

LMBBS Conference Report 2013

Contact Details

National and Conference Co-ordinator

Chris Humphreys: (01633) 718415 or by e-mail at chris.humphreys4@ntlworld.com

Newsletter Editor and LMBBS Clinic Support Worker

Tonia Hymers: (01255) 551886 or by e-mail at toniahymers@btinternet.com.

Secretary and LMBBS Clinic Support Worker

Julie Sales: (01892) 685311 or by email at Kevin.julie1@btinternet.com

Fundraising Co-ordinator

Emma Oates: 01765 676578

The LMBBS web address is www.lmbbs.org.uk. All of the above contact details are posted on our web site.

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Foreword

Another year, another conference and those who attended will agree, an excellent time was had by one and all. Once again, this is thanks to the the amazing support of our members and their fundraising and donations; as you will have read in your Summer Newsletter, we have seen some incredible feats of courage and determination in the name of fundraising this year.

We are delighted to report that at our Annual General Meeting, Steve Burge was voted in as Vice Chairman; Steve is a long standing committee member, a contact point for those with BBS and is also a volunteer at the specialist Clinics at Guys Hospital, London. With our excellent line up of speakers and facilitators, there was, once again, something for everyone. We had a theme of 'Exercise and Healthy Living' and a highlight of the presentations was the personal perspectives from Shane Ryan and Hannah Young, who with their success in the Sporting World have shown what can be achieved through exercise and healthy eating.

It would be remiss of me not to thank our voluntary care team, who come back year after year and enable our children and young people to make the most of their weekend. Without their commitment each year, our weekend family conference just could not happen, our heartfelt thanks go to you all. We were fortunate enough to receive a grant from BBC Children In Need to pay for the children's outing and activities and our thanks go to them for their support.

A special mention goes to two very special young ladies, Emmy and Holly Anstee, who spent many hours over the weeks leading up to the conference making little LMBBS keepsakes. They had their own table during the weekend, inviting delegates to make a donation in return for one of their gorgeous little craft items and made a whopping £75 for the Society. Well done girls! Our heartfelt appreciation goes also to a friend of Laura Dowswell, who donated a big box of handmade greetings cards. They were absolutely beautiful and must have taken many hours to make. We were overwhelmed at the generosity of this very special lady.

We have stressed in the past, the importance of the evaluation forms, as they form the basis of future conference planning. Unfortunately, very few were returned this year, so if you still have a form, it is not too late to return it. Alternatively, e-mail one of the committee with your thoughts 'good or bad'; it is your conference!

Finally a big thank you to YOURSELVES, for without you and your continued support, our conference would not be the resounding success it has become. We look forward to seeing faces old and new at Conference 2014.

Chris Humphreys

Update on Research and Study of LMBBS

Professor Philip Beales

Professor of Medical and Molecular Genetics

Wellcome Trust Senior Research Fellow in Clinical Science

Honorary Consultant in Clinical Genetics

President of LMBB Society

We are in our third year and hold clinics across four centres in Birmingham and London. Every clinic is delivered by a multi-disciplinary team and altogether, there are around 70 people involved; without the support of the LMBBS, our clinical nurse specialists and the admin team that runs behind the whole clinic service, it simply would not exist. We have around 440 patients attending and we seem to be receiving a healthy number of new referrals. The challenge for LMBBS is that we are having to increase the number of clinics and are beginning to creak at the edges. The managers are breathing down my neck asking how we are going to pay for this, so where does the funding come from?

Our funding comes from the National Specialist Commissioning team which is part of the Department of Health. There is a lot of uncertainty concerning the future of services like ours because the NHS has been restructured and the group of people who looks after our service has been dissolved. Next year we will have to start re-negotiating our package again, ready for 2015.

The 152 Primary Care Trusts have now been replaced by more than 200 GP organisations called the Clinical Commissioning Groups (CCGs). The CCGs will be responsible for close to 60% of the NHS budget and every GP surgery has to be part of a CCG. It is hoped that these changes will enable the NHS to become more efficient and improve quality of care.

CCGs will decide whether or not to pay for any hospital care a GP thinks the patient needs, much as the PCTs have always done, however the government thinks the CCGs will be better placed to make these decisions because doctors and nurses will be involved in the process.

So how does this work? Under the old system, the Health Secretary set the policies and the Department of Health passed those policies down through the Strategic Health Authorities and on to the PCTs, who had to ensure that all of these policies were implemented. Under the new system the Clinical Commissioning Board will take charge of overseeing the NHS from the Department of Health. They will work from their base in Leeds through four regional centres which are then devolved to 50 local offices around England. Local councils will have more of a role in tackling public health problems such as obesity and a new body, the Health and Wellbeing Boards, have been developed to work with those particular councils to link

with the work of the CCGs. Another national body called 'Healthwatch England' has been developed as an independent consumer champion for health and social care in England. Working with a network of 152 local Healthwatch teams, their aim is to ensure that the voices of consumers and those who use services reach the ears of the decision makers, who by law, have to listen and respond to what they have to say.

Moving on to our service and in particular, genetic testing, this is a unique component of our clinical service, which we are very fortunate to have; none of the other specialised services have a genetically defined component or a laboratory that is set aside to do genetic testing. There are now 16 or 17 genes that have been discovered and, as most of you know, you just have to have two faults in any of those genes to have Bardet-Biedl Syndrome or LMBBS. Beth Hoskins, long time member of LMBBS, is the lead scientist for our genetic testing service and she has set up some very advanced new testing which has worked extremely well. We have tested 308 patients, out of 440 and have confirmed diagnosis in around 70%.

We can say with real conviction that BBS1 is the gene which is most broken/faulty in the UK, followed by BBS10 and then BBS2 and BBS12, however there are still a significant number of patients unconfirmed, which means there are still unknown BBS genes to find.

Surprisingly, there are around 7,000 known rare diseases and about 100 new ones are described every year. Around 1 in 17 people will be afflicted with a rare disease, that is 3½ million people in the UK, more than the number of people with diabetes. Thankfully, governments, particularly in Europe and North America, have now recognised this fact and are pushing funding in this direction. Pharmaceutical companies are intensifying a lot of their research into rare diseases; 250 drugs have now been approved by the Food and Drug Administration (FDA) in the US, for more than 200 rare diseases.

So how do we approach the treatment of a rare disease? If we know which gene is involved and the type of mutation, we can start to make some suppositions or guesses as to what might be the best type of treatment. We can then drive forward and think about funding and so forth. There are some critical pieces of information that you have to have in place to be able to go forward with treatment, and as we already know the genes for most of BBS, we are in a good place. We know also that there is a pre-symptomatic window, which means there is a period of no symptoms. We know that most BBS children are born with normal vision and this period of time, before there is retinal damage, is very valuable to us. Looking ahead to potential treatments, which approaches are applicable to BBS? There are many, but I'm going to discuss three, Gene therapy, Genetic therapy and Drug therapy.

Gene Therapy: The idea is simple, you've got a broken gene, let's replace it with one that isn't broken. The actual execution is very difficult and involves a lot of people. The gene has to be of a very high quality and pass all sorts of regulations. The surgeons have to be very adept at injecting the gene into the back of the eye, where the photoreceptors sit, because it is the photoreceptors that become damaged over time in BBS and these are the key cells that receive the light. If we can get a normal gene back into those particular cells to make the correct protein, we may be able to restore an element of vision.

A navigation demonstration of the first person to receive gene therapy shows how effective it can be. The experiment involved directing the patient into a maze and recording his

progress. Pre gene therapy, the patient made 8 navigation errors and took 77 seconds to complete the course. Six months post gene therapy and it took 14 seconds to complete the course with no navigation errors. We do have to be circumspect about these kinds of studies because they are still a work in progress and it's not necessarily amenable to absolutely everyone, however it is something that we feel we need to explore. Some of our colleagues in the States have demonstrated effective gene therapy in BBS mice and we are looking at pre-clinical studies with the possibility of developing gene therapy for the common BBS1.

Genetic therapy: this is not the same as gene therapy, genetic therapy is drug based. Genes make proteins and they are just a linear code, a part of the DNA. They have a particular message or a code that says, 'start the protein here' and they all have a stop codon, a signal that says, 'stop here because you don't want to make a protein that is too long'. So that's what we all have, stop mutations in a particular place, in all of our genes, so that we can produce the right type of protein. We know that roughly 40% of folks who are coming to clinic have at least one of these stop mutations, or 'faults' in the wrong place, which has the effect of doing two things: either you get a short protein or you get no protein at all. Either way, it is a problem and you end up having the difficulties associated with BBS.

Ataluren (it used to be called PTC124) is a medication that has been developed to find those premature stop signals and ignore them; no-one really knows exactly how it works, it is what is called 'read-through therapy' and it reads through to hopefully produce a normal protein. Ataluren has been trialled now in two well-known conditions, Duchenne Muscular Dystrophy and Cystic Fibrosis; the results show promise. It's important that we should go forward and set up a trial of this particular compound, in a group of patients who have nonsense or stop mutations in the wrong place.

Drug Therapy: Our question is, 'are there ready approved medications that might be valuable for folks with BBS?' Most drug therapies will not cure a particular condition, however they may alleviate it in some particular way and that is what we are proposing: to slow down deterioration in vision perhaps or try to prevent kidney problems or weight gain. We have decided to take two approaches. The first is to base the type of drug we want to test on present knowledge of the disease mechanisms. The second approach is a blind screen, in other words, test thousands of drugs, which are already FDA approved, to see whether any of them might have some benefit and 're-purpose' them. It takes a lot of money and at least 15 years for any promising drug to make it to the market, which is why we have decided to focus on existing approved drugs. Thalidomide is a classic example of that. We all know the problems of Thalidomide which was used for sickness during early pregnancy, but now it works very well in the treatment of leprosy. Our research to date has shown that there are drugs that are looking promising, promising at least in the BBS zebra fish, so you can see, we still have a way to go.

ADDENDUM:

National clinic service: Since the conference, we have met with senior commissioners for highly specialised service in NHS England. The really great news is that the uncertainty around the continuation of the BBS service has been resolved. We do not have to go through a renewal process next year as planned, the service will be reviewed and monitored episodically. This is really good news for the third sector i.e. LMBBS Society as their role is retained.

Transition: growing up with a health condition

Marie McGee

Transition Co-ordinator,
BCH Paediatrics and QEH, Birmingham

Marie McGee is the Transition Coordinator at Birmingham Children's Hospital and was at the Conference to tell us about her role and the process of transition. Her background is in education, working in Birmingham inner city schools for the last 24 years. Marie has worked with a variety of social and community groups, particularly with young people with special needs, and is now taking that vast experience into the world of health; she has been working with the BBS team for about six months.

Our young people start working on what we call their transition skills when they get to about age eleven to twelve. This is when they move from primary to secondary school, so transition effectively means change and coping with change. It is about them developing their own identity, managing the changes that come with that, achieving their own independence, developing their confidence and self-esteem and being ready vocationally, whether they are going to go on to college, university or going to do an apprenticeship.

We have all been teenagers, growing up in our own way, maybe as Bay City Rollers fans, ex punks or Goths! We have all gone through different changes, and for the young people that we see from age eleven onwards, it is no different. They have all got the same types of problems in different ways, which is why we work with them very much as an individual. Our youngsters with long term health conditions can suffer from a number of barriers, some of which leave them feeling quite isolated, particularly from their peer group. This makes it difficult for them to manage relationships, so some of my work and the Psychologists' work dovetail into each other.

Quite often our youngsters have limited opportunities for what they want to go on and do, and I'm a great believer in, if we see a barrier, how do we get under it, over it, around it and not be defeated by it? That is easy for me to say, because you are living those barriers every day with your children and young adults. However, it is how we can give you the information to support your young people to get past those barriers that can make a difference.

I also work for another team and we had a young lad in clinic who wasn't doing any housework at all because, he says, "I've got arthritis. I don't have to do it. My mum and dad don't think that I need to do it." He was quite well, so I suggested he got involved with a household chores rota. He looked at me in horror and said, "You're supposed to help me, not get me involved in stuff like this." But actually, I'm there to ensure that he has a full life and that includes housework and all the other things that come with it. The young person's identity can end up revolving around the condition, as opposed to the young person living their life and the condition being a part of it. It doesn't have to be the ultimate and the only thing in their lives.

The definition of transition in a health setting is 'developing our young person's skills and knowledge to support their long term health condition'. It is about managing all the different changes that come up, and that includes the transfer over to adult services. A lot of people think that transition is transfer, but it's not. Transition is a whole process and transfer is part of that. We need to work with professionals to make them understand this, as well as making plans with the young person and their families. The aim is that the young people will hopefully have uninterrupted health care that is coordinated.

We want to promote that our young people can be more confident, have more independence and can start to take some control where they can. We are looking at maximising lifelong function and developing our young person's best potential. We are all involved with transitioning our young people; the young person is at the centre and we have a huge variety of services around them as well. I am developing some transition plans that you can share with your local professionals, because we need those people on board and to be having these conversations and educating our young people in the same way. Once the plans are finalised, they will be used in clinic and will also go on the LMBBS website.

The plans are set up for around ages 11-13, 13-15, and then 16-18. We've called them 'Set up', 'Get up' and 'Go'. The plans are a way of assessing a range of skills and knowledge that the young people may or may not have. These plans are just the tip of the iceberg, but they allow us to start conversations that maybe nobody has ever had, or the young person has never thought of. The young person can do the self-assessment themselves. The good thing about the plans is they trigger things for the young people, but this may set off a whole chain of thought patterns that families aren't prepared for, which is why I think it is important to share the questions with families and let them see the sort of things we are looking at.

The plans are to help and support the young person as they prepare to move to being adults. I'm working to put something together where the young person writes their own referral to the adult team and that will accompany all the information that is sent by the health professionals, so when the young person goes to an adult clinic, the professionals will have a little insight into who this person is, as well as all their medical information. It gives the young person the tool to be able to set the scene, explain who they are, what they are about, what their dreams, goals and aspirations are, what they have got confidence in and what they are able to do.

For young people who have learning difficulties, we have developed the plan 'My Life, My Future.' This is an overview of their life which we can start to build on and find out where they need additional support. One of the sections is about asking questions. When the young person is with the Consultant, they don't always have the ability to ask questions, but they could prepare something in advance. They could bring a little list or a diary, or an audio file where they have recorded questions beforehand. It may be that they have never been prepared to do that before because it is assumed that mum and dad will do all the talking.

Helping our young people to be as independent as possible is a huge part of the transition process. Just getting some young people to clinic is hard enough, as is trying to have conversations with them that they don't want to have. It may be that if they think about it and

talk about it, they have got to start to accept it. Then, if they start to accept it, what's going to happen? What will the future look like? It is almost easier for them not to have those conversations. The Psychologists can work with us very closely, and use different strategies, such as music therapy. Certainly with some of our young men, talking is just not going to happen. You sit them down to talk, or you want to ask them something, or you want to tell them something and just the word 'talk' seems to terrify some guys! However, if you were doing an activity and you just bring it up inadvertently, it works far better, and that's how we hope to go about trying to impart information to our young people, or get some information out of them.

For me, the parents and carers are the most important part of the jigsaw in terms of supporting the young people, because we can't do it without you. You need to have confidence in us as professionals and we need to be able to give you the right information so you can go off and ask further questions of your local support services. Being part of that transition process means helping to let go of your youngsters, allowing them to make mistakes so they can evolve and develop. Over protection from the family, is something which we all quite naturally do, however it is very important for our young people to start developing, at least in clinic settings.

One parent said, "I think it's hard for us as parents to stand back and say 'well I'm not going in' because we really want to know what's going on. You know they're not going to say everything you want them to say."

The ideal clinic situation is where a young person goes in and sees the doctor or a nurse, for a period of about five to ten minutes, on their own so they can talk about anything. They may not even talk about their health. They might talk about what they did at school that day, but it is giving them the confidence and the independence to start being able to talk to the professionals and express themselves in their own way. Then the parents go in, and the young person recaps on what has been said, maybe with the doctor's prompt, and that way we can check the young person's understanding and their memory. We would also explain to the young person what confidentiality, consent and duty of care means, and explain that to the parents too, because there may be conversations that young people want to have with doctors that will stay between them, because they have that right. Of course, those conversations and what needs to be shared back to parents can become a minefield and we would need to address that individually.

So transition is about many changes, and those changes come in different guises: health, home life or vocational challenges. It is so important that we all work together. A young person said to me that his family is the ice cream and the medical team were the ice cream cone, which is lovely. I think what he was trying to say was if you get lots of support with your condition it will make an awful lot of things a bit easier and a bit better to manage. What I do, and other people like me, we are the sprinkles on top of the ice cream and if you get some sprinkles on your ice cream it makes it a lot better.

Uncovering the Emotional Needs of the BBS Population: observations and interventions

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 the psychological aspects of living with Bardet-Biedl Syndrome:

"My role involves doing a brief assessment of people's needs and then looking at the impacts of the disorder, possible levels of depression and how people are coping. Because it is quite a new service, a lot of my work has been about trying to identify some common themes, in order to get some sort of sense of what people with BBS are really dealing with on a psychological level. Psychologists teach people coping skills and techniques for how to handle difficult emotions and accept their condition. We know that if people are able to come to terms with their physical disability, they become more positive and their life becomes a lot easier to live.

I think most people who have BBS would agree that physical health and mental health are strongly associated. To date, there isn't any research about the emotional impact of BBS, but just from hearing your stories and seeing the work that other people have done over the years, I think it is very obvious that there are mental health difficulties associated with BBS, particularly the slight over-emotional presentation, as there would be with anyone trying to cope with a physical disability. However, everyone's ability to deal with stressful events differs and having BBS does not necessarily mean that the ability to cope will be different from the general population.

Based on the years of experience that Professor Phil Beales and his colleagues have of meeting with you all, I find that emotional immaturity seems to be a recurring theme, as is poor reasoning, psychological inflexibility, obsessional thinking and rigid routines, which can be signs of autistic presentation. Roughly 25-30% of the adults seen in clinic over the last year have had referrals. The majority have been referred for Cognitive Behaviour Therapy (CBT) but there have also been a couple who have been referred to Occupational Therapy, Social Workers and organisations/charities that offer supportive services such as disabled day centres.

At Guy's, I am looking into the emotional impact of the syndrome, particularly at adjustment issues that come with the emotional response following diagnosis. This is actually a very normal part of receiving any kind of bad news or dealing with anything that is difficult. Adjustment disorders should typically pass after a couple of months, but sometimes they don't and this can lead to depression, anxiety, panic attacks, anger and poor emotional control. Typical signs of depression are low mood, not feeling like doing anything, and withdrawing and isolating from other people. Someone who has panic attacks might think that their anxiety symptoms (typically palpitations, chest pains and shortness of breath) are

part of something even worse, for example a heart attack. Some people who have had an accident as a result of their impairment, might find it scary to enter a particular environment, but these fears can be treated very efficiently with therapy. There has not been enough research yet to know how much of the emotional problems are part of the syndrome and how much is due to the impact of it, but I'm fairly sure that it's a bit of both.

From the time of diagnosis, or even pre-diagnosis, to 7 years of age, typical themes that we see in clinic include obsessional thinking, rigid routines, lack of communication, challenging behaviour, inability to form friendships, problems in school or nursery, feeding issues and obesity, all of which cause a lot of worry and emotional issues for family members and of course also for the patient. Between the ages of 7 to 12, there is typically an increasing awareness of the disability and the feeling of being different is starting to emerge, particularly in the school setting. At this sensitive age, youngsters may have difficulty with friendships, obesity and bullying, resulting in their self-esteem being affected. Fears, such as the daily management of any behaviour difficulties, education, living an independent life and having a relationship, start to set in for both the person affected by the syndrome and their family. Fitting in socially is very important and the teenage years are when relationships and sexual functioning are major issues. Emotional detachment is common for any teenager, so suddenly not wanting to share everything or discuss things openly anymore can be difficult for parents who want to help. As with the general population, hormonal changes can lead to developing mental health problems, with people feeling lonely and unhappy.

In adulthood, common presentations are: worrying about medical conditions, not feeling in control of emotions, apathy and social skills problems, such as lack of a social life and finding a job. Some people don't seem to show emotions at all; with the more autistic presentations there can be a lack of understanding for other people's emotions and an inability to read social cues, which makes it harder to relate to other people. Independence is an issue for some, but not for others. Some people are able to do anything they want to do, whereas others find themselves understandably frustrated about the limitations that come with BBS.

When we look at intervention, we look at the factors that determine how well people cope. Our backgrounds make a big difference, as does an understanding of the issues we are dealing with. Some people are extremely well read-up on what BBS is all about and what kind of things you can expect from it and that often makes it a little bit easier to cope with. If you are new to the condition, you have the LMBB Society to support you and I think that makes such a difference for us in clinic as well, because it acts like a bridge between clinicians and the patient group.

Possible interventions for children would involve assistance with school integration, behaviour therapies, speech therapy, family support, management of symptoms and sensory and emotional support. It is important to get the associated learning difficulties diagnosed, which could then lead to help from other areas. I'm thinking particularly about autism now. We see signs and traits of autism in such a large number of people and if they get help from an early age, it could really change their future.

It is really important that parents receive support too. If a parent is depressed and anxious about things, they are not going to be able to offer adequate support to help their son or daughter. We understand that taking care of your child is paramount in your life but people often seem to think that they should get on with things and try to cope on their own. Actually, it is better to look for help early on, before problems get out of hand. There are lots of helpful services out there, especially Cognitive Behavioural Therapy, that can be accessed through local GPs and local support centres. Don't be scared to ask for help when you recognise the signs, such as fluctuating or low moods that go on and on, and hold people back from living a full life. Although parents always mean well, some of the things they do can actually hold children back from developing independence, so parents may need help in identifying how they can give the best support to their child.

Cognitive Behaviour Therapy is one of my favourite topics! Traditionally, CBT has been used to treat things like depression and anxiety, but these days, CBT can be found in almost every setting in a hospital and I find it is a very useful framework to use in order to try to understand and treat mental health difficulties. It looks at thoughts, feelings, behaviours and physical reactions and how these interact and often form negative vicious circles. For example, someone who worries about socialising will find speaking to people an anxiety-provoking situation. On a physical level that means they might be sweating and have an increased heart rate. If they decide to not speak at all, they will never find out whether saying something would have been fine, and the vicious circle of thinking they can't converse with people will just continue and often generalise to other situations as well.

There are things in life that we cannot control, like having BBS. In CBT we focus on the things that we **can** control by making changes to the way we feel by working on thoughts and behaviours. Worrying and focusing on negative events can make us less able to cope, so we look at distraction techniques and other techniques that can assist in breaking the vicious circle and improve peoples' mood. We look at things like eating patterns because we know that obesity is going to lead to negative thinking, low self esteem and negative moods. Using activity charts for scheduling activities and having a routine can help people feel better and this leads to them to feel more accomplished and less depressed. If we can't get the actual patient on board we might be able to influence people around that patient to contribute to changes in the behaviour.

CBT will assist you in identifying and modifying negative behaviours in order to minimise the impact that it has on your emotions. It tries to strip away 'avoidance behaviours' – things people do to try to avoid emotionally painful experiences. Examples are alcohol, isolation, avoiding certain places or people, or trying to avoid one's own thoughts and feelings. Avoidance behaviours are a very natural way of responding to stressful events, but are actually very counter-productive. So let's say, for example, standing up to do my presentation, I felt so nervous that I just wanted to run away! Running away would be an avoidance behaviour and it wouldn't serve the purpose because I would come away thinking "I never delivered my speech!" – 'I cannot cope with public speaking'. So when you start taking away avoidance behaviours, people are forced into situations where they realise that they can do things differently and then that has a nice knock-on effect on their mood and gradually also on their confidence in being able to cope in life.

CBT can also encourage you to make changes to the way you feel through physical activity. I cannot emphasise enough how important exercise is for mental functioning, and this is not just true for those with BBS. It has so many positive effects, like endorphin release and learning how to better tolerate uncomfortable mood states. Some of you might not be able to do a lot, but try to find something that is at least getting you outside the house, even just a little walk. Trampolining has proven to be fun and not too strenuous and is suitable for people of all ages.

I just wanted to say that I had no expectations when I came into the BBS clinics. I ended up here very much randomly but I decided to stay. I often think that when people are faced with adversity it makes them get their priorities right and grow on a personal level and that is certainly obvious in this population. There is a fighting spirit amongst you and I think that's wonderful. If you have any kind of problems, you are always welcome to contact me for advice on how to proceed. If you know of any support groups or initiatives in your local area, please let me know so I can pass the information on to other patients."

Finally, here's a lovely quote that I thought represents a lot of what I see in you guys –

'The most beautiful people we have known are those who have known defeat, known suffering, known struggle, known loss and have found their way out of the depths. These persons have an appreciation, a sensitivity and an understanding of life that fills them with compassion, gentleness and a deep loving concern. Beautiful people do not just happen'.
(Elizabeth Kibler Ross)'

Achieving International Cricket

Hannah Young

"I'm really into sports and when I found out I had LMBBS, I didn't think playing sports would be possible. However, a whole new world was about to open up for me. I went along to support a friend's son at a cricket game and my husband, Sean, saw a poster on the wall about Blind Cricket; he asked me would I like to give it a try and I said yes. I gave the number a call and spoke to a chap called John Garbett. He invited me along to a development weekend in Leicester and I was asked to play for the Berkshire Stags. I'd never played cricket before and it was the first time I'd met any of the team. They made me feel very welcome.

I went off to play the first game and after a few pointers from my team mates, I went in to field first. I was right by the bat, with balls whizzing over my head. It was a bit scary and I was a bit nervous but I loved it. To my delight, in my first over, I got a wicket. I was very proud, especially when all my team mates came over to celebrate. Then it was my turn to bat. We only needed a couple of runs to win the game. The opposition crowded me but I just hit the ball over their heads and we won the competition; to my delight I won the best new player award.

After that I went to train with the Berkshire Stags and soon after, I was asked to go and train with an international ladies team. I had a try out and was then asked to trial for the men's team, however I needed a bit more practice fielding. I decided I wanted to play for the girls anyway, because girls are best! At the end of April, the new season begins and we play our first game against Somerset. Unfortunately, they are a very good side so I'm going to be very nervous."

Hannah, with assistance from her husband Sean, showed the delegates the different equipment used in Blind Cricket. There are two different sized balls, a size three football, used in county cricket and a normal sized ball used in international cricket. The balls contain ball bearings so the players can hear their location. Players are classified according to their vision, from B4 (partially sighted) to B1 (Blind). If the person in bat is classified as B1, the ball has to bounce twice in front of them, otherwise it just bounces once. In international cricket, the ball has to drop halfway along the pitch and then roll along the ground so the person in bat can hear it coming.

Hannah explained that the person in bat, along with all those fielding close to the bat, have to wear a helmet. Hannah said, "You do get hit on the head but after a second or two it doesn't hurt really!" The bat is just a normal cricket bat, however the wickets are larger and in field, B1 players are allowed to let the ball bounce once before they catch it. B1 players in bat have a runner, however B2s and up have to run. B1s get double the score so if you hit a two, you get a four, and so on. Aside from that, the rules are the same as standard cricket.

Hannah brought her presentation to an end, but had information for those interested in trying Blind Cricket. She said, "You can contact John Garbett and he'll be able to tell you if there are any teams in your area. In September there is a development weekend in Leicester and anyone can come along, you don't have to be able to play cricket, you just turn up, join a team and give it a go." John's email address is garbett.j@sky.com.

Dietary Update: Three Years in BBS Clinics

Sarah Flack

Principal Dietician, Great Ormond Street Hospital

As you know the BBS clinics started three years ago, with the first clinic held at Great Ormond Street Hospital in April 2010. I'd like to share with you our findings from these 18 clinics.

I have been looking at information you would expect a dietitian to - weight, height and body mass index (BMI). If you come to the London clinic you will have seen and hopefully completed an 'All About Me Diary'. This allows me to collect information about individuals' dietary intake and also their activity patterns. Why should we fill this in? After clinic, families get a tailored report which focuses on ways to either make their child's diet adequate, for example improving iron intake or ideas to help control their child's rate of weight gain. The completed diary each year helps highlight both positive changes in activity and dietary patterns.

So what is body mass index and why do we use it? BMI is a simple, easy to calculate measurement of a person's weight, compared to their height. It is calculated by dividing a person's weight in kilograms by their height in metres squared. (Weight (kg)/Height (m²)). It is used clinically because it provides a good 'rough' indication of the level of body fat a person has. The healthy adult BMI range is between 18.5 and 24.9 kg/m². Generally adults with a BMI of 25 or more can be classified as overweight. However, I am sure we all know of examples when BMI has been shown by the media as not being a reliable measure. Athletes, for example, may have a high BMI, because of their muscle bulk, not their body fat levels. BMI should also not be used for pregnant women, as there is more than one person involved!

So what about BMI in children? In children, BMI is used from two years of age, however there isn't a standard healthy BMI number range as in adults. When we look at BMI in children, we still compare weight with height, however we also need to consider their age and sex. This is because boys and girls have different growth patterns and body composition varies with age. Girls and boys have separate body mass index charts, which you may see when you attend your clinic appointment. (Girls have pink charts and boys have blue charts). On each chart there is a shaded area which represents the healthy BMI zone. The BMIs represented by the shaded area changes with age. For example let's look at a BMI of 20:

- This we know would be in the healthy BMI range for an adult.
- For a girl of 7 a BMI of 20 is outside the healthy shaded area. As this falls above the shaded area this girl would be classified as overweight.
- However if a girl of 12 had a BMI of 20 this falls within the healthy shaded area and so this BMI would be considered healthy.

I use the changes in an individual child's 'BMI score' to see how a child is getting on at their follow up appointments. In an overweight child, we are hoping for a reduction in their BMI score. The BMI score change is the only way to show positive changes in a child. This is because weight in young children should be increasing as they are growing and ideal BMIs vary with age.

So what results do we have? We have now seen 27 children at least twice in clinic. Of these children, 26 had weights above the healthy BMI zone at their first appointment. At their follow up appointment (12 to 18 months later) 16 children, more than 60%, had reduced their BMI score.

11 children reduced their BMI score by 0.25 or more. A recent study of adolescents attending a specialist obesity clinic in Bristol found this level of reduction leads to health improvements. These benefits include better blood pressure and improvements in blood fat levels.

Six children achieved a reduction in their BMI score of 0.5 or more. The same study found this resulted in additional health benefits, including a reduction in body fat levels, reduced waist size and improved blood fat levels.

Four children actually reduced their BMI score to within the healthy BMI zone (the shaded area on the chart) - which is fantastic!

After all those numbers and facts, it is time to move on to look at how some children and families have actually achieved these positive changes. I should point out that most of the following examples are boys. This isn't because it is only boys that have achieved these BMI score reductions, but because we have seen more boys for follow up appointments, than

girls; it does not mean if you have a daughter, that they won't be able to achieve the same outcomes!

Firstly child A is a boy who was seen initially when he was 4 ½ years old and then again when he was about 6 years old. His BMI score had fallen by 0.83 points which is a great achievement. The key dietary change was that he was 'eating better'. In clinic his parent mentioned that he now liked 'proper food' which meant major changes to his diet. His Mum found that he was much more co-operative and listened to 'No' which made managing his diet much easier. Really positively, his Mum felt this understanding of 'no' could be used with "diet rules" and she expected even better results in the future. So I hope for some of you this will reassure you that developmental age and understanding can make a huge impact with dietary control.

Next, Child B was initially seen when he was 5 ½ years old and he returned for his follow up appointment aged 7 years. His BMI reduction again was very impressive - it had fallen by 1.18 into the healthy BMI score zone (the shaded area on the BMI chart).

I bet you are thinking this must have involved lots of changes to achieve this? However the changes were actually quite small but have been successfully continued for the long term. The big dietary changes were small, the family offered lower calorie snack options and 'treats' were kept for weekends only.

The other major change this child made was to significantly increase the amount of exercise he did. He now regularly plays football, attends football training, goes swimming and at home, plays active computer games like a Wii Fit or Xbox Kinect.

Just to prove girls can do it too, child C was initially seen aged 3 and then again when she was 4. She has managed to reduce her BMI by 0.25 which is associated with the positive health changes mentioned before.

Her family had excellent portion controls in place. Her 'All about me diary' showed that she was eating the same calorie intake so her portions hadn't increased as she got older. She was also doing more exercise, which again shows how important keeping active is to help keep our weight gain in check. At her last clinic appointment she was active after school three days a week and on both weekend days - so active on at least five days a week.

All of the examples so far have been younger children, so child D has been included to show that older children can do it too! This is a 14 year old boy who was seen again when he was 15 ½ years old. Again he had achieved an impressive reduction of his BMI score of 0.46. This reduction was achieved by good portion control, cutting back calories from snacks and increasing exercise.

He had fantastic portion control - using the half plate of vegetables rule. Vegetables have a high water content and also a high fibre content, both of these help to fill you up for longer. So half of the plate is covered with vegetables, the remaining half is covered equally by protein foods (meat, fish, chicken etc) and starchy foods (potato, rice, bread etc).

His other dietary change was cutting back on crisps. Instead of crisps he was eating a piece of fruit, therefore reducing the amount of calories he ate each day. By cutting down to having crisps occasionally rather than trying to stop eating crisps altogether, he was able to maintain this change in the long term.

He again had increased the amount of exercise he did each week and was regularly attending the gym. Gym programmes for young people can be really motivating as progress is clear each time the programme is reviewed.

Three main themes stand out from these examples, healthy snack choices, watching portion sizes and the importance of activity! Let us look at these in a little more detail starting with healthy snack ideas.

The snack market is big business and in recent years, manufacturers have launched a number of healthier snack options. Look out for snack foods based on fruit, for example fruit bars and individual bags of dried fruits. It is worth looking fairly regularly when you shop because new products keep hitting the shelf. My children, for example, have recently discovered cola flavoured raisins - they like them because they think they are a little bit naughty! Flavoured popcorn is meant to be one of the hot new foods for 2013 and in some supermarkets you can find small individual bags containing just 50 kcals per bag. This is a great calorie saving compared to other savoury snacks, such as potato crisps, which have 100 kcals more per small bag.

Sometimes presentation of food can be key to getting our children to try new foods: children love things that look interesting, fruit kebabs for example, or you could try making caterpillars out of grapes on a stick, put the face on the front, and it suddenly looks more exciting than a plateful of grapes.

Portion control is vital to maintaining a good healthy weight. Our plate size has grown massively in the last 30 years. Today our plates are 30cm, that's a ruler size. Going back 30 years ago, to when I was growing up, our plate sizes were 23cm. Obviously, if you have a bigger plate you can fit more food onto the plate, so just by changing your plate size you can cut your calorie intake by about 20%. This is such an easy thing to do compared with trying to change your diet completely.

Pictures of food portions on different sized and different coloured plates, demonstrate the tricks our eyes can play on us. Each plate has exactly the same portion, however it feels far more satisfying to have a full, smaller plate rather than a half empty large plate. There is a lot of research going on at the moment about plate colour and whether that has any impact on how we eat. Some research says if you want children to eat more green vegetables, put their food on green plates.

A simple way of controlling our portions is to cover half the plate with vegetables, a quarter of the plate with protein, and a quarter of the plate with starch. Vegetables have a high water and fibre content, both of which help to fill us up, so overall you have a meal that is going to be more filling, which hopefully means later on in the day, you will feel less hungry and will have eaten fewer calories.

A big theme that is emerging from our kids in clinic is the importance of activity. For five to eighteen year olds they should be doing an hour of exercise each day, but it doesn't have to be an hour all at once. You don't have to say "Right now you're going to go and play football in the garden for an hour." Or "We are going to go swimming and we are staying in the pool until we have finished our hour." The exercise can be done in 10 or 15 minute blocks, which should be built into the day. When I walk to school with my kids, although we have been ready to go for ages, all of a sudden no one has any shoes and it is time to go! So we do a fast walk to school, which counts as exercise, however we tend to amble home, so that doesn't really count. When we get home, the kids will hopefully go out onto the trampoline in the back garden so that I can cook tea.

So it is using those little blocks, counting the bits where you have been most active, whatever it is. If you go by car to the shops, park at the furthest end of the car park to extend your walk; if you go by bus, maybe walk one extra bus stop, it is making little

changes to build some activity into your day that can make positive long-term changes. It is also worth trying out new sports and clubs to find something you really enjoy doing.

So this is all very positive, but are these changes long term? At the moment we only have data for two children, both boys, that have been seen three times. The first child has continued to reduce his BMI score, each clinic it has come down and down and he is in the healthy BMI zone now. The second child did really well at his follow-up appointment. His BMI score had reduced, however at his last appointment, his BMI score had increased and it was higher than when we first saw him. The child had changed schools and his activity levels had reduced massively. He hadn't changed anything diet wise, so his weight loss had all been to do with the activity he was doing: he was no longer playing football at school, going to football training or playing football at weekends. The young patient's attitude, however, was encouraging. He started thinking about ways he could change things as soon as he left the clinic, so hopefully when we see him again next year, his BMI score will be down again.

If you attend the Great Ormond Street BBS Clinics, I send out 'All about Me' diaries in advance. It would be really helpful if you could complete and return these as I am hoping to compile the information and provide advice on portion control and calories in relation to age. In the meantime, it is very easy to use the 'half plate vegetable rule', consider your plate size and get active.

Q. 'I completely agree with everything you're saying, but are there some children that just cannot lose weight? Our son has an underactive thyroid. He lives on low fat mousses. That's his diet 24 hours a day. There is no deviation.'

A. 'I have presented the best outcome and as you know BBS is a real spectrum. What works for one may not necessarily work for others and there are some children that are much more severely affected with their weight. We do have several older children coming to clinic that are eating very little, yet are struggling with their weight. We are doing minor tweaks to see if we can help, but until we see them again, we won't know how effective it has been.'

Memory Study: an update

Dr Torsten Baldeweg

The part of the brain most important for our memory is called the hippocampus and we know this little structure deep in the brain is important because the patients we see who have injury or epilepsy in this region tend to have severe memory problems. Studies on mouse models have shown that cilia may be important in the development of the hippocampus and because many people with Bardet-Biedl Syndrome, a known ciliopathy, complain

occasionally of memory or learning problems, myself and Kate Baker decided to look at this systematically.

Quite a few BBS patients volunteered to be scanned and assessed and we are very grateful for their contribution because we were able to do the first systematic study in the world that looks at the brains of young people with Bardet-Biedl Syndrome. One of the things we have found is that the hippocampus is a bit smaller in some people with BBS. Other areas also appear to be affected, the temporal lobe is also a bit smaller in some patients and that is also important for memory.

At last year's conference, we conducted a memory study that asked questions such as 'Who is the Prime Minister?' We had 38 volunteers with LMBBS and we also had 13 parents and siblings. What was reassuring was that the natural memory deficiency that occurs with age happened at the same rate in both the BBS and non-BBS population. Quite a number of people with BBS however, demonstrated a mild to moderate deficit in their memory scores, which we had suspected, based on what people were telling us. Pleasingly though, many BBS patients performed as well as the unaffected parents or siblings, or even better, which is great. Ultimately, we would like to understand why some people suffer more than others and at some point be able to help.

Cilia are important to make memories permanent, so that we may remember things from our childhood, from our school days and so on. Cilia probably don't support what we call short term memory and many BBS patients may have noticed that their short term memories can be very good, maybe as a way of compensating.

So, what can we do to improve our learning and our memories? I think the obvious answer is to do puzzles, learn new things and so on; challenge the memory. From a medical point of view, we could try to develop new drugs, in fact there is a whole industry out there trying to develop drugs for Alzheimer's Disease and other degenerative disorders, where memory loss is a key feature. We could transplant stem cells into people's brains, which is a bit radical, but has been effective in stroke victims and those with severe Parkinson's Disease, so this is clearly an option.

One of the most simple things to do, is to exercise our bodies, because studies show that physical exercise helps to grow new stem cells in the hippocampus. This is something that has been discovered in rats and mice and replicated in biological experiments many times; after running on a treadmill, we see a flurry of new born cells in the brains of rats and mice and it has now been discovered that the same thing happens in elderly people, in young people and in children. To begin with, improvements can be seen following just a small amount of exercise. In some of the early studies, getting elderly people to exercise at least once a week showed some benefit that could be demonstrated using brain scans and of course, the amount of exercise can be increased over time. We are now thinking that maybe we could develop some sort of exercise programme for young people with LMBBS.

Of course, it really is important that we exercise our brains as much as we can, by listening to music, learning an instrument, learning another language, doing maths puzzles and so on.

Workshop 1

Professor Philip Beales (Professor of Medical and Molecular Genetics, ICH)

Professor Tim Barrett (Honorary Consultant Paediatric Endocrinology and Diabetes, BCH)

Dr Shehla Mohammed (Consultant Clinical Geneticist, Guy's Hospital, London)

Dr Denise Williams (Consultant Clinical Geneticist, Birmingham)

Our thanks go to everyone who contributed to the question and answer workshop sessions during the afternoon of the conference, a selection of which are included below:

Delegate: What can you tell us about gene therapy?

Speaker: A ten year project looking at inserting genes into cells in the retina has sadly, in the last year or two, come to a grinding halt, effectively because there has been no progress. The team involved are now thinking about how they can do things differently in a way that might work this time. It has to be a very cautious process, because these treatments can go wrong as well as right.

Delegate: What about drug therapy?

Speaker: At the Ciliopathy Alliance Conference last May, it was really interesting to see that there are so many people researching this family of conditions called ciliopathy diseases which includes Bardet-Biedl Syndrome. It is reassuring because it may well be that someone will come up with a treatment for one of the other members of the ciliopathy family that will actually be helpful for Bardet-Biedl as well. I guess we are all hoping that someone is going to come up with a cocktail of medicines in the next few years that might have some benefit. The hope is to slow down the degeneration, or have some kind of holding solution until something better comes along.

Delegate: How much money is needed for funding and where do you propose to get that funding from?

Speaker: We are talking well beyond a million pounds. We usually have to turn to private charities for this and there are a few foundations around. This is partly why we have created the Ciliopathy Alliance because a bigger group will lead to more prompt action, more enthusiasm and better access to funding.

Delegate: How is BBS different from other ciliopathies?

Speaker: From a vision perspective, with all ciliopathies, it is not just the central vision, or peripheral vision that is affected, it is a little bit of everything. We have a microscope which takes a laser photograph of the eye (Optical Coherence Tomography or OCT). Using OCT we think we can separate a person with a ciliopathy from a person with retinitis pigmentosa. If you have BBS, usually you will also have reduction in your colour vision, and you will quite likely start having problems at night. And then year on year, very slowly, everything just changes a little bit and that happens differently in some other ciliopathies.

Delegate: So should we use the term retinitis pigmentosa in BBS?

Speaker: No. We should call it retinal dystrophy, which is progressive.

Delegate: Can we say anything about the preference for the rods over the cones?

Speaker: Children who have particular problems with reading the optician's chart early on, but who do not have night vision problems, their cones will be affected a little more than their rods. Children who are short sighted and have night blindness, but aren't so affected during the day, their rods have been more affected than their cones. We can see these changes using OCT.

Delegate: I always assumed BBS night blindness was almost always in the pattern of progression for BBS. Are you saying you have seen a few children who might not have that?

Speaker: For some children, their visual acuity, their ability to read the chart is affected well before the night vision issues start.

Delegate: When is it advisable to get my other children tested for BBS?

Speaker: I guess it really depends on how old the children are and how well they might be able to understand what the significance of that information is. We would be guided by you as parents but normally we suggest around the teenage years when they might be thinking about reproductive issues. We would be very happy to meet with the family, as would your local geneticist, to help explain the ins and outs of having a test. You can ask your GP to make a referral to the local genetics service.

Delegate: Is that advice free?

Speaker: Yes, regional services are funded to provide that advice which would be available throughout the country.

Delegate: What happens to our blood samples when they go to the lab in London to test for BBS?

Speaker: When we get the blood sample, we extract the DNA and do an initial test for the four common mutations that happen in the largest population of BBS patients. If one of those mutations is present, we write a report to Professor Beales who will then send the result to the patient; the process takes around four weeks. If further testing is required, or we have a backlog of patients, the process will take longer.

Delegate: Have you identified the characteristics of each gene so that you know what conditions you are going to have if you have BBS5 for example?

Speaker: We have been looking through our data with a fine-tooth comb and have found some subtle differences, however, it is too early to use this data in a predictive way. With

BBS1, it seems that the average age of onset for severe visual problems such as being registered blind, seems to be slightly later, however this might be more of a function of the actual type of mutation in the gene as opposed to which gene is actually involved.

Delegate: What other approaches can we take towards managing the condition that are more relevant to someone who has already lost their vision?

Speaker: The computer people and the assistive technology people are doing a huge amount in terms of the eyes for people with retinal dystrophy/retinal degeneration like BBS. Families, individuals and doctors need to enthuse and help those scientists, computer people and engineers by showing them and telling them about our individual needs and circumstances so that they can develop the technology that is going to help us in our everyday lives.

Delegate: What can we do to help our children control their condition?

Speaker: We recognised that there are lots of different problems connected to BBS, which is why Professor Beales set up the multi-disciplinary clinics; we believe that we can get children to a much healthier state by paying attention to all the little details. Once a year, we check the kidneys and the blood pressure and make sure protein levels are okay. We check the eyes and try and utilise all the modern visual aids that are available. We do a psychological assessment to identify emotional issues. We want to stop children getting diabetes, if at all possible, and keep them as healthy as we can by encouraging regular exercise and a healthy diet. We are also looking for any problems with feet which might impact on exercising. The clinics have only been in existence for three years, but already we can see the benefits of this multifaceted approach.

Workshop 2

Annika Lindberg, Clinical Psychologist

Annika held three question and answer workshop sessions throughout the afternoon, which were all well attended. Our thanks go to Annika for her excellent presentation and contribution throughout the weekend; a selection of discussion threads are printed below:

Delegate: Is there anything you can suggest to help with over emotional responses?

Annika: When things don't happen in line with our expectations it can become a big problem. At times you might be feeling over emotional, however at other times you might be wondering, 'why am I not feeling anything?' So it is not always consistent. It is common for people with BBS to have a plan for how something is going to happen in their mind and when things don't go to plan, it becomes a lack of control. A lot of negative emotions come when we, as human beings, don't feel in control of things, like our surroundings and our own emotional state.

If you think back over the past week or so, you will notice a pattern: when you are busy, you are less likely to experience emotion, because you are focusing your attention elsewhere. However, when left to your own devices, sitting on the couch at home, maybe alone, that is when you are focusing on your own thoughts. If those thoughts are negative, that can lead to a negative emotional state. Inactivity is a big problem.

Delegate: So what sort of things can we control?

Annika: We know we can't change having BBS and we know we can't change other people. The one thing we can change is the way that we respond to things and we can do this by changing how we relate to events and other people (attitude) and changing how we spend our time (behaviour).

For example, if I spill my mug of coffee over all my papers and my response to that is, 'these things happen', I'll get over it, have another cup and I will probably be okay. If instead I think, 'oh why can't I ever do anything right, I'm a total failure, everything I touch just crumbles', then obviously I'm going to get quite sad and upset and the whole day will probably feel a bit unpleasant. So you can understand how the way that we relate to things that happen to us on a day-to-day basis is going to make a huge difference to how we are feeling.

Delegate: What can we do to get over a fear of something?

Annika: The traditional way that a psychologist treats fear is by exposure. Start by breaking the task up. Think about what will be the first little step that you could take and follow that with baby steps. When you can actually complete a task, that's going to add to your confidence and the next time you face a situation like that, you're not going to be quite as scared. Using this step-by-step process, you can begin to build layers of your confidence and ultimately, you will be able to achieve the thing that you set out to do.

One thing that is within everyone's control is the power of trying. It is important to keep an open mind and try different things but make sure you have plenty of support.

Delegate: What can I be doing to help my child with socially inappropriate behaviours?

Annika: Looking at these kinds of behaviours is hard. Try to correct them to the extent that you can, like explaining when something is not going to go down well in a situation. However, there are certain behaviours with BBS that we cannot do anything about and I think the best approach to those is to try to accept that that is how your son/daughter behaves in certain situations. I think ultimately you're bringing more stress on yourself if you're trying to make your child fit in to every single situation and you know it's never going to happen.

Delegate: I think every parent wants their child to be socially-adept and socially-accepted.

Annika: In childhood, you can change a lot of behaviours using modification therapies, because the brain is still growing. Later on in life, when these behaviours are engrained, it's

going to be a lot harder to change them. If you are finding that you are literally hitting your head against a brick wall and no matter what you do it isn't going to change, then for your own sanity, you're going to be better off trying to learn ways to come to accept it. You need to manage your anxieties by getting as much support as you possibly can for yourself, so that you in turn can support your child.

Empathy is something that is hard to teach. You cannot make someone feel something that they are not feeling, but you can teach consequences to behaviour, like giving rewards.

Delegate: Is there a link between BBS and Autism?

Annika: It's early days with the whole Autism/BBS link but I don't doubt that it's there. There are a lot of therapies available for Autism so if you can get a dual diagnosis (see your GP about this) you would be able to access this help. Some of the Autism therapies are extremely proactive in helping children modify their behaviours. There is a book I'd like to recommend called 'No Fighting, No Biting' by a Danish Psychologist, Else Holmelund Minarik. This book gives little anecdotes of difficult situations for children with disabilities like Autism and it provides a variety of ways in which you can intervene. What really stands out is that you cannot assume that you understand these children, because how they think has nothing to do with how we are thinking.

Delegate: How do you talk to your child about BBS?

Annika: Not all children are going to be able to conceptualise and I don't think there is a right or wrong answer: Is it better for them to know? Is it better for them to be in blissful ignorance? I think that comes down to each individual case and also to each individual parent: what do you feel comfortable with? What do you want them to know? I think the beautiful thing with children is that they will quickly adjust to almost anything, whereas adults take a lot longer. If you feel comfortable waiting, I think you can leave it until your child starts to ask about it, or when you notice that they're ready for hearing little bits and pieces then that might be the right time.

Delegate: How can you treat compulsive tendencies?

Annika: When we treat Obsessive Compulsive Disorder (OCD), part of the treatment is to expose the person to the things that trigger the obsessions. After a while, you teach the person how to better respond to it. The individual also needs to understand that both intrusive and obsessional thoughts can be ignored, you don't necessarily have to act on them. A lot of patients who have BBS are not necessarily finding OCD stressful, it is the people around them who are finding it a worry.

Routine is a difficult area to approach. We know that the sameness of a routine means security, however sticking to some routines isn't necessarily going to help individuals cope with the outside world which is always going to be changing. We know that if you let go of the routine completely, it's probably going to be a disaster. What you can do, to increase psychological flexibility a little bit, is to introduce new things, but give your child some way to

prepare for it. For example, if you are about to try a different route to school, tell your child beforehand, so they have a bit of time to prepare themselves mentally. I think we can all relate to that, even if we don't have BBS. If someone takes our routine away from us we are going to be struggling, and that struggle will go on for some time until we adjust to it and learn the new habits. In that respect, people with BBS are not dissimilar, it is just that their routines are far more rigid than ours.

Delegate: Could you give us some examples of helpful and unhelpful techniques for dealing with things?

Annika: I have seen parents who, in their worry for their children, have become so overprotective that they are not letting their children or young adults experience things on their own. They fear their child will fail at it or not be able to do it safely. I must say that from the patients I have seen, the ones who are happiest and most well adjusted are the ones who can do most things for themselves. There is a definite correlation there and that does not mean that those are the ones who are least affected by BBS, it just means they have learnt or perhaps been forced to learn to deal with it.

Paralympic Experience

Shane Ryan

My name is Shane, I'm from Limerick in Ireland and I am going to give a quick overview of how I overcame my sight loss with my sporting achievement. Going back to my earlier life, I went to normal mainstream school through primary and secondary, and I played mainstream sports up until around 17-18 years of age. As my sight deteriorated, I had to stop playing those sports because I wasn't getting any enjoyment out of it; I had to find alternative sports. I tried a bit of blind soccer which never really worked out for me as it was just too far away; it was up in Dublin, which was a long trek up and down. I received a letter about adaptive rowing and we decided to go and have a look at it, and I suppose, as the phrase goes, I took to it like a duck to water.

The support network was fantastic, headed up by a man called Joe Cunningham. He is still involved and he was our team manager of the Irish crew. So I started rowing in 2007 and since then it has taken on a journey of its own really. At the start, all my training was on the rowing machine with a bit of practice on the water at the weekends, but then as I learnt the technical aspect of it, we were on the water a lot more; I prefer to be on the water than on the rowing machine. I competed in several different events, individually and as part of a crew. Individually I went to a couple of indoor competitions which are done on the rowing machine. You set the distance and it is a time trial against yourself and all the other guys that are competing, so it is quite a good way of finding out where you are physically and mentally.

The Irish team was set up in early 2009, and at the time there were only four people involved, two managers and a cox. I got into that fairly quickly, however as we were coming

towards qualification for London, we started to get a lot more interest, so I really had to keep up the training and fight off the competition for my position in the team. To get to London 2012, we had to qualify the year before in 2011, in Slovenia. In order to qualify we had to come within the top eight countries, finishing fifth gave us automatic qualification.

The Paralympics is a fantastic organisation. There are some great people involved and they really know what they're at. There are dieticians, psychologists, a leading team doctor that literally is on the go 24/7. He knows everything about every athlete on the squad. We had physios as well, to keep us injury-free. Pre-camp was in Portugal, which enabled us first to get away and also to get a good vibe running through the camp. We got to know everyone from the other sports individually, as we were encouraged to spend meal times with different people. After Portugal, we flew to London and went down to the rowing venue at Eton Dorney, where we stayed until after our event.

We had three races in three days. Only the team which comes first in the heat goes straight to the final, with the remaining teams racing again. We had to go to the B final where we came fifth, which meant tenth out of twelve countries, so that was kind of a unique experience. The noise level was quite unique as well. Literally the whole race was so noisy.

In the boat we have got different athletes with their own disabilities. Our boat is known as a LTA4+ (leg, trunk and arm) which means there are four people using one oar each and a cox. The cox steers the boat and gives motivational calls. We had two visually impaired athletes, all the visually impaired athletes are blindfolded to make it a level playing field, so we had to get used to that and it takes a long time to get the rhythm together. Of the other two people on the team, one guy was missing 3 fingers on his left hand, and another girl had MS and Addison's Disease.

Questions:

Q: Did you go to the opening ceremony as well? What was that like?

A: It was electric. As our country walked in, they said 'Here's Team Ireland' and we got the biggest cheer, so that kind of typifies the support that was out there for us. However, we were competing two days later, so we had to leave fairly quickly and return to camp to get into race mode.

Q: *Where would you say you are in your rowing career? Have you peaked or will you have the chance to go to Rio? If you do, do you have right of passage or do you have to make your place on the team again?*

A: I'm only 25 this year, so my rowing career can go on for another couple of years if I can keep my place in the boat. Every year you have to go through a set of national trials with your country and it is open to anybody that has any kind of disability that's classified within the range of the boat, so we have to go through a set of trials and whichever crew is the fastest, that is the one that is going to be there for the year.

Q: How did you find the training for it? Did you find that you could control your weight while you were training and still keep enough energy?

A: I always had the weight issue and the training has brought my weight down massively. Since I took up the sport, I have lost about two stone in body fat and replaced it with muscle. The more lean muscle you have, the better you are as a high performance athlete. My training involves getting up at around 6am to be at the gym for 7am. I do a weight session on Monday, Wednesday and Friday; you never do them back to back, just to let the body recover. So a full weights session takes about an hour and a half and then I go and get some food. As I wasn't studying or anything at the time, I was able to go back to sleep. They say sleep is the best recovery that you can have, so I suppose I was having power naps through the day. I get up and go training again, either on a rowing machine or a bike. Most of the week would be endurance type training, long sessions of around 8K to 10K. Over a week I would probably do up to about 24K to 30K. Following that, if we are lucky and there is enough light around, we would be on the water in the evenings. So that would be either two or three exercise sessions a day with one day off a week to rest.

Q: Shane, do you think that you suffered post-Olympic blues, and if you did, how did you get over that?

A: I suppose I did really. We finished our races quite early in the programme, so we were able to go up to the Olympic Village and really get into party mode. We supported all the other Irish athletes that were competing at the time and when we came home, there was the big homecoming into Dublin Airport. When I got home, I went to bed and slept for about a day and a half. I took some time off and then got straight back into training.

Delegates Comments

"I would like to say a big 'thank you' to everyone who made me feel so welcome at my first conference. I met some lovely people who will stay in my life and heart forever. Really looking forward to next year. Much love and respect to you all."

"It was an awful lot of information to take in all in one morning."

"Hello to my good friends and new friends of which I was happy to meet and greet. My sincere thanks to the LMBBS team of whom put an awful lot of hard work and effort in to making our weekend such a fantastic event. Thanks again for such a wonderful time. With good and kind wishes to you, all take care."

"It was a very good conference. I found the psychology part the most interesting..."

"Thanks everyone for making the conference such a great time. It was really good to see old friends and make new ones."

“Phil Beales’ talk on the condition was good. I haven’t been at conference for a while, so a summary of the ‘now known medical facts’ would be good.”

“Hotel and food was absolutely fantastic, could not be faulted.”

“Too long with not enough breaks!”

“I can't wait a whole year to see everyone again! It was such a wonderfully, positive, fantastic experience and we have made some great new friends and feel very lucky.”

“We had an amazing weekend, it really is a well organised event where all involved work so hard to make it special. A big ‘thank you’ to you all. It was so nice to meet up with some truly amazing people who are such an inspiration! Big love to you all!”

I hope you have enjoyed this conference report, don’t forget, all of the contact details can be found at the beginning.

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