies were received from Lindsay and Grant Mapley, Marie and Mark Hughes, Graham Lilley, Trevor Frounks, Ellen and Stella O'Rourke, Claire and Lee Eccles, Kjell Arne Høeviskeland, Richard Oedegaard, Chetan and Aruna Meshram, Emma and Steve Oates, Vicky Merchant and Wayne Fitzgerald.

Minutes of 2014 AGM

The minutes of the last AGM of the Laurence-Moon-Bardet-Biedl Society, having previously been circulated, were confirmed to be a true record of the proceedings.

Election of Honorary Officers

Phil Humphries, Chairman and Chris Humphries, Conference and National Co-ordinator both retired as Honorary Officers. Kevin Sales, Treasurer, Julie Sales, Secretary and Tonia Hymers, Newsletter Editor also retired as Honorary Officers, however they will continue to perform their day to day duties for the foreseeable future. The remaining Officers, Steve Burge, Vice-Chairman and Emma Oates, Fundraising Co-ordinator were eligible for re-election. Nominations were received for Steve Burge as Chairman, Laura Dowsell as Treasurer and Emma Oates for re-election as Fundraising Co-ordinator. No other nominations had been received for these positions and all were duly elected unopposed.

Nominations were received for Richard Zimble and Graham Longly for the position of Vice-Chairman and/or trustee. Voting was by ballot and resulted in Richard Zimble being elected as Vice-Chairman and Graham Longly as Trustee.

Election of Committee

Of the current trustees, Richard Zimble and Alan Clarke were due to retire, however they were both eligible for re-election. Claire Eccles

had a further year to run in her present role. Nominations were received for Alan Clarke for re-election as trustee and Stefan Crocker, Dianne Hand and Rob Hymers for election as trustees. No other nominations were received for these posts and all were duly elected unopposed. The Chairman welcomed the new Trustees to the Board.

Chairman's Report

The Chairman read out his report:

"It has been a year of goodbyes.

Twelve months ago at our last conference, Stephen Sherwood, whom many of you will have known from attending conferences with his father Nick, from an early age and latterly as an adult attending on his own, enjoyed a great weekend here with his friends. I can still see his smiling face as he left on Sunday morning. We were therefore shocked and saddened to hear shortly after, that Stephen had passed away in his sleep. Along with other LMBBS members, I attended his funeral in Herefordshire, which was a celebration of his short but full life. Stephen will be sadly missed by all who knew him. Nick is with us today and I convey all of our heartfelt condolences to him and his family.

In June 2014, we received the sad news of the passing of Terry Crotty, who had been Vice Chairman of the Society and Director of LMBBS Clinics before his resignation in 2012. Terry volunteered with the Society for 8 years along with his wife Anne who had been our Fundraising Officer until her retirement in 2013. We send our condolences to Anne and the family.

In October we said goodbye to Ali Bokhari. Many of you will know Mr & Mrs Bokhari and their family, who attended many conferences over the years. Condolences were sent to the family.

In January 2015, we were saddened to hear that Mrs Anuradha Kochhar, a lovely gentle lady who regularly attended conferences with her son Abimanyhu, passed away in hospital after a short illness. Condolences were sent to the family. We hope that Abi will attend future conferences with his family."

The Chairman asked the delegates to take a few moments in silence to remember their friends.

The Chairman continued by extending a warm welcome to all new members and acknowledged that first visits to conference can be daunting, however he hoped that many new and lasting friendships would be made. He continued:

"My thanks to all those dedicated to furthering the aims of the LMBBS, in particular Phil Beales and his team. I sometimes find it hard to believe the advances made over the past 22 years. We are now in our 5th year with BBS Clinics, with numbers rising annually. To Tonia, Julie and their team of volunteers and all professionals involved, thank you for this continued and valued service which has made such a huge difference to the lives of our members.

Thank you to my committee for their outstanding commitment throughout the year. My thanks go to Drina and Michael Parker, who represent the Society on the Board of the Ciliopathy Alliance. Drina and Michael, along with their son Richard, attended the Ciliopathy Conference in Paris on our behalf in October 2014. To our team of volunteers, who work silently behind the scenes representing us at Sight Village in London and Birmingham, attending day courses and conferences and of course our fantastic team of Childcare volunteers, our heartfelt thanks to you all.

A huge thank you, which I know you will all endorse, to Chris, Tonia and Julie for the organisation of this valuable weekend. I am only too well aware of the amount of work that goes into this successful event, as will Robbie and Kevin (Tonia and Julie's husbands). Finally, thank you to the Hilton Team who look after us so well, catering for our every need.

I would like to welcome on board Jacquie Kearnes and Angela Scudder our Child Development Officers, employed as a result of our successful Jeans for Genes grant, they are both proving to be an asset in their work with

families, clinics and future activities.

We have had another year of generous donations and fundraising and a huge 'Thank You' goes to all concerned."

Treasurer's Report

The Treasurer presented an overview of the Society's accounts:

"For the financial year 1st January 2014 to 31st December 2014, the Society received an income of £35,319, which was a decrease in income of around £6,500, compared to 2013. The Society's expenses for the same period reduced to £38,420, a decrease of just over £4,000 compared to 2013. The decrease reflects the reduction in the number of publications; the Society did not produce a winter newsletter and worked on a medical booklet instead. Overall, the charity made a loss of £3,000 in 2014.

Also noted in this year's accounts, we have over £1,800 of unsold merchandise. The Society aims to sell as much as it can at this year's conference, at a competitively-reduced price. The marathon bond as shown in the accounts of £480, relates to our silver bond place in this year's Virgin London Marathon, which is next weekend. We've received our first instalment of our grant for Jeans for Genes and have employed two part-time Child Development Officers. The second instalment was due in March 2015.

In financially-changing times every charity relies on its regular donors and regular income to enable the core of the work to continue. We have our Friends of the LMBBS scheme, with many of our members making regular donations on a monthly basis and holding fundraising events. Around £3,000 each year is paid into the charity account by standing order by our 'Friends'. We aim to increase the awareness and subsequent membership of the scheme, which will strengthen the charity financially. Although there are several fundraising ventures in the pipeline, Conference 2015 will leave the charity short of funds and we are unsure at present whether Conference 2016 will be a weekend event.

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 summary the Society's main expense is always going to be the Annual Family Conference, however we have seen the costs reduce quite considerably over the last year. Our second largest expense is always going to be the distribution of our publications. We produce two newsletters and a conference report annually and all editions are available in hard copy, audio, CD, via email and on the web page. The reproduction of leaflets is also a considerable expense. As a committee we are truly grateful to all our volunteers and fundraisers for their continued support of the charity, as without their concerted efforts we would be unable to meet our goals and objectives."

The Chairman thanked Kevin for his contribution to the charity, as treasurer and trustee over the years.

Appointment of Auditor

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

Any Other Business

It has become apparent in recent years that we need to change the name of the Laurence-Moon-Bardet-Biedl Society. The medical

establishment consider the condition to be Bardet-Biedl Syndrome and all referrals are made in this name. It is therefore confusing when patients, family and medical and educational professionals are trying to find information and we are called LMBBS. The present Committee therefore proposed to change the name of the charity from Laurence-Moon-Bardet-Biedl Society to Bardet-Biedl Syndrome UK. Voting was by a show of hands and the motion was passed unanimously.

Agreement was also required for the subsequent changes to be made to the wording of the constitution. Laurence-Moon-Bardet-Biedl Society will be replaced with Bardet-Biedl Syndrome UK, LMBBS will be replaced with BBS UK, and the Society will be replaced with either BBS UK or 'the charity'. 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 date of the next meeting was set for Saturday 23rd April 2016.

The full LMBBS Trustees Report for 2014, including accounts, is available to download from www.lmbbs.org.uk or from the Charities Commission.



Steve Burge
Chairman



Richard Zimble
Vice-Chairman



Laura Dowswell
Treasurer



Emma Oates
*Fundraising
Co-ordinator*



Allan Clark
Trustee



Stefan Crocker
Trustee



Claire Eccles
Trustee



Dianne Hand
Trustee



Rob Hymers
Trustee



Graham Longly
Trustee

Promoting Vision by Retinal Repair and Regeneration

Professor James Bainbridge MA PhD FRCO phtth

Professor Bainbridge aims to develop effective new treatments for retinal vascular and neurodegenerative diseases. His programme of research extends from the laboratory investigation of mechanisms of disease in experimental models, through preclinical development of novel therapies, to clinical trials of new medical and surgical interventions. He performed the World's first gene therapy for inherited blindness and Europe's first stem cell transplantation for macular degeneration. In 2012 he was appointed Chair of Retinal Studies at UCL and in 2012 was appointed NIHR Research Professor. Professor Bainbridge's presentation covered the new treatments that can be expected for the sight-impairment caused by BBS, and other similar conditions, in two key areas: gene therapy, and cell therapy. The exciting potential of the treatments, as well as their challenges, and possible side effects were explained.



Professor Bainbridge began his presentation with an introduction to gene therapy in the eye. The eye, in particular the retina, has many advantages as a target organ for gene therapy: it is small, so fewer genes are required, and it is self-contained, improving the chances of the genes staying there, and not spreading round the body, where they could cause side-effects. Our eyes are essentially spheres, similar to cameras, with the retina being the 'film. In reality, the retina is a layer of light-sensitive cells that line the back of the eyeball, and consists of a number of different types of cells. The two most important for this therapy are: photoreceptor cells, and pigment cells.

Photoreceptor cells are the light-sensitive cells, which receive light and transform it into electrical energy, causing nerve impulses to be sent down the optic nerve to the brain, enabling us to see. In many inherited eye diseases, vision is affected because a particular gene, which is responsible for many of the numerous proteins needed for the normal function and survival of this photoreceptor cell, fails to be inherited. This is the case in BBS.

It is in the cilium of the photoreceptor cell that the gene defect has its main impact. In these cells, the cilium acts as a transport system, moving proteins from one part of the cell to another. Gene defects in the cilium mean critical proteins cannot be transported to the light sensitive part of the cell. There are two

different sorts of photoreceptor cells; rods, and cones. Rod cells are used to see in the dark, and to some extent in dim light. The cone cells allow us to see in normal daylight, and to perceive colour. The BBS gene defects that cause problems with the transport system in the photoreceptor cells initially affect the rod cells, causing difficulty seeing in the dark, and with peripheral sight. As the disease progresses, the cone cells are affected, meaning the ability to see in normal daylight, colour vision and fine detail are impacted.

Changes in the retina can be observed with a powerful microscope, which can identify individual cone cells. This enables the course of the disease to be studied, and to see how new treatments bring potential benefits, and any side effects they may cause.

Professor Bainbridge is working on two types of potential therapies: gene therapy and stem cell therapy.

In **gene therapy**, cells that have a defective gene can be given a new, normal working copy of the gene. Modified viruses are used as a packaging system to carry the new gene to the cell where the defect is. The virus containing the gene is injected into the retina, using a long surgical needle, through the front of the eye, to the cells underneath the retina, where the genes are needed. It is a delicate operation, that takes about 1 hour. One disadvantage of the procedure, is that it causes temporary

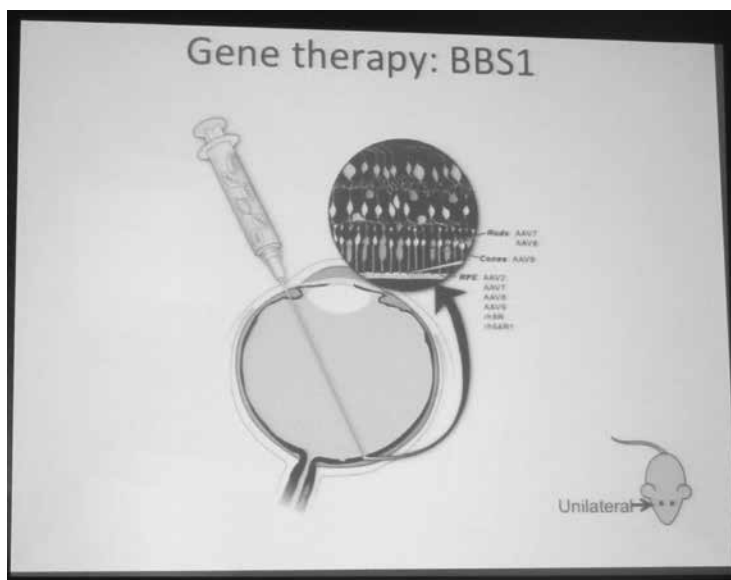
detachment of the retina, which can cause loss of sight, and could cause possible side effects. In Professor Bainbridge's team, they have seen no immediate side effects, in terms of function, but are aware that cells under the retina can be harmed. In the future, it may be possible to design an improved procedure to deliver the genes.

This procedure has been shown to improve sight, even after a few months, in one particular condition that has been trialled. The area of the retina that has been "infected" by the virus shows increased sensitivity six months after surgery, and this is still present, even after three years. The patient's other eye, that does not receive the working copy of the gene, shows no such improvements. Unfortunately, vision does not improve in all patients, there seems to be wide variations, and the recovery is not always sustained for as long as would be liked.

In theory, retina cells stay with us our whole life, so a long-lasting benefit should be possible, as seen in some animal experiments. However, in humans, the peak improvement in sight is not maintained as well as in animals. It is suspected that a bigger dose of the new gene is required by the cells where it is missing and this would bring a more reliable, persistent, and sustained benefit. In the condition that has been studied, the improvement has only been seen in the rod function (better night vision), but no change in daylight vision (cone cells). More work still needs to be undertaken. It is suspected that the same would be true for BBS.

A video was shown, of an interview with a family whose son had undergone the procedure described above. Similar to BBS, the condition this child suffers from causes sight to deteriorate with age, possibly leading to blindness. The outcome of the operation was positive, the parents say the child is "more himself", and is able to move around more easily. They said deciding to operate was an easy decision, they felt they had nothing to lose.

In summary, the trial has proven that gene therapy can improve some aspects of sight. The two key challenges are to demonstrate a sustained long term benefit, and to try and protect daylight sight. The group have already designed and manufactured a new version of the virus carrying the gene, which it is hoped will



be more powerful, and lead to sustained sight improvement.

One significant limitation of gene therapy, when used to treat photoreceptor cells, is that due to the progressive nature of the eye condition (like BBS), the photoreceptor cells gradually die, meaning the new gene has no target, and brings no benefit. An alternative is trying to regenerate these cells, using stem cell therapy. Stem cells are cells that can give rise to many different types of specialised cells in the body (including retinal cells), and can also renew themselves. There are sources of stem cells in both embryos, and within the adult body. In Professor Bainbridge's lab, stem cells have been used to generate light-sensitive photoreceptor cells, which have then been transplanted into animal models, including mice, where photoreceptor cells are lacking. The new cells have been shown to integrate into the retina. There is evidence to suggest that these cells improve the sight of the animals. This process has yet to be tested in humans.

However, underlying pigment cells have been successfully transplanted into humans: these pigment cells support the survival of the photoreceptor cells, by regenerating important chemicals the photoreceptor cells need to function normally. In some eye conditions sight loss is due to these underlying pigment cells dying, resulting in areas which show loss of pigmentation. In a process currently under investigation, stem cells are used to generate pigment cells in the lab, which are then injected into the eye, and appear to produce areas of pigmentation in the retina, indicating that the

cells survive. It's too early to know whether this process improves, or protects sight, but data suggests that retinal sensitivity is preserved, similar to that in the untreated eye, indicating the new cells cause no harm.

The focus now is to ensure there are no unwanted side effects from the implanted cells for example, inflammation of the eye, an aggressive reaction rejecting the cells, or the cells causing harm through the growth of tumours in the eye, or indeed spreading to other areas of the body. The next step would then be to see whether some aspects of sight can be improved, or if it is possible to preserve aspects of sight in people with residual sight remaining.

Research in the lab is now at a very exciting stage, not only can specialised cells be generated from stem cells, but also tissues, including a 3-D retina. This provides the opportunity to study the mechanism of diseases, including the causes of the loss of sight in conditions like BBS. It also helps in identifying cells, or even tissues that could be used in transplantation with the aim of improving sight.

Professor Bainbridge believes, in theory, there is good reason to expect that conditions like BBS could benefit from retinal repair by gene therapy, when performed at an early stage in the development of the condition. For those with relatively advanced sight loss, it may be possible that in the future they can benefit from transplantation of retinal cells. There are two key issues to be addressed with both gene therapy, and stem cell transplants before clinical trials can be designed: firstly, the potential benefit must be confirmed, and secondly any side-effects must be known and evaluated. Once the benefits and risks have been closely evaluated, a clinical trial can be designed. For BBS, this could be in the foreseeable future. The key is to design a trial that is affordable and fundable; the trial would need to extend over a period of years, to ensure the intervention is protecting sight long-term.

The talk concluded with the following questions:

How long could one expect the benefit of a gene therapy treatment to last? Could the therapy be performed more than once?

Animal models have shown that the benefit can be sustained long-term in certain conditions. Whether this would be true for BBS is unknown. A sustained benefit from a single treatment



would be preferable; the treatment is invasive, and therefore carries some risks.

Given gene therapy uses viruses, would it be suitable for people with compromised immune systems? The risk of introducing an infection with gene therapy is very low, as the viruses are disabled. However, in stem cell therapy, people receiving transplants are at greater risk, as they will rely on immune suppressive treatments, which could be an issue if an additional underlying condition is also present. Health is definitely an important consideration with both these therapies.

Is there an age limit for the treatment? In theory there is no age limit. The key is to identify an optimal window of opportunity. If treatment is performed too early, the risk of harm may outweigh the benefit. If it is left too late, particularly in gene therapy, there may be inadequate surviving target cells. Children would certainly be candidates for the treatment.

How is it decided who receives these treatments? The treatments described are not even at the clinical trial stage yet. Currently, 'experiments' are being performed on people to evaluate whether these treatments really work, and how to start developing treatments that could be more widely available. So far specific conditions, other than BBS, have been carefully selected. BBS did not fit the criteria, at this early stage, for a condition where the benefit of a treatment, or the potential risks could be measured easily or accurately. However, it is hoped that within the next five to ten years, BBS is likely to be one of the conditions that could be addressed by gene therapy. The team would be grateful to know of anyone who might be interested in being a candidate!

Dan the Man: a personal perspective

Daniel Evans



"My name is Daniel Evans, I'm 29, and I have BBS. My main issues with BBS are with my eyes and weight. I had a lot of sight when I was younger, I played on my PlayStation and tried to beat my brother at football. Then when I was 15 I started using a cane which helped a lot. I still had some very useful sight, until about four years ago. Now, I can just make out shadows, shapes, and people in front of me. My partner, Anna, helps me get around and I help her with things too as she has a learning difficulty, we work as a team.

I've gone through life fighting really. I went to a mainstream school and I was picked on, again because of my eyes and weight, though I still managed to get a few GCSEs. The best thing in my life was going to Queen Alexander College in Birmingham. When I went there at 16, I had no friends, no social life, and I didn't really know much about life but still to this day, I say it was probably the best five years of my life. I made friends and that's where I met Anna. At QAC, I studied IT, Admin and Customer Services, English, Maths, and I also did an English speaking course which is probably why I am able to stand here and speak to you all.

It was a residential college, so I learned how to do things for myself. When I first turned up they said Dan, What can you do? Can you clean, cook, get up in the mornings? No. So what can you do? The first term I actually learned how to make a sandwich, to cook basic things, change the bed, iron, also how to make friends, I made friends very easily when I went to QAC.

In 2007 my funding finished and I had a choice to go home or stay in the area. I chose to stay because I knew the area, had learnt routes, had friends at the college and I had a better chance of finding work. My parents helped me find a flat initially, but I now live in a house with Anna, still in Birmingham.

In my spare time I am on Facebook, and I spend a lot of my time in the gym as my weight is a big issue. When I first went to the BBS clinic in 2010, I weighed in at 26 and half stone, which had to change. I dealt with it by training hard with a personal trainer who batters me. I've managed to get my weight down to under 18 stone.

I enjoy doing half marathons and marathons. I did the London Marathon back in 2010 which was very painful but I got to the end, nine hours and 15 minutes later! Anna's done a couple with me, we got round Newcastle in 3 hours 25.

I'm a member of British Blind Sports and they found me a nice lady to train with. Last October, we did the Birmingham half marathon in 2 hours 55, which was the second time I've ever gone under three hours. I then did the Silverstone half marathon in March in 2 hours 55. I'm doing a 10k run in May, another half marathon in October and then possibly London again next year, as I turn 30.

Being a member of British Blind Sports, I run the club for Birmingham, organising the trips and so on. We have a Bowling winter league and a summer league; the winter league is a triage league and the summer league is a

pairs league, both of which me and Anna have won. The summer league is a lot harder than the winter league, especially the finals where you have to bowl eleven games. I had to bribe Anna towards the end with double vodkas, because she was starting to tire. I said okay, you're starting to tire, you can have a double vodka and coke at the end if you get a strike or a spare and do something decent after it. And she did, somehow we managed to win the whole competition.

We also go to lots of concerts; Anna likes JLS, One Direction and Union J, whereas I prefer The Saturdays or Girls Aloud. We go to theatres though I'm not keen on the theatre, but I go to support Anna. We saw Shrek the Musical, Phantom of the Opera and Jesus Christ Superstar.

I prefer sport and in July I am going to Edgbaston to watch England play Australia (Ashes). I've been a couple of times to see it when it was in England in '09 at Cardiff and Edgbaston. I've been to the football loads of times too and I've been to watch the darts at the NIA. We go to the cinema too as I don't like sitting at home, I'd rather be keeping myself active.

One thing I do find hard is finding paid employment. I work as a lunchtime receptionist at the QAC, but trying to find full-time employment is really hard, even with getting support from Action for Blind and Remploy.

Anna is a great support though and we have a lot of fun together. We're actually going away on our own together to the Sol Pelicanos in Benidorm. We went last year and as long as you go with a rep it's not a problem. If people want to go away on their own who are visually-impaired it is doable, you've just got to find the right support going abroad.

I have a couple of bits of advice. When you go travelling make sure that the train has coffee, I completely forgot that the 7.00am train we were going on did not

have coffee, so instead of Anna getting coffee at 7.00am she had to wait until lunchtime and she wasn't the happiest woman. Also as we were boarding I said to the assistance what seats are we in? Oh you can't book seats on this train! So I ended up sitting one side and Anna sat on the other side and we didn't talk the whole journey.

I think my second advice would be when you go out with the in-laws don't have the biggest dessert on the menu. As Anna would tell you, we went to the local pub with her dad, probably the first time I met him, and I had the biggest dessert going! I sat and ate it all myself, and he wasn't too impressed. So every time I go out with him now, I make sure I don't have the biggest dessert because he doesn't like sitting there watching me eat!

And that's about it really, I'm on Facebook and Twitter. If you want to follow me on Twitter, it's @tenpinkingpin."



BBS Young Person's Focus Group: Have Your Say!



We were very excited to launch the BBS Young Person's Focus Group at this year's Conference. It is so important that our youngsters have a voice within the Charity and BBS Clinics and the first meeting brought together a group of young delegates to choose a name for the group and to find out what they would like from the charity, conference and clinics in the future.

Angela Scudder organised and facilitated the event, along with Marie McGee, our Transition Co-ordinator from Birmingham Children's Hospital and Jacqueline Kearns. Three tables were covered in paper from a roll and scattered with stickers and pens. There was also a small amount of playdoh available for restless hands. The young people were able to scribble and doodle on the tables and play with the stickers and playdoh and after many suggestions, the group came up with a short list of:

- Be Heard!
- Our Life, Our Way!
- Shout Out !
- Have your say!
- It's our Life!

We took a vote and 'Have Your Say' was chosen. The group were then asked for their thoughts regarding the Charity, Conference and Clinics.

At Clinic

The group said they wanted more to do at clinic as they found all the waiting boring. They felt what was on offer now was too young and suggested:

- Art
- Nail painting
- Tee shirt painting
- Teen Mags
- Nintendo

We have forwarded the request on to the clinic teams and have already noticed an improvement

in craft activities at Gt Ormond Street Children's Clinics. Birmingham Children's Hospital has bought Ipads for their young people to use and we are waiting for approval to buy some for the Great Ormond Street Hospital clinics – Watch this space!

At the conference

The group came up with many suggestions for on and off site activities including:

- Bead making
- Trampolining
- Go-carting
- Badge making
- Treasure hunt
- Archery

Charity

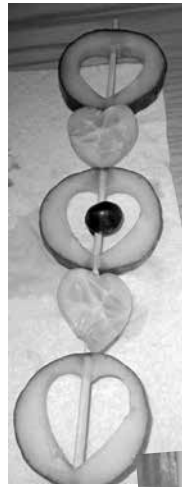
When thinking about the charity and what they would like from it, the group really wanted their own magazine, including their stories and achievements, competitions, recipes and game reviews. They also wanted to be able to go on day trips and short breaks together.

We have already had a family day at London Zoo in May, which was a fabulous day out for our families. We have another planned for Twycross Zoo in July and two activity weekends after the summer at Avon Tyrrell and October at the Calvert Trust. We look forward to seeing many of our young people there.

Looking ahead, we would like to develop a 'Have Your Say' Newsletter and to do that, we need the help of all our young members. Please send your stories, achievements, pictures, jokes, recipes, game reviews and so on, to Tonia at the address at the back of this newsletter. We can't wait to hear from you.



Weekend Round Up





Welfare Reform

Ray Perry

Area Benefits Officer
Kent County Council Social Services

Ray Perry is an Area Benefits Officer with Kent County Council Social Services Department, based in Tonbridge, and has over 30 years' experience, giving advice and training on Social Security Benefits. He is a member of the National Association of Welfare Rights Officers (NAWARA) and the London Welfare Rights Officer Group (LWROG). He has a particular interest in BBS and has already successfully

taken many BBS and registered-blind cases to appeal. Ray can provide BBS members with a benefits check to ensure that their benefits are being maximised. He is also able to offer advice on procedures and tactics if you are considering taking a case to appeal. Five years ago the coalition government announced its intention to make wide-ranging changes to the welfare system and Ray's presentation outlined these developments and their implications.



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"Ian Duncan Smith stated that the proposed reforms would incentivise employment for those able to work, limit eligibility for a range of benefits and streamline the system in a way that would achieve a sustained reduction in the levels of spending. He has been very successful in the latter two proposals, with the government targeting the Social Security budget with unprecedented cuts of £22b a year. In 2008, Ian Duncan Smith introduced Employment and Support Allowance (ESA). Income Support, Incapacity Benefit, Severe Disablement Allowance and so on, are all being reassessed under Employment and Support Allowance. The test is designed to reduce the number of people who qualify for benefit, and about a third of claimants are found fit for work with many others placed incorrectly into what's called the work-related activity group, rather than the support group. An independent review commissioned by the Department for Work and Pensions (DWP) found that the test was impersonal, mechanistic with poor decision-making and a high rate of appeals. I spend my day, sorting out the DWP's decisions.

One of the other big changes the

government made was to Contributory ESA, which is for those who have paid National Insurance contributions. It is now time-limited to one year for most claimants. This particularly affects couples, where one is in full-time work because the sick or disabled person loses their independent income. They become reliant on their partner and lose their independence.

The biggest development is the replacement of Disability Living Allowance (DLA) with Personal Independent Payments (PIP) and in the next two or three years, this is going to affect all who claim DLA. PIP has started to replace DLA with a new points-scoring assessment and it's for those of working age. For the time being, children will stay on DLA, but for anybody of working age, the aim is to reduce the number of people claiming by half a million.

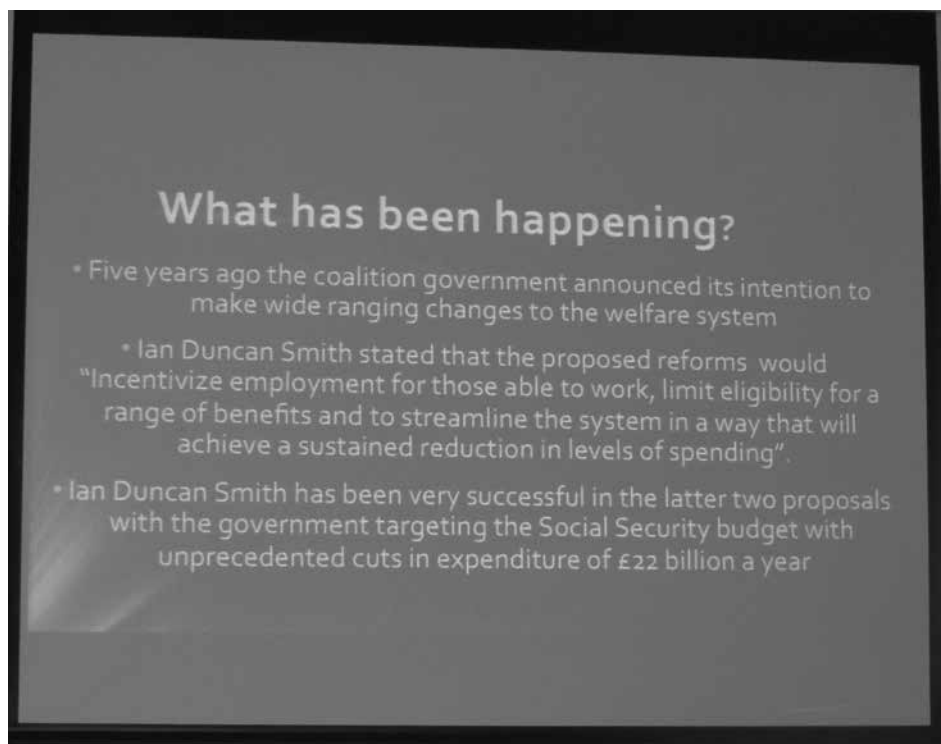
Other changes introduced include the Bedroom Tax, with Council Tax localised and reduced by 10%. The government also abolished Community Care Grants and Crisis Loans, and made various changes to the Tax Credits system, and the level and length

of sanctions. For those who have been placed in the 'work-related activity group', they have to engage with the DWP otherwise they can lose their money. This has had great effect on a lot of clients that I work with.

Universal Credit was the government's flagship reform, but unfortunately the introduction of Universal Credit has only made a limited appearance, for single jobseekers and only then in selected parts of the country. Press reports state that the introduction of Universal Credit has been plagued with IT problems, because it is a combining of the benefits system and the Tax Credits system plus Housing Benefit.

Universal Credit is eventually going to replace income-related Jobseeker's Allowance, income-related ESA, Housing Benefit, Working Tax Credits, Child Tax Credits and Income Support. It is going to combine all of them into one benefit. It has got to be applied for online and it won't be paid fortnightly, it will be paid monthly. It is going to be paid to those in or out of work. Claimants will be required to accept what is called a 'claimant's commitment', and this new commitment will be tailored to their individual needs, regularly reviewed and states what the claimant must do in return for benefit. It will make them very much aware about what happens to them if they don't meet these responsibilities and sanctions all the way through.

I wish I could be more optimistic about the future of welfare. The Conservative government have pledged to make a further £12b a year cut to the welfare budget. They haven't said yet how they're going to do this but unofficial reports think they will achieve



this by taxing disability benefits, like DLA or AA or PIP; scrapping the Industrial Injuries Compensation Scheme and contribution-based Job Seekers Allowance; restricting Carer's Allowance to those on Universal Credit, resulting in 40% of those who currently get Carer's Allowance will not be able to do so. It has also been suggested that the government will make restrictions to Child Benefit, limiting it to the first two children. These are all unofficial at the moment."

The complete transcript of Ray's presentation can be found on the Bardet-Biedl Syndrome UK YouTube Channel.

Further information, including Ray's contact number can be found on our website: www.lmbbs.org.uk

An Overview of Communication Difficulties in Children with Autism

Abigail Mance

Abigail Mance has worked as a Speech and Language Therapist for eleven years in a range of settings including mainstream and special needs schools and a specialist clinic for children who stammer. She currently works in the Wolfson Neurodisability Service at Great Ormond Street Hospital for Children NHS Foundation Trust and has an interest in the assessment and management of children with autism spectrum disorders. Abigail's presentation was about Autism and included an overview of some of the communication difficulties experienced. She explained that communication is fundamental to what Autism is and how it might impact on what we see, in terms of behaviour, and how it is relevant to Bardet-Biedl Syndrome.

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"Autism is a life-long developmental disability that affects how a person communicates with, and how they relate to other people and it affects how sense is made of the world" (National Autistic Society website).

"You may have heard lots of different terms - Autism Spectrum Disorder, Asperger's Syndrome, Asperger's and Autism. Essentially people who are known by these terms have difficulties in three key areas and this is called the Triad of Impairment and this theory was developed by Lorna Wing. The triad is that people have difficulties with: social communication, social interaction and imagination, which can include having repetitive or fixed interests.

Alongside this triad there are often sensory difficulties in that a person can be under- or over- responsive to sensory information, and that can be information coming in through all channels; sight, hearing, taste, touch. Someone may, for example be over-responsive to certain sounds like the sound of an over-head projector. This sound may be overwhelming. Or a person may find the taste and texture of some food very unpleasant, so that they may have a very restricted diet.

Someone with difficulties in the area of social communication may have difficulty using facial expressions, not readily showing whether they're happy or sad, or they might be quite exaggerated in their facial expressions, or they may have difficulty reading body language.

Quite a lot of social information is unspoken and so you will often pick up on how someone is feeling through their facial expression or what sort of posture they're holding, and it can be very difficult for someone with autism to crack the (well-hidden and unspoken) code to identify how people are feeling. They may find it difficult to take turns in conversation. Someone may talk way too much about something of interest to them, but won't ask anything about the person they're talking to. Or it may be simply that a person does not realise why it's important to use conventional greetings like 'hello' or 'goodbye'. It may be very difficult for someone with social communication difficulties to understand jokes or sayings, like 'it's raining cats and dogs', so understanding language which may have more than one meaning may be quite difficult, and a more literal interpretation of that language may be favoured. A saying like 'they cried their eyes out' might actually produce quite a horrible image for someone with a social communication difficulty, who may take that saying quite literally.

In terms of the other part of the triangle (in the Triad of Impairment), social interaction, someone with an Autism Spectrum Disorder may stand a bit too close, or they may hold eye contact in a way that is too fixed or intense and they may have difficulty picking up on how someone feels, or ask questions that aren't appropriate and so as a result, they may come across as rude or insensitive. Making friends

can be quite hard and people with Autism may spend time alone and come across as a bit of a loner or aloof.

The other aspect of the triangle is imagination and difficulties in this area may be more obvious. At around a year old, children start to understand that something can represent something else and so begin imaginative play. It may be quite difficult for someone with Autism to imagine one thing represents another thing and to evolve their play so that it becomes more complex. For some children they prefer to line things up because they just don't know how to play in a more imaginative way with the toys. People with Autism spectrum disorders tend to prefer routines and anything that changes their routine can be quite hard to manage. As an example, coping with a new or supply teacher or coping with something else unexpected may be quite difficult. They may have very strong interests in princesses or trains and the intensity of these interests can be striking. Their interest in a particular topic may dominate what they like to talk or think about. Often you see repetitive behaviour, and this may be quite simple repetitive behaviour, having a desire to flick lights on and off or it may be more complex, so they may have a little ritual that has to be done before leaving the house.

So how would this impact on what is seen? The social world can be quite a confusing place. Someone with Autism is trying to fit in, but it can be very difficult and if they don't understand people's intentions and they can't read them easily, then it can lead to frustration and sometimes withdrawal; it just becomes too much and it's easier to spend time alone. In addition, people who have difficulty communicating verbally may have difficulties asking for things and they may resort to less conventional methods, like taking their parent's arm and using it almost like a tool to get the thing that they want.

As a Speech Therapist, I'm trying to look for ways that can help, and often in Autism you do get stronger visual skills than non-verbal skills. Spoken information can be backed up with pictorial information such as hand signs, photos or symbols. This can aid understanding of spoken information and expression. The Picture Exchange Communication Systems (PECS) is a relatively well known approach in the field of autism, where symbols are used

to help children communicate messages like 'I want a banana'. Visual schedules convey pictorially how to complete a simple task. They also have the advantage of encouraging independence - a child can go and have a look at a schedule and see by themselves what to do. Visual timetables convey to a child what they will be doing during the day / week. They can be used to prepare a child for a change in their day-to-day routine. They can be used at home or school.

Adaptations of visual support strategies can be made for children with Autism who have sight impairment. For example, tactile cues could be used instead of picture symbols.

Aside from communication, there are other factors that impact on behaviour:

Limited flexibility: I've talked about how children with Autism often have a preference for routines and knowing what they're doing and disliking change and that change can really upset. It could be a change in something that a child may hold in their mind as being quite fixed such as a change in the design of a favourite cereal packet which could be quite upsetting and interfere with daily life.

Social situations: can generally be quite anxiety-provoking and I think it is often down to the unspoken code that lots of people intuitively know how to interact, but it can be difficult for someone with social communication difficulties to pick up on.

Sensory difficulties: I've talked about sensory difficulties as well, which can make it very uncomfortable for someone with Autism to go out, particularly into busy environments with lots of noise and light.

The Diagnostic and Statistical Manual of Mental Disorders V (DSM V), published by the American Psychiatric Association, offers a standard criteria for the classification of mental disorders. It attempts to classify the severity of Autism Spectrum Disorders on the basis of social communication impairment and restricted and repetitive behaviours on a scale from level 1, requiring support, through to level 3, requiring very substantial support. So far, I am not using this scale clinically. People with Autism Spectrum Disorders are very individual and a general descriptive scale like this doesn't effectively capture what is going

on, or consider the environment of the child. You could have someone in an environment that is very well-suited to their needs and so they don't show as many difficulties, versus an environment which has not adapted to their needs and so the difficulties are more evident. It is quite difficult to say someone has mild, moderate or severe Autism. It is quite a varied condition and it is often a hidden condition.

Autism affects one in 100 people. It affects more men than women and it affects people from all nationalities, cultures, religions and social backgrounds.

It is worth saying that a proportion of people with Autism Spectrum Disorders will have learning disabilities, however not all, and the degree of learning disability can vary. The focus of my talk is on communication so it is worth pointing out that a learning disability can inevitably have an impact on communication. For example, it will have an effect on what somebody understands, how they understand and how well they process instructions, explanations that are given and stories they hear. It will also impact on how they can express themselves, the kind of vocabulary they can use, the words they can use and how they learn vocabulary. So you can have an Autism Spectrum Disorder and learning disability and together they will both have an impact on communication.

I reviewed some of the literature out there on Bardet-Biedl Syndrome and Autism. Barnet et

al's article in 2002 used something called the Childhood Autism Rating Scale, and 4 out of 12 parents indicated a significant level of autistic-like symptoms on that scale. The Childhood Routines Index showed increased level of routines and rituals, compared to typically-developing children. Forsyth and Beale's article in 2013, in their clinical overview reported that children with Bardet-Biedl Syndrome often are reported to have labile behaviour, which can lead to outbursts of frustration, and many prefer fixed routines and have elements of obsessive compulsive disorder, and lack social dominance. Others develop Autism spectrum disorders, and psychosis. Brinkman et al 2013, in their study reported that most participants (80%) were reported to have social skills deficits but there was no indication of severe Autism Spectrum Disorder.

To conclude, children with Autism experience difficulties in key areas – social communication, social interaction, imagination and sensory difficulties. It is probably best thought of as a *spectrum* disorder, with people being affected in different ways but having difficulties in all those core areas.

Before I finish, I would just like to recommend, if you are interested in finding out more, the National Autistic Society which has a wealth of advice on what Autism is and how to get assessments, a diagnosis, if appropriate, and strategies to cope. For more information go to: www.autism.org.uk"

Running Up That Hill

Tonia Hymers

Daniel came into our world on the 11th July, 1996. The most beautiful little slip of a thing with shocking red hair. My husband has black hair and I have brown, so of course I had to put up with endless jokes about the mythical ginger postman! After a rollercoaster pregnancy, I was relieved to finally hold my little man.... to my confusion, Dan had one extra little finger, perfectly formed and although we didn't know it at the time, it was the start of his and our BBS journey.

Dan's failing kidneys were picked up at around

five to six months and many tests followed at Great Ormond Street Hospital, with Dan being diagnosed

at around ten months old. There was no explanation of what BBS was and in those days, no internet. In the local library I found a book explaining the symptoms of BBS, in out of date and brutal terms. Time stood still a bit. We knew about the kidneys, extra finger and weight issue, however the potential learning



difficulties and visual impairment was a shock. It was our genetics counsellor who told us a bit more about the syndrome and the LMBB Society and this was when we first made contact with Drina Parker.

After the wait to get pregnant and the roller coaster of pregnancy and childbirth, when things are not as they should be, it is important to allow time to grieve for the life we thought we would have and accept the one we are living. Raising a child battling illness and disability is an incredibly intense experience, especially when there is delayed development; every moment and milestone is so precious. As many BBS parents have experienced, having a child with disabilities tends to sort your friendship group out for you. It is hard to raise a baby with developmental delay, obesity, kidney failure and visual impairment in close contact with other, apparently healthy babies. There is too much comparison and too many competitive and sometimes unkind or thoughtless parents. I withdrew for a short time and my parents cocooned us and provided the love and support needed to get through the difficult early days of diagnosis and acceptance. They are our greatest cheerleaders and we simply could not function without them. Over the years, we got a grip on all things BBS, accepted our lot and woke up to the fact that very few families and people in life escape unscathed.

By the time Dan reached school age, I realised that it wasn't about me... there is nothing like the pain of watching your child struggle and suffer... but ultimately, we can leave the BBS mantle at home and go out socially and recharge. Our children have to live with BBS for the rest of their lives. Our role is to be their parent, their friend, their advocate and *their* cheerleader and help them make their lives as good as they could possibly be. There is no one else in the world who knows our child like we do, and we have to take this on board and make ourselves heard. Maintaining good relationships with the key people in our children's lives is vital, but we have to *believe* that we are an equal partner with medical,



social and educational teams, whether they do or not. We have to use the resources available to us, to give our children the confidence, courage and self-esteem to know that they have a great deal to contribute to this world.

Sometimes this is easier said than done and there are just too many hills. I was at the House of Commons recently for the Rare Disease Reception and an MP spoke very passionately about the delegates in the room, who were all either parents or rare disease patients. She praised and thanked us all and asked us to keep pushing our MPs, to keep rallying and protesting and demanding to be heard. This made me quite cross. Isn't it enough that we have to fight at a local level for our children to have access to decent health, education and social care, while at the same time cope with the demands of looking after our families, in between hospital stays and appointments? And of course, we are not immune to the life events that affect everyone else, in fact, families like ours are more susceptible to illness, relationship break-downs and financial hardship. I think one more glass of wine and I may well have started a riot!

As a family, we had three major worries when Dan was little, his weight, his eyes and his kidneys. Between four to five months, Dan ballooned from normal weight to clinically obese and like other BBS mums I spent the next X number of months and years explaining

to all and sundry that I really wasn't over feeding him! He was an enormous baby, however we were strict, maintaining portion control and limiting treats to special occasions and although he was always chubby with a BBS tummy, we managed to get his weight under control. At around 15, Dan finally started shooting up and the excess pounds melted away. Dan has grown up with a conscientious attitude towards food and now manages his diet beautifully.

When Dan was three, his little brother, Connor, came along. Con was a gorgeous little boy, a ray of sunshine and he and Dan were good friends. It was hard on him though, as it is for all siblings of children with illness or disability; there was no child on this earth more loved, however he perhaps inevitably felt that Dan was more important and his self-esteem suffered. We worked hard at getting things back on track and although it took a while, we got there. Connor has grown up into a funny, loving, typically moody teenager, who loves his brother and is passionate about disability rights and the LMBBS.

Over the years, Dan became increasingly unwell and in early 2006, a viral infection put him on the path to transplant. My kidney was transplanted into Dan at Great Ormond Street Hospital. Daniel was only ten years old and was incredibly brave. He had been needle phobic his whole life and during this period, endured so many needles, canulas and tubes with absolute courage. By day two, Dan was off morphine and on paracetamol using the Playstation as a distraction. Meanwhile across at the Royal Free, I was hyperventilating because the morphine drip wasn't working fast enough!

I missed Dan and Con so much but thankfully once Dan was discharged to the GOSH patient flat, we were able to be a family again. It was a tough few months for all of us, however we muddled through and came out stronger, individually and as a family. Dan's kidney continues to do well.

Once we had all settled down after the transplant, the next worry reared its head, Dan's vision. Dan was diagnosed with retinal

dystrophy at around a year old and we settled into the routine of yearly check-ups. Not long after the transplant, at an ophthalmology appointment at our local hospital, the consultant was quite frank in front of Dan. Although Dan knew about BBS and his vision, it was a shock for him to hear it like that. The transplant had taken his mind off the long-term implications and he suddenly had to face it again. This was one of our more painful moments, seeing that realisation hit home. Running up that Hill is a song about swapping places – there have been so many times when myself and other parents like me have wished that we could do just that. We made our way home, all lost in our thoughts, when out of the silence, Dan pipes up 'Horse walks into a bar, barman says...Why the long face!' We were laughing and crying – a touch of hysteria I think.

You could be forgiven for thinking Dan was a joker and happy go lucky little boy but that couldn't be further from the truth. Like many with BBS, Dan struggles hugely with his emotions. When he was little, emotional outbursts were a regular occurrence – the terrible twos (and threes and fours) for a BBS child, (and parent) are truly terrible! This phase passed, however the emotional difficulties remain, which makes it all the more remarkable how well he has coped with the various challenges over the years. I didn't know whether to laugh or cry when I first saw him hook his finger over the cup to judge when it was full, it showed that his sight had deteriorated, but also showed a natural resourcefulness that would be invaluable.

When Dan was little I found the uncertainty difficult; we knew he would become visually impaired but we didn't know when, we knew he would need a transplant but we didn't know when. I wanted to know it would all be ok in the end. It was painful and difficult at times, but we got through it... You can rely on the clock to keep ticking and eventually it is over. The treats helped too! We had great fun with our caravan, we visited Legoland many times and Disneyland Paris thanks to Dreams Come True. We also threw caution to the wind and re-mortgaged to take the boys to Disneyworld Florida and to Egypt so Dan could fulfil his

wish to see the tombs and pyramids.

So where are we now? Dan has endured much in his 18 years. There have been difficult, ignorant teachers, bullies, endless medical tests and interventions. He has had to cope with understanding the condition he was born with and the implications it holds for him and his future. But during this time, Dan has kept his identity. He isn't defined by his weight, his vision or by BBS. He is a bright, funny young man who grew up on Disney, Dr Who, Star Wars, Lord of the Rings, Lego and computer games. He is developing an interest in politics, he loves Ancient History, Miranda, QI and comedians in general and he does not suffer fools gladly. Despite the many challenges, Dan has kept an eye on his future and has developed the drive to take him there. A failed sixth form placement gave him the determination to try boarding at the Royal National College for the Blind in Hereford where he is studying IT, Braille and Business. He is going to university in September to continue studying IT and we couldn't be more proud of him.

Dan is not alone though. I have met many children, young people and adults who have BBS at conference and clinics over the years and their bravery, courage and determination in the face of disability and illness is inspiring. I have also seen so much achievement within our group, whatever the passion. We have successes in business, sport, in the workplace,

(paid and voluntary), music, art as well as academic, there is a great deal that can be achieved. As a family, we were fortunate to learn this, early on at our first conference when Dan was just two years old, which gave us so much hope for the future. We met Steve Burge and shortly after, Graham Longly and Richard Zimbler and over the years more and more of you have shown us all what a great future our children can have.

We have come a long way since then and owe a debt of gratitude to so many. Thank you.



Adults Reception

Richard Zimbler and Steve Burge once again hosted a reception on the Friday night of the Conference for adults living with BBS. They wanted to try and add some structure to the event and decided to introduce table hopping, which would enable everyone to meet as many people as possible. There were four tables covered in paper, with five questions across the four tables, to get everyone talking and sharing ideas. After twenty minutes or so, everyone moved to a different table and discussed that table's question and comments. The questions

are listed below, along with a selection of some of the suggestions that were put forward.

What would you like from the BBS Clinics in the Future?

- Better communication and administration
- Food selection could be better
- Medical staff covering other aspects of the syndrome, for example joints, feet etc
- Results and referrals are not being picked up by GPs

- Results should be in layman's terms and not GP language.
- Clinics should be all day with the follow-up clinic a half day

What are the major struggles of living with BBS

- Access to information
- Diet
- Exercise
- Doctors don't understand
- Accessing public transport
- Isolation
- Complete lack of energy

What would you like to see from an adult support worker?

- Emotional support
- Help with form filling
- Help with benefits
- Organise regional meetings
- Help with access to sports
- Support with dealing with diagnosis
- Support with employment
- Meet up more often

What would you like from the charity in future from an adult's perspective?

- Regional meetings

- Bring adults who are computer literate together with those who aren't
- Coffee mornings for fundraising – bake sales etc
- Regular e:newsletters
- Running technology based events – learning IT for example
- Theatre Trips

Fundraising ideas

- Sponsored Swims
- Domino Bingo to raise money
- Karaoke night
- Auction of some kind
- Quiz supper evening
- Guide Dog Sponsored Walk
- Tandem cycling

There was a lot of chat and laughter coming from all the tables and although some felt it was a little formal, it was a great success with the majority of participants. The comments and suggestions have been fed back to the clinics teams and committee and will be used for future planning. If you have an idea for next year's reception, contact Steve Burge; his details can be found on the back page.



Delegates' Comments

What did our delegates say about Conference 2015?



"Kids had a fantastic time and carers superb as always."

"Speakers were all very good and informative. Found autism talk specifically very good..."

"Great as always, committee works hard, but sad that Chris and Phil are leaving committee – they are very special."



"Good to hear about research developments and especially the personal perspectives."

"Childcare facilities are faultless – excellent carers in the crèche."



"The team are worth their weight in gold, they do a marvellous job every year. Many, many thanks."

"I really enjoyed making mosaics, it was fun, calm and peaceful. I enjoyed the crafts."



"Staff are brilliant, always smiling and very helpful."

"Thank you for all the continued work that has gone into organising a fabulous conference."

"Myself and Shaun would like to thank Sandra Dale and the care team for looking after Savannah at the conference on Saturday. She really enjoyed herself. She is looking forward to seeing you all next year, and she is so excited about it. Lots of love from Savannah."



The BBS UK Annual Family Conference is a fantastic weekend full of learning, networking and social opportunities. A booking form for Conference 2016 is enclosed with this report, so don't delay, complete the booking process as soon as possible to guarantee your place on this invaluable weekend; we look forward to seeing you there.

Goodbye Chris and Phil

A Tribute from Julie Sales and Tonia Hymers

At the end of Conference 2015, we said a heartfelt thank you to two very important people within the LMBBS family, Chris and Phil Humphreys. Chris and Phil have been a huge part of the Society for 21 years and were there in the early beginnings with Drina and Michael Parker. They have seen the Society grow and go through many changes through the years and during this time have been an amazing source of support, not only for their own family but for countless LMBBS families, including all of us on the Committee.

Both have become a special part of our lives and, in the words of the founding member, Drina Parker, 'we thank you for your amazing 21 years of devotion to LMBBS and for being a very supportive friend'. Drina continued, 'I'm sorry I can't be with you today, Chris, to say these words in person and to let you know how highly I value all that you have done for LMBBS. The saying 'if you want a job done, ask a busy person' is so true of you. No matter how many tasks you have to perform you never refuse a request for help and you always do it with a smile. No wonder you are held in such affection by all members of LMBBS, not just me.'

Tonia and I have known Chris since we joined the Committee 18 years ago and have worked very closely with both Chris and Phil over the years, with conferences,

m e e t i n g s ,
fundraising and the
BBS clinics. Chris'
work ethic, passion
and dedication will
never be matched.
She is an inspiration
and will be sorely
missed. We know
Chris will continue
providing support
to us all over the
coming months and
years, her experience
is invaluable.

Chris and Phil should both be incredibly proud of the legacy they have helped to build. A thriving charity with a strong, new Committee, exciting events in the pipeline and clinics going from strength to strength. This has all been achieved thanks to their endless energy and enthusiasm. We all wish them both a restful retirement. Chris and Phil, know that you are highly thought of and thank you from the bottom of our hearts.

On behalf of the Society, Julie and Tonia presented Chris and Phil with a crystal vase and a photo album full of conference photos from the past 20 years.

Farewell from Chris Humphreys

21 years ago last month, the Humphreys family attended their first pre-diagnosis day conference at Exhall Grange in Coventry. James was 12 years old. It was a day of initial euphoria at recognising the features of LMBBS; we were welcomed with open arms and those friendships have grown over the years. Suddenly we were no longer alone dealing with the unknown. These words will sound familiar to many, if not all of you attending today. We are all part of a large extended family. We soon became members of the LMBBS Committee, roles we have cherished over the years. As National Co-



ordinator I became the first point of contact for the Society, and in particular newly-diagnosed parents. This has been a particularly poignant role for me as I well remember the evening hours spent talking to the then National Co-ordinator, Drina Parker. I recently felt that I had succeeded with this role with a comment sent in by a member on hearing of my impending retirement, I hope she won't mind me sharing it with you. 'I still treasure the letter you sent me when my son was first diagnosed and I was in a very bad place. Touching things that you have done like that will stay with people forever and please know that you will always have a special, grateful place in my heart.'

We have been overwhelmed by the emails received since our retirement was announced and thank you for your kind comments. We have so many special memories to look back upon since that first day, so many friends, both personal and professional, to thank for your friendship and support. You all know who you are. And so it is with sadness that we are leaving today, but the time is right. You have a new and vibrant Committee who will take the Society from strength to strength. We will always have some involvement with the Society and attend future conferences, James would not allow us not to. He has grown up as part of the LMBBS family but for now, and again I quote from an email received, 'it is not the end of an era but the beginning of a new chapter'.

Farewell from Phil Humphreys

What I'm going to say comes from the heart, it can't come from anywhere else.

Our James is adopted; we fostered him when he was just a few months old, when he was a bouncing baby boy with a mop of curly blond hair. When he was about two years of age, all of a sudden one day, he had a hell of a temperature. From that time on, he changed as a baby and we just didn't know what was the matter with him. He had an extra digit on his foot, which was removed. We went from

place to place, doctors, professionals asking what is it, what's wrong with him? It was due to Chris's tenacity, when James was 11 years of age that we found out that it was LMBBS. The doctors said to Chris 'well, now you've got your label'. It wasn't a label we were looking for, but as all mothers will know, she knew there was something wrong with her child, something different, and eventually we found out what. I couldn't be prouder as a dad of our James. He's done so much for the Society with his sponsored parachuting and other activities.

We became involved with the Society, our first one being in Exhall Grange in Coventry, 21 years ago, where the first people we met at the conference, like us, were from Wales, and that was the Begleys; we've been friends with them ever since. We met Drina and Michael Parker, who founded the Society. If it hadn't been for those two this society wouldn't be here today; the work they've put in over the years has been fantastic. Michael was Chairman of the Society and I've got to be honest I was in awe of him. I never thought that ten years after, I would take his place as Chairman. It's been a pleasure, a real pleasure, and not only that, it's been an honour.

Chris and Phil presented Julie, Kevin and Tonia with a personal thank you gift and flowers from the Society for their help in the organisation of the Conference.



Friends of BBS UK

The financial stability of our small charity relies on regular donations and we are often asked why we don't charge for membership. This would indeed provide that much needed regular income, however it has always been our policy that membership is entirely free.

Several years ago, we set up 'Friends' of the LMBBS, a fundraising initiative, giving those members who wished to regularly support the Charity, the means to do so and we have slowly built up a small, loyal band of 'Friends'.

If you would like to support BBS UK in this way, contact Kevin Sales for more information: kevin.sales1@btinternet.com.

BBS UK Needs You!

Over the past three years, BBS UK membership has grown hugely, largely due to the success of the specialist clinics. With a growing and diverse membership spread across all of England, Scotland, Wales and Ireland, not to mention Europe and the rest of the world, it is hard for one committee to look after the needs of everyone as well as to keep driving the charity forward to bigger and better things.

If you feel you have skills that could benefit BBS UK, please do not hesitate in coming forward. In particular we are looking for those with charity, management and human resources experience. For more information or to express an interest, please email: toniahymers@btinternet.com.



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