# Conference Report
## 2014

Laurence-Moon-Bardet-Biedl Society

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# Foreword

After every conference we ask, 'can we improve on this success?' The answer is, of course, 'Yes' and 2014 was no exception. We had one of the highest attendances to date, with something of interest for everyone. On Friday evening we had 'Resource Central,' an opportunity to drop in and chat to the professionals and facilitators in one large room. Steve Burge and Richard Zimbler hosted a reception for adult delegates who have BBS and Claire Anstee and Laura Dowswell once again hosted the New Families Meeting, bringing all first time delegates together, to chat informally and share experiences, which is always so appreciated.

Saturday morning saw the little ones happily playing in the crèche while the older children and young adults went off to the Theme Park or Ten Pin Bowling with their brilliant carers; it was time to start on the serious business of the conference. The LMBBS Annual General Meeting saw Claire Eccles welcomed on to the Committee. Claire, mum to two young boys with the syndrome, has been actively working behind the scenes at Sight Village, Birmingham and with friends and family, fundraising for the Society. We know Claire will be an asset to the Committee.

The AGM was followed by an excellent mix of Speakers and Personal Perspectives. Dr Helen-May-Simera flew in from Washington, USA, to speak about her research and to tell us her exciting news; Helen has been chosen to receive the 'Sofja Kovalevskaja-Award, which will enable her to start her own research group in Germany, looking specifically at cilia in the eye and possible treatment options from late 2014. There were also presentations covering research, emotional well-being and speech and language issues, in addition to moving perspectives from Stefan Crocker and Rebecca Goodman. You will be able to read edited versions of all the presentations within this report.

The morning culminated in a splendid lunch, followed by an afternoon of small informal workshops. In no time at all, we were once again greeting our tired and weary children, adults and carers, returning from their fun filled day out. We are so grateful to our incredible care team, who return year after year, and without whom, parents would be unable to fully enjoy the conference, gleaning much needed information to assist them in their day to day coping with the syndrome.

The resource, merchandise, tombola and raffle tables were all well supported. Hollie Sales made a selection of mosaic craft items and Sandra Dale and friends made a range of celebration cards, all to raise funds for the Society. Our delightful up and coming young fundraisers, Emmy and Holly Anstee also sold handmade items on their own fundraising table. Our grateful thanks go to everyone who donated or made items, took time out over the weekend to oversee the stalls or supported them, you are all stars!

Following an early tea, the Care Team supervised craft and games activities for older children, including a very popular mosaic craft table; many thanks to Chrissie for supplying the materials and helping throughout the weekend. A highlight of the evening was the hugely enjoyable music jamming session. Richard Zimbler and Connor Hymers played a variety of songs on the drums and guitar respectively, supported by many children and their parents on various percussion instruments. The room was a sea of happy faces and some of the children danced to their heart's content. As always, the evening finished with a big family quiz and raffle. The LMBB Society and Family Conference Weekend continue to go from strength to strength, which is only possible with your support and that of our amazing team of professionals, carers, volunteers and fundraisers.

This very special weekend is so valued and continues to grow year on year. We would love to see lots of new faces next year, so why not make it your New Year's Resolution to attend. Booking information and forms are included with this Report, so don't delay and miss out. We look forward to seeing you next year.

***Chris Humphreys***

# Update on Research and Study of BBS

**Professor Philip Beales**

"As most of you will know, the specialist BBS Clinics are held in two centres in Birmingham and two centres in London, providing a multi-disciplinary service to all of the BBS patients in the country. We have over 450 patients now attending this clinic, which I think is a phenomenal success. The clinic is funded by NHS England, which means we are funded directly by the Department of Health.

Coming up to our five year renewal, with all of the changes that have been happening in government and in the NHS, we were, to say the least, a little bit worried about the fact that these highly-specialised services, and there are 70 of them in the country, could have been at risk. We have been told very loudly however, that we are safe and we do not need to apply for renewal of this particular service. We do, however, get judged every year on the quality of the service that we are providing.

For those of you who are not familiar with the clinics themselves, we run a one-stop clinic, allowing patients to see six or seven different clinical specialists all in the one day, rather than have to attend individual appointments. The clinics are supported in a very unique way by the LMBBS Society; Tonia and Julie and many other people help run these clinics and without their support, we really could not cope. The involvement of the LMBBS really has added an extra dimension, so much so, that other services around the country are looking towards this one as a model of how clinical care should be delivered. We are ably supported by Kath Sparks, clinical nurse specialist, in London and her equivalents in Birmingham, and behind the scenes, there are administrators, secretaries, managers and clinical scientists.

In July of 2013, we held our annual audit day. This is a requirement of the clinical service, however it is also a really useful opportunity for us to review how the service is running and helps us to determine where problems are arising. We have to be able to improve year on year; commissioners from the Department of Health attend the meeting and listen to the presentations from the various specialists who run the clinics. This year our main focus was on ophthalmology and clinical psychology. In addition, Dr Elizabeth Forsythe and Kath Sparks presented a report of a clinical survey they had undertaken. We also heard from the dietetics team; Sarah Flack and Waseema Azam have been working very hard with our young patients and have had a lot of success. At least 61% of the children attending Great Ormond Street clinics have been able to reduce their weight by working with the dietitian. What we need to know now, is how well this is sustained into the adult clinics.

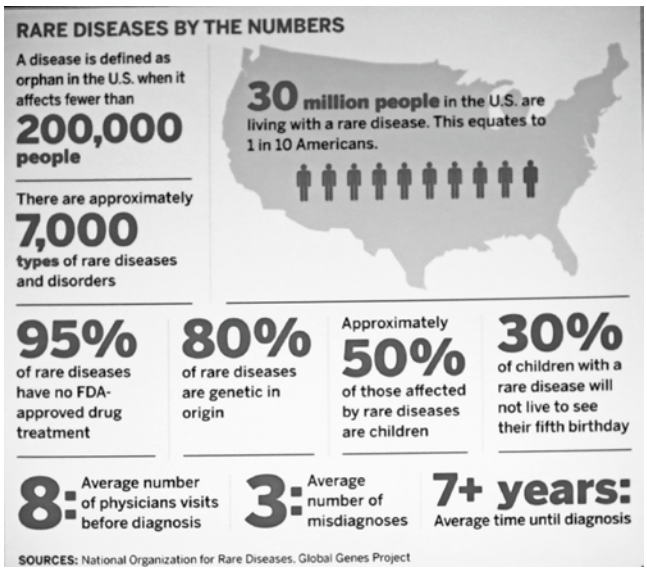
For many years I have spoken at conference about research and the big issue of funding. We depend on grant funding to conduct research, to understand why BBS occurs and, in particular, how we might be able to treat it in the years to come. There has been a huge seed change in thinking in the last 18 months and essentially, a move away from the National Health Service's preoccupation with common diseases. The NHS is designed for looking after patients with common diseases, rather than people with rare diseases, however, we know that there are a huge number of rare disease sufferers around the world, approximately 350 million people. Governments, by and large, have been ignoring rare diseases, at least up until the last couple of years; politicians really care about cost and they have now realised that the number of people with rare diseases are actually costing the Health Service quite a lot.

In the EU, a rare disease is a condition that affects less than 1 in 2,000 people. LMBBS is one rare disease out of around 7,000 that have been documented and 80% of these are

genetically determined. What is also very important is that approximately 65% of rare diseases affect children and 30% of those children will not live to see their fifth birthday. For 95% of these rare diseases there are no treatments, no FDA-approved drugs, and now, the politicians have woken up and are realising that we should be doing something about that.


We are now realising the significant impact of rare diseases. There are 3.5 million people with a rare disease in the UK, and on average it takes 8 physician visits and more than 5 years before a diagnosis is made. In addition to improving the patient's pathway to diagnosis, we need to know what the cost is to patients with rare diseases. One estimate is that 31% of people with a rare disease have had to use their savings in order to be able to just cope on a daily basis with having a rare disease in the family. Then there is the emotional toil, the anxiety and isolation and so on that comes with having a rare disease, and these are impacts that we need to be measuring and costing up at a national level. The EU has really been instrumental in driving forward the recognition that rare diseases are very important and, in 2009, the EU Council decided that all of the member nation states had to come up with a national plan for rare diseases; we delivered the UK plan for rare diseases in October 2013.

The first thing the UK plan suggests is that for every patient, a clear personal care plan should be developed, to bring together health and care services to provide more support for rare disease patients and their families; I hope we are already part of the way there for BBS patients, however for most other conditions this level of care and support isn't there yet. We need to develop specialised clinical centres to offer the best care and support and again, we have had our specialist clinics since 2010, but this needs to be recognised and rolled out for other rare diseases. Better education and training for health and social care professionals has also been flagged up and better access to treatment as well.




There are 51 recommendations in total, but what is really important is that all four countries of the UK have committed to this plan. Alistair Kent, Chairman of Rare Disease UK and Director of Genetic Alliance UK is chairing a committee which I have been asked to join, to oversee the implementation of the plan throughout the four different countries that make up the United Kingdom. It is going to be a huge challenge because they all do things differently; prescriptions in Scotland are free and elderly care in Scotland is free, whereas in England it is not.

Moving away from politics and onto talking a little bit about the genetics of rare diseases, of the 7,000 rare diseases, the genes causing the conditions have been found for about 50% of them. We expect that by 2020, the remaining 50% should have been discovered and this has been completely driven by technological scientific advances in the field, mainly through the development of new machines that can sequence all DNA in one single experiment. For LMBBS, 80% of the genes involved, if not more,

have now been discovered.

David Cameron stood on the steps of Number 10 over a year ago and said that we are in a pretty good place with the NHS; we have a joined up healthcare service and we have some of the leading genomics or genetic scientists in the world and should therefore be the first country to be delivering 'personalised medicine'. My prediction is that you will be going to your GP within the next ten to fifteen years and your GP will be able to look at your genetic sequence on the computer and the computer will suggest a particular drug based on your genetic sequence. That is where we are heading in the health service and internationally, and is how medicine is going to be delivered in the future.

So a year ago, David Cameron announced the 100,000 Genomes Project which involves the sequencing of the whole genome and they have decided to do this in 100,000 people who have cancer, rare diseases or infectious diseases. Many other countries have decided that they want to do this too; Abu Dhabi, Saudi Arabia, Qatar, and the US, all now have their own 100,000 Genomes Project. In London, we have had significant involvement with this particular programme and I'm really pleased to say that LMBBS, again with the Society at the forefront and through our clinics, is contributing to this very cutting edge international programme.

Last year I talked about how we need to engage pharmaceutical companies more and I can safely say that we have now engaged two of the big pharma companies. We are about to sign a deal with Pfizer, which will allow them to assist us in our research and also give us access to their expertise. Some of our research is aimed at finding medications or drugs that might at least slow the progression of visual impairments or kidney problems in LMBBS. We can only get so far however, because we are not chemists, we don't have that kind of expertise. Once we find the chemicals or the drugs that might help, we can ask the big pharmaceutical companies to give us access to their chemists who can make that particular drug maybe safer or more effective.

Beth Hoskins has been working really hard to develop genetic testing as part of the BBS clinical service. There are now over 18, maybe 20, different genes implicated in BBS and, of the 376 patients that we have now tested within the clinical service, we have a confirmed diagnosis for 80% of them. We have also been able to confirm carrier status in 80 parents and 21 other siblings or relatives, so it isn't just about developing a test to confirm a diagnosis in an individual, although that is very important. We have also carried out a number of pre-natal tests to check whether babies in the womb are going to be affected or not.

We continue to do a lot of basic science; Dr Sonia Christou-Savina, Senior Research Associate, UCL, has been doing some ground-breaking work, looking at why there are learning difficulties, memory issues and so on in BBS. Sonia has found that some of the faults may lie in the synapses, the connections in the brain, and is looking at whether simple measures such as exercise might improve learning. This is something that has already been discovered and published many times in the general population. Over the past year, funding for basic science has dried up in favour of funding for translational research. We are moving into translation work and use zebra fish in the lab to understand the biology of BBS and to test drugs that are already approved and in use. Dr Dan Osborne and Dr Miriam Schmitz have been instrumental in pushing forward screens to find drugs that might help in terms of delaying the onset of visual impairment, renal abnormalities and so on. Dr Victor Hernandez, Research Associate, ICH, has been researching weight gain in BBS and also gene therapy, which is starting to mature very nicely in the lab as well.

An indication of how much research is being done is the number of papers being published and BBS publications have been rising significantly over the past few years. In the year 2000, the year we discovered the first BBS gene, there were only around 17 papers published about Bardet-Biedl Syndrome or anything connected with Bardet-Biedl Syndrome. Since then, there has been a steady rise and by 2007, there were about 50 papers a year being published. We reached a peak in 2011 and there were just under 70 papers published and then, in 2012-2013, the figure sat at around 60-70 papers. By April 2014, we are already up to 40 papers, so I imagine by the end of the year, we should see around 80 papers published on BBS.

So if we wind the clock back ten years, no-one knew anything about this condition, but now, there should be no discerning biologists or doctors who have not heard of BBS, and this really is very much down to the work of this group, this Society, in getting out there and making the condition known. I am part of this effort as well and am somewhat shameful in publicising where we can,

not only our work, but also where possible, the LMBB Society; we had a nice piece published earlier this year in the New Scientist, one of the very high brow research journals and together with some LMBBS members, I contributed to a Horizon programme, 'Big Data' on BBC2 last year. Although much was cut to suit their own programme, we still got a little bit of airplay. I also did a programme recently with BBC Radio 4 and, again, I promoted our work on that.

So back to publications, BBS publications which mention therapy have been few and far between. There were no publications before 2008, when just two were published, followed by just one paper in 2009/10/11. In 2012, there were a phenomenal three papers, including one of ours on gene therapy. I expect, over the next few years, we are going to see much more in this area, which reflects the funding and also reflects the emphasis of my lab, because although we are still interested in doing the basic science, we aim to capitalise on the funding that is available for translational or therapeutic research. We are focusing on redefining medicines or compounds that are already out there, that we might be able to re-use or repurpose for our benefit. We are also doing a lot of work around gene therapy."

**Q: You talked about gene therapy, gene engineering. I remember, when we were first organising these conferences, we were always grasping for that straw… I just want to make sure that people appreciate that the therapies will be helpful to future generations, but are unlikely to turn the clock back…**

A: I absolutely agree. We have to be cautious about raising too much expectation around the therapies and so forth because we are perhaps talking about the next generation who may benefit the most and I think that's a given.

# How Cilia Can Help Us See

**Dr Helen May-Simera**

*Dr Helen May-Simera completed her PhD in Professor Phil Beale's laboratory at the Institute of Child Health before moving to the States in 2008, to continue her research. Helen has recently been chosen to receive the prestigious Sofja Kovalevskaja Award to start her own research group in Germany, looking at cilia in the eye and possible treatment options. Helen's talk was about her research on cilia (singular cilium), in particular cilia in our eyes.*

"A cilium is like a little hair sticking out from a cell, and is thought to be like a signalling antenna. It helps cells communicate and function. Lots of different cells in the body have cilia on them. BBS is caused by cilia not working properly. There are two types of cilia. The one people are most familiar with are the *motile cilia*. There can be many of these type of cilia on a cell's surface. They move, bend and wave, and move fluid across surfaces. They can be found in the brain, lungs, fallopian tubes, and in our ears helping us to hear. The second type of cilia, are *primary (or sensory) cilia*. Cells that have a primary cilium have just one very specialised cilium. Until recently, scientists did not think this type of cilium had any function; only recently have they realised how many functions they have, and how important they are. It transpires that some cells in the eye have one of these highly specialised primary cilia.

As previous talks have mentioned, there has been a huge push to identify and understand which genes are causing BBS. Genes contain all the instructions a cell needs to make proteins. Proteins are the building blocks of every cell in the body and are needed for our cells to work properly. A cilium is made up of lots of proteins

stuck together. If a gene has a mutation, some of the proteins will not be made correctly, and the cilium may not work as effectively as it should. In most patients with a ciliopathy, the cilia exist but do not function optimally because the proteins involved in making the cilium function are disrupted in some way.

Of all ciliopathies, BBS is one of the most studied, as it exhibits so many of the symptoms common to ciliopathies, for example, retinopathy, polydactyly, obesity and cystic kidney disease. Almost all the different ciliopathies include some form of visual impairment, the reason for this is explained below.

The eye captures information that we see, processes it, and sends it on to the brain. A key part of this process is the retina, sitting at the back of the eye. The retina is a thin, transparent layer, a few millimetres thick, containing many different layers of cells. Right at the back of the retina exists a highly specialised type of cell called a photoreceptor, each of which has one *primary cilium*. These cells capture light, and transform it into an electrical signal that is transmitted to the brain. If you isolate just one photoreceptor, it does not appear, at first, to have a typical hair-like cilium. Under a microscope however, the photoreceptor looks like a long rolling pin, and it is the arm of the rolling pin that is, in fact, a cilium.

There are two different types of photoreceptors, *rods* and *cones*. The *rods* look a bit like rolling pins, and the *cones* like ice-cream cones. If you peel the back of the retina off, so the photoreceptors are exposed, it looks like there is a sea of little cells sticking out, which are the cilia, in between which are a few upside down ice-cream cones (the *cone* cells). If the cilia are not working optimally, the photoreceptor cells begin to degenerate and stop working; the retina becomes thinner and causes RP (retinitis pigmentosa). It is hard to predict the rate of degeneration of the retina in someone with RP as it seems to vary greatly.

The two types of photoreceptor cells, as well as looking different, also have different functions. *Rods* help us detect the difference between dark and light. *Cones* help us see colour. In fact humans have three different types of cones that help us see colour, allowing us to differentiate between all the different colours of the rainbow.

As mentioned above, a cilium is made up of different building blocks, or proteins. The BBS genes contain the instructions to make these proteins that form the cilium. We want to try and understand the biological function of each of the individual proteins within the cilium of the rod, and the cone. We know the rods and the cones have different shapes (ice-creams and rolling pins), and very different functions, so the protein building blocks that they are made up of must also be doing different jobs, in order for the cell to function. If we could understand some of the biology behind the function of the cilia in the eye, it may also help us learn more about cilia in other parts of the body. Like the different shaped cilia in the eye, those within the rest of the body also have varying shapes."

# Coping Better with Bardet-Biedl Syndrome

## An introduction to coping skills for the affected individuals and their loved ones.

**Annika Lindberg**

BBS Counselling Psychologist, Guys Hospital, London.

*Annika Lindberg is a Counselling Psychologist in genetics and assesses adults as part of the Guy's BBS Clinic in London. Patients are referred on to local services if needed, so that treatment can be accessed and funded locally to where people live. Annika spoke at the LMBBS Family Conference about coping skills for those affected by BBS:*

"In last year's presentation we looked at the occurrence of particular mental health problems in relation to BBS and found a high prevalence of depression, anxiety and Obsessive Compulsive Disorder (OCD). We discussed whether these psychological illnesses are part of the syndrome, or whether BBS patients are more susceptible to developing mental health issues as a result of having to cope with the many complex medical issues that come with the syndrome; we still don't know the answer to this, but it is likely to be a combination of both.

I think it is important to acknowledge the lack of services available for helping BBS patients with psychological health issues. There are just a handful of clinicians who work in this field and unfortunately we don't often see patients outside of the BBS Service, so we are only assessing and referring. In Psychology however, we look at 'Transdiagnostic Underlying Processes,' meaning things that underlie all mental health problems, across diagnosis, so it doesn't matter if a patient has BBS, or other medical issues, or even no medical problems at all, the processes that underlie mental illness are the same in everyone.

How well people cope psychologically with having BBS is not related to the severity of the condition. If someone is more affected by BBS it doesn't mean they will be more mentally disturbed, because we all respond in different ways.

For example, in Sweden, our country has the highest level of welfare in the world, but also one of the highest prevalence of suicide. People who have a lot of convenience and have easy lives, often don't practice their coping skills a lot and as a result, they may be less able to cope with difficulties when they arise.

Much work has been done in the area of Psychology relating to chronic illness and chronic pain. Psychologists working in these areas then look at ways of dealing with pain and illness issues, not necessarily with a view to cure the conditions, but to help people handle the pain/illness differently. Other mental health services, such as those dealing with OCD, depression, panic disorders, grief or loss of health and so on are all examples of services that BBS patients may be able to get some help from.

There are, however, ways in which we can help ourselves and some thoughts that people have in relation to illness are more helpful than others. For example, if you have a diagnosis of BBS you may think, 'Why me? I don't deserve this. Why is this happening to me?' These thoughts could lead to feelings of

anger and possibly sadness in the long term: 'This is chronic, how will I ever feel okay with this? How am I ever going to be able to cope with this in the future?'... These thoughts can lead to a particular set of emotions, including sadness, hopelessness, fear, anxiety and so on. It is important to understand that these kinds of feelings cause us to act in particular ways, many of which are intuitive to all humans. When we experience sad or anxious feelings we tend to act in ways to avoid the bad feelings, because we don't really feel good sitting with them. Often these behaviours we engage in become very counterproductive; basically we do things in order to alleviate the stress, but what we are really doing is prolonging it. Although avoiding feelings works well, and does indeed offer short term relief from them, in the long term the feelings will persist, and sometimes turn into moods.

More positive ways of thinking are: 'I may not like this condition but I'm going to do my best. I'm different, but if I believe in me, others will too. I'm not my condition, I'm also a person.' This would lead to emotions of hope, contentment, satisfaction, happiness and empowerment. It is not the condition in this particular example that is influencing how we feel, but the way we think about the condition- so basically it is never the actual event or situation that is happening, but how we perceive and interpret it, that determines how we are going to be feeling about it.

Looking at the underlying processes, we previously looked at the content of our thoughts, which is the output of what goes on underneath. We all engage in negative ways of thinking from time to time, although some people do it a lot more than others. One of the most common negative thought processes is worry and those who have a medical condition will typically worry about their body symptoms and health and the impact on their future. For example, 'What if I'm having kidney failure? What if I end up on dialysis? What if I die? What if nobody will want to marry me?' These kinds of thoughts are referred to as Type 1 worry. When we think these thoughts, we end up feeling anxious and we will look at the consequences of that in a minute. Another type of worry, which is often referred to as 'Type 2 Worry,' is the worrying about our own worrying. For example, you start worrying about your own internal processes like thoughts and feelings- 'If I continue worrying like this I will die from stress. If my thoughts don't stop they will drive me mad. Thinking like this means I'm weak and that I'm not coping as well as other people do'. I've seen a lot of people with medical problems who start comparing the way they are coping with their condition to the way their peers are coping. They may start feeling that they are not dealing with things quite as well, so they start to worry about their own coping responses.

When we talk about worry from now on I will be specifically referring to the thinking process rather than the feeling of worry. We all have negative triggering thoughts from time to time, but worrying is the persistent negative future oriented thinking that often starts with 'what if…' and typically become increasingly catastrophic. The negative consequence of worrying, is that it causes anxiety, leaving us in a state of paralysis when we want to accomplish something. There are things you know you should be doing, but the feeling of anxiety prevents you from feeling able to do so. When we are anxious, our ability to think logically is greatly reduced. Let's say you're off to a social event, you really want to go, but then you start worrying about it - 'What if nobody likes me? What if nobody talks to me? What if I'm going to make a fool of myself?' You start getting into such a state that withdrawing from the event becomes a real temptation. Usually by the time people become that anxious, their ability to think logically is so reduced that they are unable to think straight and it becomes impossible to complete their task and some form of avoidance, perhaps in this case by withdrawing or not going, becomes natural. This kind of scenario will make us more likely to avoid a similar scenario in the future and it will also make us less confident in ourselves over time.

Worrying also leads to procrastination, the tendency for people to put things off for another day or for later because they are a bit anxiety provoking. This can be problematic in individuals with BBS as many already struggle to get things done due to the condition.

Worrying increases feelings of helplessness and hopelessness, particularly over time. It leads to very negative mood states, including depression. As time goes on people lose confidence in their ability to do things because

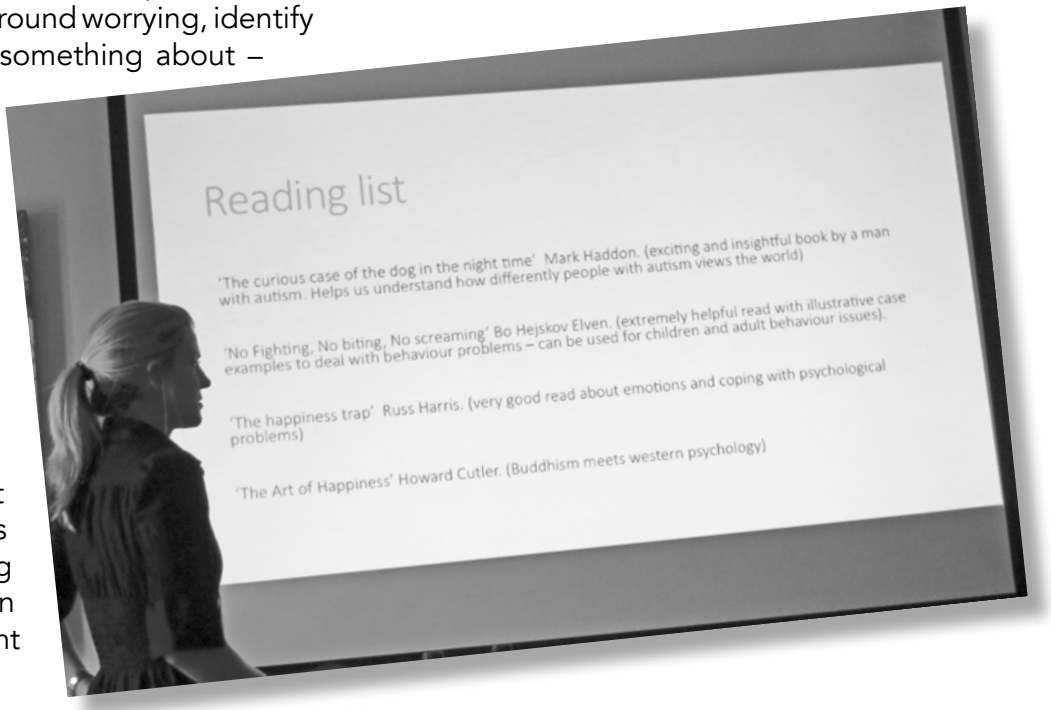
while they are worrying, they are not able to do anything else, so they are losing valuable skills as time goes by. That doesn't mean that it can't be reversed, however it is still quite a negative consequence.

One might wonder why people would continue to worry when the consequences are negative. Many people would argue that worrying is uncontrollable and unstoppable – but this is not the case. The truth is that many people confuse worrying with other similar but fundamentally different thinking processes such as problem solving. They might think 'thanks to worrying so much I acted straight away when I developed symptoms in my kidneys'. If you really analyse what happened here you will find that the person was first worrying ie. 'What if something is wrong? What does this symptom mean?' but at some point the process changed and the person thought 'I better go and see a doctor to check' – the last one here is not worrying, it is problem solving. Problem solving is a constructive thinking strategy, that is attempting to move forward from an issue- and it has none of the negative consequences on us emotionally as worrying does.

A very useful technique that I can't recommend enough, is called 'Worry Tree'. Next time you start worrying in response to a trigger, ask yourself, 'what am I worrying about and can I do anything about it?' If the answer is yes, then change your thinking strategy and engage in some problem solving. This takes time to perfect but generally people can become quite good at it, so rather than sitting around worrying, identify the things you can do something about – the things where you have some control. Start gathering information, maybe seek advice, improve your fitness levels or speak to a friend. Do something that can help your situation. If we go back to the worry tree, and ask ourselves the same question but find that the answer is 'no, I can't do anything about this worry', then there is still no point in worrying, as the worrying will only lead to anxiety and all the other negative consequences we talked about earlier. The recommendation here would be distraction, maybe some activity scheduling, plan something nice for the future or just do the very best you can to make yourself feel better about what is going on. Gathering information can be really useful. It is also good to have an understanding of what is going on medically, which can give us a sense of control.

Some people develop so called 'maladaptive coping strategies', alcohol is one of them. In the short term it makes us feel fantastic, it makes our problems go away. In the long term however, this is not the case; alcohol is a depressant drug and will make us more depressed. All forms of avoidance fall into the category of maladaptive coping strategies. Other forms include social withdrawal, procrastination, emotional suppression and other attempts of 'running' from feelings.

Another type of avoidance is called thought suppression - for example, people who avoid their own experiences of thoughts. For example: 'I'm having a bad thought about this, I must stop thinking about this, I must remove this thought from my brain'. This process causes the thoughts to bounce back harder because they have been given too much importance. Some people may have very disturbing thoughts for example, 'What if I pick up this knife and hurt somebody?' They respond to that thought by thinking, 'this is terrifying, this means I am



going insane'. In reality, that particular thought had no more importance than a thought about a green apple that was left on the breakfast table, however the way we respond to it is what determines how we are going to cope. What is important to learn here is that thoughts in themselves are never the problem- it is the way that we react to them that cause distress. If we dismiss a thought as simply being just a thought- it is not going to disturb us. If we believe our thoughts are facts, we will also believe the negative ones, and react with distress.

So what can we do to break the vicious circle between thoughts, feelings and behaviours? One technique that is easy to remember is called 'Opposite Action'. All feelings have a tendency to want to enhance themselves: when we feel negative, sad or worried, our minds will tell us to do all the wrong things such as stay indoors or don't get out of bed and so on. By doing the very opposite of what our mind is telling us, we can break the cycle and a really productive approach is to use distractions such as activities, social engagements, sports and scheduling of tasks.

A lot of people I have seen in the BBS clinic have very high levels of inactivity, spending enormous amounts of time indoors, doing very little. This leaves room for our minds to go a bit wild and we start worrying away, or ruminate about negative things that have happened to us in the past. Having a structure to the day and week is very important in order to reduce the opportunity to sit around and 'overthink' so setting ourselves goals to go out every day, do sport or socialise at least once a week is really important. Sleep is also very important as it affects our mood greatly, so try having regular bed times.

Being proactive in your attitude and your behaviour about the things that can be changed, and recognising one's limitations are also very important. This is particularly true for parents who have a natural tendency to give up everything for their children, in the belief that they are doing the right thing, however in the long-term the parents may fall ill, or become stressed, leaving them less likely to be of any help. A good analogy of this is the aircraft safety instructions, where you are told to place a mask on yourself before you assist your children. Children depend on their parents for both emotional and practical coping and parents of a child with BBS will know how dependent they can be. We need to think about the short term versus the long term consequences. Yes, I can give up my life right now, do everything I can for my child, but in the long term, is that really going to benefit my child in the best way possible? It is a balancing act and it's not an easy thing to do.

To summarise, I want to explain again why I chose this topic to talk about. I want our BBS patients and their families to have a more flexible way of thinking around the services that can be used for BBS by making people realise that many of the underlying processes that occur in individuals with BBS who have anxiety and depression, are the very same as for people in the general population who suffer these problems. As we all know, NHS services are slimming down rather than enlarging, so the chances of the BBS provision increasing any time soon is quite slim, and that's the realistic outlook. So rather than focusing on the handful of BBS Psychologists in the world who could help, trying to think of this as a psychological problem in relation to BBS rather than one that is for BBS patients. You could seek help for depression and anxiety from most Psychologists who, although they may not know anything about BBS, will know how to treat the anxiety and the depression- which is the part that they would be treating anyway. It is also common for psychologists to need to learn from their patients the things that may be specific for that patient medically or personally, if these factors complicate or impact on the ability to treat that patient. This is part of our job."

Finally, I have put the following reading list together which may be of some use:

*The Curious Case of the Dog in the Night time.* Mark Haddon

*No Fighting, No Biting, No Screaming.*
Bo Hejlskov Elven

*The Happiness Trap*
Russ Harris

*Art of Happiness*

The Dalai Lama & Howard C. Cutler

# Stefan Crocker: Personal Perspective

"I was diagnosed about 10 years ago in 2003. When I was 17, I went to check my eyes out to see if I could learn to drive and was told that I couldn't and that was quite annoying because I'm a sports car fan and Formula 1 fan. I left it on the backburner for a few years and thought I'd go and get some laser eye surgery because at this point I only knew that I had astigmatism. Around this time, I was told that I had something more severe wrong with my eyesight, and they sent me to Moorfields where I had some very interesting tests, including sitting in a darkened room for half an hour. The professor said that I had LMBBS and gave me some interesting advice, he said if I had a good circle of friends that liked swimming and playing chess, then I'd be perfectly fine; I'm not sure if that's worked.

What I'm covering mostly in this talk is independence as a young person, involving university, independent travel, and socialising in general. I used to be taken around quite a bit by my mum until she had a bad ME relapse, then I had to start being independent, which was a bit scary at first. Taxis were perfectly fine, however buses are a bit unreliable because they sometimes don't tell you when it's your stop even when you do ask them to which isn't very helpful. There's a new RNIB campaign, which I think is helping to try and make them better. I find trains a lot easier because you can book assistance. People who know me will be surprised because everyone knows I'm a fairly confident traveler now, but when I first started out, I was quite nervous. Now I travel everywhere and as long as I research in advance and communicate with people, everything goes fine; you can also apply this to flying. If you talk to the airport they'll help you collect your luggage and get to your destination; you can go abroad without having to have a chaperone, be much more independent and go with friends on package holidays.

At first, I thought talking about how to make friends was going to be difficult because my experiences are mainly from university days where my socialising was with clubs and societies, but then I thought that the most important thing was communication, so it could apply in general. I met my friends Tom and Dan when I went to a Rock Society social event, but then, I met more friends through them, because I didn't let my sight be a barrier to trying things, I just had to adapt to social events too. If you are at university or are thinking of going, contact your local sight charities and councils, as they can help you with social events and clubs. While I was caring for my mum, I found out about a disability football team, so I contacted them and explained the situation and joined the team.

Finding the right University can be quite a complex thing. I would say that campus university for a visually impaired person and a BBS person is not necessarily a bad thing. My own personal experiences are that a city campus is probably the best option because everything is nearby. My experience at Hertfordshire was that the disability support itself was excellent, however everything was far away, so it was difficult, whereas at Portsmouth University, everything is in the city, so you can walk everywhere which is brilliant. If you, or your children are planning on going to university one day, it's important to do a lot of research in advance; contact the university disability services and they'll help you.

I also thought I'd cover a bit about socialising in general. I've had some experience of going to gigs, with friends and talking to customer services in advance is usually very helpful. My best experience is with the O2 in London, which are fantastic to a point where I could probably go on my own. I don't know if I'm going to do that yet, the thought of it is a bit scary but I'm possibly going to do it in the future. It

always comes down to communication with people and good communication allows you to be much more spontaneous when you go out to restaurants, and pubs, and theatres and so on. If you just talk to people and explain the situation, someone will help you go through things, and this also extends to clubbing too. You can go out pubbing and clubbing with your friends as long as you've got reliable friends. If you are queuing to get in, tell a bouncer or member of staff that you are visually impaired and then if you lose your friends they tend to be quite good at finding and reattaching you."

**Q: As a young boy, has your journey been as you thought it would be, or have you surpassed what you thought you could achieve?**

**A:** I think the university is the biggest thing. When I first started a long time ago I was never really convinced or certain if I could get through with my disability, but now I'm quite proud of myself that three days ago I handed in my dissertation and I'm at the end of my university degree, and I've achieved that. I'm a lot more sociable… I thought I wouldn't be quite as sociable as I am now.

**Q: Now you've finished your degree, what does the future hold for you?**

**A:** Work, hopefully. I want to do politics. I sometimes consider going into parliament but I'm probably too honest, so I can't. So yes, it's on to employment next and carry on with life.

**Q: What did you study and what was your dissertation on?**

**A:** My dissertation was on the descriptive representation of women in the British parliament, and I was looking at how you could increase the number of women in parliament.

# An Introduction to Speech and Language Therapy Within BBS Clinics

**Caroleen Shipster**

*Caroleen Shipster is a paediatric speech and language therapist at Great Ormond Street Children's Hospital. She has worked with children from infancy to adulthood in a range of settings, which include pre-school settings, child development centres, community clinics, hospitals and different types of schools.*

*Caroleen has worked with children who have all types of speech and language problems including those caused by hearing impairment, structural difficulties, neurological problems, and learning difficulties. Caroleen has worked in the UK and the USA and has published many articles on speech and language difficulties.*

*In 1999, a paper was published which indicated for the first time that children with BBS had speech and language difficulties. In 2010, Professor Beales invited Caroleen to join the BBS Clinics team at Great Ormond Street. Caroleen was invited to hold workshops at the Family Conference and spoke during the morning programme about her role and findings to date:*

"It is a privilege and a pleasure to be part of the LMBBS Family Conference. Before I started attending the BBS clinics at GOSH in 2010 I looked at some of the literature on children with BBS. I found that there have only

been two studies published so far on speech and language skills in children with BBS. The first one was on a questionnaire study of 109 adults and children with BBS and the second one was a more detailed study looking at 21 children, aged between 3 and 18. The papers reported that about 50% of the individuals in the studies had some form of speech and language difficulty. The main difficulties reported were speech impairment and delayed language development. A few children also had hypernasal resonance (sounding as if there is too much sound coming from the nose) and others had a high pitched voice. Many of the children in the studies also had global developmental delay.

Since I've been at Great Ormond Street, I have seen 74 children with BBS. 37 of those children have had repeat assessments, which has been fantastic for me as a clinician, because I have seen at first hand, how the children change over time and heard the concerns as well as the progress made by the children reported by the parents. In total, I've done 111 assessments, and the age of those children has ranged from 10 months up to 17 years. The assessments in the BBS clinic are quick screening assessments, so we are just getting a snapshot of each child's abilities when they come to clinic. Some parents bring their children's local reports to the clinics, which is extremely helpful as it gives a lot more information as to how those children are developing.

What I have learnt is that there is a huge variation in the speech and language abilities in children with BBS. Not only are there many different types of communication problems but those problems can range from mild to severe as well. I am now going to describe some of the common patterns that I have seen. The first common pattern that I have observed is that the early speech and language milestones can be very delayed in some children with BBS. First words usually start to appear in children between the ages of 12 and 18 months but within the clinic, you often hear parents say their child didn't speak until they were over 2 or 3, or over 4 years in some cases, and sometimes it can be even later than that. Overall, there is a trend for expressive language milestones to occur quite a lot later in BBS. Alongside that, there can be a delay in other areas such as understanding of language, and the ability to pay attention to language based activities.

Looking at the patterns in a bit more detail, out of these 74 children, 31 of the children had no speech and language difficulties and their parents reported they had no current concerns about their children's communication skills even though their early language development had been slow in some cases. This implies that even though the early milestones can be delayed, a lot of children go on to develop normal speech and language skills as they get older. The 43 children with speech and language problems had difficulties, which ranged from mild to severe, across a range of areas. Problems with expressive language included slow acquisition of new words, difficulties putting sentences together, and difficulties describing a sequence of events or telling a story. Problems with receptive language included difficulties with understanding words and sentences; and difficulties following stories and more abstract language within the classroom and at home. A number of the children had social communication difficulties. Social communication difficulties is a term used to describe difficulties with social interaction such as not knowing about the rules of social interactions (i.e. what are the appropriate things to say to other people and when to take turns in conversation etc) and difficulty interpreting the emotion of what other people are saying. Some of the children had a diagnosis of autism.

The speech (sounds used in words) difficulties are extremely interesting in BBS. There can be delayed acquisition of speech sounds and some children do not babble when they are young. Other children do not develop speech sounds in the expected pattern, or have unusual speech patterns (this is called disordered speech or dyspraxia) or they can have a "favourite" sound that they use in a lot of words. They may leave sounds off the ends or the beginnings of their words or have vowel distortions. They might sometimes have a halting rhythm to their speech. Sometimes the speech can be quite difficult to understand. If speech difficulties are present, speech therapy can often really help to improve the clarity of the speech and you can see this improvement when the children come back to the clinic after they have had the therapy.

There are some factors that influence speech and language development such as developmental delay, learning difficulties, hearing impairment and autism. The school environment and the amount of therapeutic support the child receives can have a tremendously positive effect on a child, and of course the home environment is always the most important influence on children's communication skills. I am now going to talk about these factors.

It's very helpful to have an overview of your child's development in other areas apart from language and then to look at the speech and language development in relation to that to see whether the speech and language skills are developing as well as you would expect given the child's overall level of ability.

Hearing impairment is really worth mentioning. I have seen that a number of the children in the clinic have glue ear, and the reason for that is that all young children in general have a risk for glue ear. If it is treated effectively, it can have a really positive effect on helping develop speech and language. In BBS and other conditions where a child has a range of medical problems and there are a lot of concerns, something like glue ear hearing loss can be overlooked. So as parents, it is very good to be thinking, 'is my child hearing as well as possible, or does he/she need a hearing test?' In clinic, where the children have had treatment for glue ear, I've had a couple of parents report that speech and language skills have really improved following treatment for this.

If autism is present, that is going to have a negative influence on the development of language and social skills. Sometimes there may be a history of speech and language difficulties within the family that has nothing to do with BBS. Speech and language difficulties can be very genetic, and we often ask whether other family members were slow to talk, just to try and understand why a child might have a particular pattern of speech and language.

In conclusion, if you have concerns about your child's speech and language skills do ask for a referral to your local speech and language therapy services. If your child already has a therapist, do attend some of the speech sessions so you can work together with the therapist to improve your child's communication skills."

Below is a list of some useful websites. The Autistic Society has got some fantastic resources and there is also information on some of the dyspraxia websites.

**ICan:** An excellent website offering a range of resources for early years to age 16. **www.ican.org.uk**

**Talking Point resources Catalogue:** Looks at the development of communication skills in children from pre-birth to 17 years old. Includes a progress checker, organisations that can help, DVDs and books, free parent resources, glossary of terms, a communication cookbook, and definitions of difficulties. **www.talkingpoint.org.uk**

**Afasic:** Explains the help available for children with speech and language impairments. Includes publications for parents, FAQs section, Parents' Helpline, Parents' Forum for parents to share information and talk about speech and language impairments, and a list of workshops and courses. **www.afasicengland.org.uk**

**Literacy Trust:** This website has an Early Years section with some good resources and information on the literacy development of children. **www.literacytrust.org.uk**

**The National Autistic Society: T**his website has very useful links for parents on therapy approaches under 'Communicating and Interacting'. **www.autism.org.uk/**

**The Dyspraxia Foundation:** Information about dyspraxia and its effect on Speech and Language and other aspects of a child's life. A very informative website with lots of useful links. **www.dyspraxiafoundation.org.uk**

**Signing - Makaton signing:** This Website is full of information about Makaton, accessing resources and courses, and the impact that Makaton can have on lives. **www.makaton.org**

# Results from the BBS Clinic Questionnaire and Future Research

**Dr Elizabeth Forsythe**

*Dr Elizabeth Forsythe is a clinical geneticist, working with Professor Beales, as part of the genetics team in the BBS clinics in London; she is also a member of Professor Beales' research team in his Cilia Disorders laboratory. Elizabeth has a special interest in understanding genotype-phenotype correlations and developing therapeutics for Bardet-Biedl syndrome. Elizabeth has been awarded a Medical Research Council fellowship, which allows her to remain with Professor Beales' team for at least the next three years, where she will continue to work on therapies for Bardet-Biedl Syndrome. Professor Beales described achieving the award as a significant watershed, because it means the Medical Research Council, one of the big funders in the UK for medical research, are putting their confidence not only in the work of Elizabeth and the team, but also in the idea of therapy development. Having worked in the BBS clinics on and off since their inception in 2010, Elizabeth talked about what has been learned from the BBS clinics so far and how that information is to be used in the future.*

"In 2011, Kath Sparks and myself, together with Julie and Tonia, put together a questionnaire to find out what really troubles people with BBS. We wanted to know how those of us who work in the clinics and those who do research on BBS in the laboratories can help. We asked about eyesight, weight management, kidney problems, and a lot of the other things that we talk about in clinic. I would like to say a very big thank you, because so many of you filled in this questionnaire and it really was very helpful.

We know there are around 400-500 people living with BBS in the UK and 235 of those completed a questionnaire, which is fantastic. The oldest person who replied was 56 and the youngest was 11 months. We had replies from all over the country, which is really helpful as we can be much more confident that what we are getting back is a true picture across all ages and backgrounds. Our aim is to look at what can be predicted about a person based on their genetic results. As you may already be aware, we all have two copies of each gene, one from our mum and one from our dad. For 8 out of 10 people who come to the BBS clinics, we are now able to identify two gene changes in a BBS gene, which makes it probably the best laboratory service for BBS in the world, and that is down to the scientist in our lab, Dr Beth Hoskins.

If you have BBS, you might have a change in BBS1 or BBS10 and so on, however we have found that the *kind* of genetic change you have in that gene can make a difference; there may be mild change, severe change or a mix of the two. The work we have done so far has really focussed on eye problems, kidney problems, and also what we call cardiovascular risk factors, for example, having high cholesterol. The foremost finding is that everyone is very individual. We did, however, identify some trends, which suggest that people who have mild changes seem to have, on average, maintained their vision for longer, are less likely to develop kidney problems, and also have slightly better blood test scores. In terms of vision, those who have the very mild changes seem to maintain their vision for about seven years longer than people with the more severe changes. However, you may get two people with exactly the same change in exactly the same genes, and they will experience BBS in very different ways and because of that, we are unable to use that information at this point in time, to predict how things will unfold on an individual level.

So now that we have collected all this information, I am sure you are interested in finding out how we are going to use it in the

future and how we are using it now. In Phil Beale's lab, we are all working hard trying to develop treatments for Bardet-Biedl Syndrome. For example, we are working with a drug that has been used with some success in other genetic conditions and we are trying to develop some new kinds of drugs, with the aim of converting the more severe changes in BBS genes into milder changes to enable that person to maintain their vision for longer, perhaps make them less likely to develop kidney problems and give them slightly better blood test scores. That is what we are hoping for.

We are testing these treatments on skin cells from BBS patients; some of you who attend the BBS clinics in London will already be aware of this, because some of you have very kindly donated a small skin sample for us. We are able to change the skin samples by a process called reprogramming, to turn them into cells that behave like retinal cells. The retina sits at the back of the eye and is very instrumental in allowing us to see and what some of our colleagues in Cambridge can do, is change these cells so they can behave like retinal cells, which means we can test out these drugs on cells that behave like functioning eye cells, which would give us a really good indication about whether these drugs could help maintain people's eyesight for a longer period of time. We can also look at people who have the same genetic change in the same gene and ask why they experience BBS in such different ways.

It has been a really amazing couple of years in the BBS clinics for me. I have really enjoyed it and I have enjoyed getting to know so many of you and because of your input, I think together we can make a difference for people who are living with BBS in the future, which is really exciting. I feel very hopeful that we are on our way to getting some of the answers to some of our questions and hopefully to developing some treatments in the future."

**Q: How close can you get in terms of generating retina cells from stem cells and why is it so difficult to replace degenerated retinal cells with newly generated stem cells or retinal cells?**

**A:** A lot of people have been working on this and a lot of progress has been made in the last couple of years. Within ICH there are a couple of specialists who have developed a new technique where they can do extremely well in terms of developing the different layers of the retina. It used to take months and months to get to the point where you could develop photoreceptors, however now they have developed a new technique where they can develop photoreceptors within 30 days, which is really extraordinary. Moving on to the second question, the difficulty with replacing these cells is that first of all you have to grow enough cells, then you have to inject them into the right place, and then the other difficulty is that they have to sit the right way round. So if we can get some of the photoreceptors in place, only so many of them will survive and only so many of them will then end up the right way round, which is why it's really important that we take lots of different angles on this, and work on different therapies.

# Rebecca Goodman: Personal Perspective

"My name is Becky Goodman, I am 33 and I was born with LMBBS. I was diagnosed very early when I was a baby. I went to a primary school in Weston-Super-Mare. Although it was a school for children with learning difficulties, I only had visual impairment. When I was 16, I went to the West of England College in Exeter. At the college I learnt how to be independent, doing such things as cooking, cleaning, and shopping. I also completed courses in further education, including an NVQ in administration. I also did work experience placements.

I left the college when I was 22 and lived in a transitional house with one other student and a support worker. I was there for about two years and then found my own flat in Exeter. I have support workers for 14 hours a week, and care workers who help with personal care. I have done various voluntary jobs, but due to health issues, I have been unable to join the job market. However, I enjoy a good social life. I go to a Monday and Tuesday club at Sense. I go wheelchair dancing with my boyfriend, who has ataxia, I go to meetings of Torch Trust, a Christian fellowship group every month and I go to church on Sundays.

I also go to Slimming World, where I've lost 2.5 stone. I don't let LMBBS get to me now. I've had it for 34 years now and I just treat it like it is normal… It's only me and my family who has LMBBS but I've just learnt how to live with it. It helps having Chris to support me as well and he's come to understand it better. Chris and my granddad say how proud they are of me because it's hard to lose the weight. By doing Slimming World for just over a year now, I've gone from size 20 down to a 14 just within a year. And even though I have diabetes and everything else that goes with it, since losing the weight I don't have to use my CPAP machine so much now and I'm hoping to come off it this year or reduce the pressures down.

All those parents who have children with BBS, it was hard growing up as a child having the syndrome, but going into adulthood it's not as bad. It has been quite hard, but I've had lots of support from my family and friends, and especially Chris. I love living independently and being able to see my friends and go where I want to. I feel that I have achieved a lot so far in my life, and I look forward to enjoying life to the full."

# Workshop 1:

**Professor Phil Beales**

Question and Answer Session

**If a BBS gene isn't found when the test is done, does it mean that the person hasn't got BBS?**

No, because diagnosis is as much about the clinical appearance. This society laid down the original criteria used worldwide for making a diagnosis and probably 80 - 90% of the time we go on the clinical findings. The genetic test is particularly useful for those few where we are sitting on the fence. A confirmatory test, to us, is not that surprising, because we are probably fairly clear about the clinical presentation, however there are still a handful of genes to be found. We have had a few referrals recently from people who have got retinitis pigmentosa and they have turned out to have mutations in Bardet-Biedl Syndrome genes which has been really surprising because the patients haven't got anything else, such as the weight problems and extra digits.

**Is there a test for being a carrier?**

Yes there is, but only once we know which gene is involved in that particular family. Carrier testing is accessed via the GP, who will make a referral to the regional genetics lab.

**Does everybody with BBS2 (for example) present the condition in the same ways?**

No, and that is what we find absolutely fascinating. We are now at a position where we have been able to marry up how people present clinically with the genetics data. There are some patterns starting to develop and we are going to have to look a bit deeper beneath the surface, but what we can say is that we don't have a really good strong correlation between the gene involved and possible symptoms. On the face of it, it doesn't matter whether you have BBS1 or BBS10 as to whether you might be prone to getting high cholesterol levels or high blood pressure and those kind of things. However, there is some indication that if you have a common mutation of BBS1, which we would describe as a mild mutation, it may be that visual impairment is delayed for up to five or ten years.

**Do you think liver scoliosis could be linked with LMBBS?**

Yes, it can be. You get cysts in the liver, like you do in the kidney and embryologically the liver and the kidney develop in the same place, which might be part of the reason. We are seeing an increase in the number of cases of fatty infiltration in the general population, due to the obesity epidemic. Essentially, it is the amount of fat that is being stored in the liver that becomes damaging to the cells in the liver. We would be very happy to liaise with liver specialists where required.

**Is there a breakthrough in gene therapies for BBS?**

Yes, I think so. I don't want to give false hope, but the fact is there are already diseases out there that are benefitting from things like gene therapy. Moorfields Eye Hospital, London, was the first in the world to have success with gene therapy for the eye. They have now started a trial programme where they inject the correct, full length gene back into the eye to see whether or not we can restore or rescue the photo receptors. We are working with Professor Robin Ali and his team to develop gene therapy for BBS1 first of all, because we have BBS1 mice in our lab. It is looking very promising.

**Is there some hope that vision could be restored?**

We can say that if you already have complete blindness and most of the photo receptors have died, nothing that we know about right now is going to reinvigorate them to come back to life again. However, if someone has a good level of partial vision, then maybe we could actually halt the deterioration or certainly slow it down. We are doing genetic correlation tests to try and work out when the eyes generally start to fail, according to which gene might be involved and so on. Then we will choose those children who we think are most likely to benefit from early gene therapy. At the moment, we think it might entail just a single injection into the retinal layer behind the eye, however we don't know how long that is going to last, or whether it needs to be repeated. Robin Ali has beautiful videos of adult patients who have got RP. They were sent into a maze before and after treatment and they were much better at navigating the maze after having the injection. It doesn't mean they are now able to read tiny typeface, however there was an improvement, but there had to be photo receptors present that were still healthy.

Stem cell therapy, which will actually restore those missing or dead photo receptors is still some way off yet. My colleague, Professor Jane Sowden, has succeeded in delivering stem cells to the back of the eye. She has shown that these stem cells will live and function in mice, but she hasn't been able to show that they can be turned into photo receptors that will then connect back to the brain. My gut feeling is that the nerves will probably find their way to make some of those connections.

There is also a lot of work going into developing artificial retinas. This approach is based on cramming high resolution onto those little CCD chips we have in our camera phones or in our digital cameras. The idea is to pop those into the back of the eye and connect them up. Early studies only had four or five pixels however they still managed to get some very blurred vision. Now I think they are up to 32 pixels, but they need to get into the thousands really.

**Do you collaborate on what you are doing therapy wise?**

Yes we do. We are about to apply for a big funding grant from the EU, probably in October, so I'm heading to Strasburg in June to meet up with four different teams. Dr Helen May-Simera is moving her lab to Mainz, Germany and will be working on a lot of therapeutic areas there as well and so their team is coming together with us, to put together a big application to accelerate this forward. As usual, what you need for any kind of grant funding, is preliminary data, so you almost have to have done the experiment before you apply for the money to do the experiment, which is annoying, but is the only way that they can be certain that you are the guys who can deliver.

**Are there therapy studies relating to weight?**

This is a key area that we need to be focusing on; we need to find a better way of treating the propensity to gain weight. There are a whole bunch of debilitating issues around carrying weight such as the strain on joints, cardio vascular risks, high blood pressure, Diabetes and high cholesterol. We now understand where a lot of the weight issue is coming from, it's not completely solved, but we think most of it is happening because of a resetting of the appetite centre in the part of the brain called the hypothalamus, which tells us when we are full. We will continue to look into correcting the weight issue.

**We seem to have a lot of support available at the clinics.**

The real reason why our clinics work is because of people like Tonia, Julie and Kevin who attend the clinics and co-ordinate what is going on. Without their support the clinics couldn't run as successfully as they do. The clinics ensure there is access to a variety of services. The doctors and other professionals talk to you respectfully and have a connection with the patient. Parents can be reassured that young adults who want to be independent can talk to the professionals safely.

**Is there any research into the dental side of the syndrome?**

There is no research into this at the moment

but dental overcrowding is a common issue.

**When my child was diagnosed, I tried to search for information online and read things, but the most beneficial thing has been coming to the Conference and hearing from the people who are actually involved in the research. This has given me reassurance that there are people out there who are working on helping my child, and that's a good perspective for me. Thank you.**

# Workshop 2: Skin Biopsies and Research

**Dr Elizabeth Forsythe and Kathryn Sparks**

Dr Elizabeth Forsythe held an afternoon workshop together with Kath Sparks, BBS Clinical Nurse Specialist, to talk in more depth about skin biopsy research and to answer questions. Dr Forsythe explained that they are looking to take skin biopsies from people who have BBS to enable them to understand the cells. The team need to find patients with the specific genetic change that they are looking for, as a starting point, in terms of testing therapies. The skin biopsy involves taking a little bit of skin, about the size of a matchstick head, from the top of the back of the arm where it is not very noticeable. Patients are given a little bit of a local anaesthetic which makes the area numb and the biopsy itself takes around 20 seconds.

**Who will be helped by these biopsies?**

Those who have some vision remaining are more likely to benefit from treatment. Our main focus is to develop a drug that will help people who haven't lost their sight yet and start with trying to slow down the deterioration.

**Why do you need the biopsies?**

We have to make sure the treatment is safe, so we first try it out on our cells, then animals and so on before it comes to humans. Realistically, this is going to take some time.

**So there's a light at the end of the tunnel?**

I think there is and it's not just Professor Beales

who is working on these different kinds of treatments. There are lots of people in different parts of the world who are working together, which is a relatively new thing in science, because there used to be a lot of competition. It is exciting to know that there are lots of different treatment options being developed that have really excellent potential, so yes, I think that's very good news.

**Where do you do the biopsies?**

The biopsies are carried out at the BBS clinics in London, at Great Ormond Street Hospital and Guys Hospital. For those not within the London area who would like to participate, we might be able to make some arrangements.

**What do you do with the skin once you've taken it?**

The sample is taken back to the lab where we will grow skin cells. We will grow some of the skin cells into retinal cells, with a view to testing out the drugs on these eye cells as well. We are also looking to see if we can find out why people have BBS in such different ways.

**Can you give me any information to take me a step further to understanding genes?**

We all have lots of genes, probably 25,000

genes, and they are like little packages of instructions that tell our bodies how to grow and develop, to tell the body that this person should have blue eyes or brown eyes, and they should have blonde or brown hair colour and so on. With regards to BBS, I think we are now up to 19 different genes that we know about that cause BBS, and in about 80% of people who have a clinical diagnosis of BBS, we are able to find a genetic change that causes the BBS.

**What is happening with research on retinal calls?**

People are working on trying to make sheets of retinal cells that you can put in the eye, but there are difficulties with this including factors like blood supply, providing a functioning, nutritious environment for the cells and the correct positioning of the cells; it is important, ongoing research. The main thing is that we try and tackle this issue from lots of different angles, so trying to make retinal cells we can put back in is one thing and trying to make drugs that will help the retinal cells survive is another thing.

**How long will it take to develop these drugs?**

It is very difficult to say because we don't know whether there are going to be stumbling blocks along the way. We are trying to think both short term and long term by testing drugs that are already on the market to see if they work and then also trying to develop new ones. If some of the drugs that work are already on the market, it will be about five years, however developing new drugs is a longer process and takes, on average, something like 15 years.

# Weekend Round Up











# My Time at the Conference

Hi all

You may remember me from a previous newsletter, but if you are new to the Society, I will briefly introduce myself to you.

My name is Aneeba Ahmed and I am 23 years old. I know what is like to live with LMBBS and what it is like to cope with it and with everyday tasks. I was diagnosed with the condition at birth and it affects my sight, my hearing, my speech, my weight and many other parts of my body as well. I also have got another condition called Spondylosis which affects my back and my legs.

I go to a specialist college in Birmingham, which is called Wilson Stuart where I am on a 19-25 LIVE programme course at the Hive, which will help me to get a job in the near future. I really like going to Wilson Stuart because I love socialising with my friends and with my TAs and having a laugh with them. I enjoyed going to Boldmere Court Care Home as a work placement, and also I like to learn new skills as well.

In my spare-time I like to listen to music, singing, speaking to new people about LMBBS, socialising with my family and friends, reading books and doing Braille.

I enjoy going to my LMBBS clinic appointments at the Queen Elizabeth Hospital as I see seven specialists who are fantastic and very helpful to me, as I don't have to take a lot of time out of college any more, which is a good thing!

This year in my Easter holidays, I went along with my sister Hena to the LMBBS conference in Northampton at the Hilton Hotel which was fantastic. It was inspiring to talk to other people who are new to the condition about the impact that it can have on you and what support can help. It was good sharing my own experience with the condition as well.

It was so fantastic to meet up with some of the people that I have been speaking to and it was also great playing bowling and making new friends. I really enjoyed making a Mosaic with Hollie which was lovely. I felt that these guys made me feel really welcome at the conference and also the staff were so friendly as well.

I never let LMBBS stop me from achieving my dreams and goals in life. At next year's conference, I hope to give a personal perspective talk and also to make some cards and wristbands to sell.

If you want to know more, join me on my LMBBS page which is called 'My Life with LMBBS' and feel free to add me as well. I hope to see you at the conference soon.

**Aneeba Ahmed**

# DELEGATES' COMMENTS

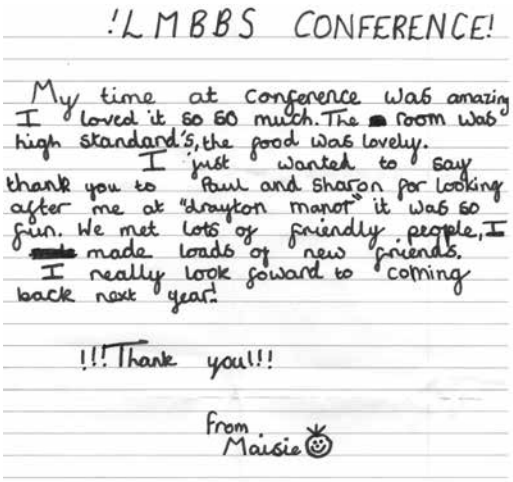
**What did our delegates say about Conference 2014?**

"Thank you – it has been a very welcoming conference and great to meet others at last who are facing similar issues that we have as a family."

"Really informative as a professional, an opportunity to hear families' experiences and update on research"

"Brilliant! Always very interesting with new things learned every time."

"Child care great! Liked the kids/carer concept, gave us parents confidence about safety; my child didn't want to leave the crèche!"


"Excellent as always, kids had a fantastic time and we really appreciate it."

"Always interesting to find out how much we are advancing in our knowledge of BBS."

**The LMBBS Annual Family Conference is a fantastic weekend full of learning, networking and social opportunities. A booking form for Conference 2015 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.**

# Annual General Meeting

**The Hilton Hotel, Northampton**
**26th April 2014**

**Minutes of 2013 AGM**
The Minutes of the Annual General Meeting on Saturday 20th April 2013, previously circulated, were agreed and signed.

**Apologies for Absence**
Apologies for Absence were received from: Terry Begley (Senior), Michelle Begley, Jenna Hook, Daniel Scott, Jill Stirland, Danny O'Sullivan and Bernie O'Sullivan.

**Election of Honorary Officers**
Of the current Officers, (Phil Humphreys, Chairman; Steve Burge, Vice-Chairman; Kevin Sales, Treasurer; Julie Sales, Secretary; Conference & National Co-ordinator, Chris Humphreys and Newsletter Editor, Tonia Hymers) all officers were eligible for re-election and were duly elected unopposed. Committee member, Emma Oates, was elected into the role of Fundraising Co-ordinator.

**Election of Committee**
Of the current Committee members (Richard Zimbler and Allan Clark), both had a further year to run in their present role. There were two spaces on the Committee and nominations were invited. Claire Eccles was nominated and duly elected.

**Chairman's Report**
It is clear from the Trustees reports that it has, once again, been a busy year. The Society continues to grow, yet despite this growth, as a community, our membership has never felt closer. Numbers continue to rise, mainly due to the success of our BBS Clinics and with this increase in membership has come an increase in the number of generous individuals committed to helping their charity move forward with purpose. My thanks go to all those who have given up valuable time over the past year to support the work we do. Without this support, we would be unable to meet our goals and objectives. I would like to thank all of the committee for their sterling work and commitment.

I would also like to thank Phil Beales for his continued dedication to LMBBS, without whom we would not be where we are today. We are now into our 4th year with BBS Clinics and as a result we are able to reach out and support so many more families and individuals. Credit must also go to our BBS Clinic Co-ordinators, who have worked so ably over the past year ensuring our patients and their families are well supported to attend this vital service. The aim of the BBS Specialist Clinics is to ensure patients receive expert attention and management and should bring about a major change in how BBS is managed, with a focus on diagnosis, early intervention and good health management. New patients are still coming through, with some parents self-referring, so raising awareness is still a crucial area for us to focus on.

One way of achieving this is by attending Disability Events, such as Sight Village, for whom we now have a team of volunteers. We also attend relevant conferences and maintain good contact with similar organisations. In June 2013, LMBBS assisted in the organisation of the first Ciliopathy UK Family Weekend Conference for families and professionals from various small rare group organisations. This was a tremendous success allowing networking between all parties. It is hoped to make this a Bi-Annual event. We thank Drina and Michael Parker for continuing to represent us on the Board of the Ciliopathy Alliance.

Conference 2013 was again a great success, surpassing previous years and thanks must go to Chris Humphreys, Tonia Hymers and Julie Sales for their commitment to this event. We now have good links with the Norway branch of LMBBS, who are keen to learn from our specialist clinics. Our Vice-Chairman, Steve

Burge spoke at the Norway BBS Conference in March and was made most welcome. The Conference continues to be the focal point in the LMBBS calendar and is looked forward to by many of our members and professionals who enjoy the chance to learn more about the syndrome, both from the speakers and the delegates.

I have kept the best until last. I would like to give huge congratulations to Tonia and Julie who earlier this year were successful in their bid for a grant from Genetic Disorders UK, as part of their Jeans for Genes campaign 2014. The grant will enable us to employ a part-time Child Development Officer to support our families further. Our most heartfelt thanks go to the families who were willing to go forward as Media Volunteers; the success of the application is thanks to their generosity.

**Treasurer's Report**

For the financial year, 1st January 2013 to 31st December 2013, the Society received an income of £41,740. Although this appears to show a decrease in income of around £7000 compared to 2012, it actually reflects the change in the Conference booking procedure. Previously, we would book hotel accommodation and guests would pay the Society, however now, guests make their own bookings, so we have less income via delegate's contributions, however we also have a smaller conference bill.

In April 2013, we received a grant from Children in Need in the sum of £3500 to pay for the children's activities at Conference 2013. Our grateful thanks go to all those involved, the children and young adults had a great time. Fundraising and donations have decreased over the year, but only by just under £1000, which, bearing in mind the current economic climate indicates a great effort by all those involved.

The Society's expenses for the same period have reduced considerably to £42,800, a decrease of £14,000 compared with 2012. As indicated previously, the main contributor towards this decrease is the new conference booking procedure. A further decrease in cost is due to adult delegates, who have BBS, now paying for their hotel room; all other conference costs are met by the Society. This has reduced the cost of the Conference considerably, bringing the total cost to the Society down to £17,000.

In financially challenging times, every charity relies on its regular donors and regular income to enable its core work to continue. We have always rejected suggestions of membership

**PHIL HUMPHREYS**
**Chairman**

**STEVE BURGE**
**Vice Chairman**

**CHRIS HUMPHREYS**
**National Co-ordinator**

**JULIA SALES**
**Secretary**

**TONIA HYMERS**
**Newsletter Editor**

**KEVIN SALES**
**Treasurer**

**EMMA OATES**
**Fundraising Co-ordinator**

**ALLAN CLARK**
**Trustee**

**CLAIRE ECCLES**
**Trustee**

**RICHARD ZIMBLER**
**Trustee**

fees however we do have a 'Friends of the LMBBS' scheme, with many of our members making regular donations or holding fundraising events. Around £3,000 is paid into the charity's account every year, via Standing Order, by our 'Friends' and we are so grateful for their support.

In summary, the Society's main expense is always going to be the Annual Family Conference; however we have seen this figure reduce quite considerably this past year. Our second largest expense is the production and 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 via our web page. 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.

**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**
In the absence of any other business, the meeting was closed. The date of the next meeting was set for Saturday 18th April 2015.

# CONTACT DETAILS

## www.lmbbs.org.uk

**National and Conference Co-ordinator**
**LMBBS Clinic Support Worker**
Chris Humphreys
10 High Cross Road
Rogerstone
Newport
Gwent
NP10 9AD
01633 718415
chris.humphreys4@ntlworld.com

**Newsletter Editor**
**LMBBS Clinic Support Worker**
Tonia Hymers
43 Balton Way
Dovercourt
Harwich
Essex
CO12 4UP
07805 685342
toniahymers@btinternet.com

**Secretary**
**LMBBS Clinic Support Worker**
Julie Sales
1 Blackthorn Avenue
Southborough
Tunbridge Wells
Kent
TN4 9YA
01892 685311
kevin.julie1@btinternet.com

**New Families Contact**
Claire Anstee
After 4pm, Monday - Friday
01604 454477 ansteeclaire@gmail.com

**Adults/Young Adults Contact**
Steve Burge
Flat 9
Pocklington Lodge
Rylett Road
Shepherds Bush
London
W12 9PQ
Mobile 07833 228 463
Skype: budge-2005
Twitter: @drumfeather.

**Special Educational Needs Contact**
Dianne Hand
0161 442 0989.
diannehand@live.co.uk

**Fundraising Co-ordinator**
Emma Oates
The Old Cow Shed
Knaresborough
Bishop Monkton
North Yorkshire
HG3 3QQ
01765 676578
07861 770164

**Friends of the LMBBS**
Kevin Sales
1 Blackthorn Avenue
Southborough
Tunbridge Wells
Kent
TN4 9YA
01892 685311
kevin.julie1@btinternet.com