Editorial

Welcome to the LMBBS Conference Report for 2006. Another fantastic conference, with numbers up to 112 delegates attending on Saturday an increase of 28 on 2005, yet again we had an excellent line up of speakers, giving us a wealth of new information. Thank you to all those concerned for the organisation of this brilliant event. This report contains an in-depth account of the talks and personal perspectives, and should prove invaluable to those who were unable to attend the conference, as well as a useful reminder to those who came.

For the first time, we employed the services of Mark Pope, Photographer, from Newport, South Wales to video the event, and DVD′s will be available to purchase, details can be found on our home page.

Once again the children/young adults visited Drayton Manor Park, the sun was shining, even if it was a little chilly, but with their fantastic volunteer carers, a great time was had by one and all. This year′s cub reporters were Hollie Sales & Chloe Maclean, and their report is on the 'Young Members′ page.

There were many activities spread over the weekend, Friday evening John Donnelly gave us a demonstration of the 'ultra cane′, Steve and Graham hosted a Q & A on the syndrome and also showed off their IT skills. For the children, the carers entertained them with crafts, and on Saturday evening a movie show, with the highlight of the weekend, a fashion show from Hollie Sales, Chloe Maclean and Alex Clark. This was a repeat of a fundraising event in 2005 which raised £395.00 for the Society. We also had the now famous family raffle, with many prizes. As always each delegate was provided with a feedback form, with a free raffle ticket for each completed form. To end the evening Dennis Clark hosted a Family Quiz, which proved to be a great success.

On the last page of the report you will find a selection of the comments received and I hope that these will inspire those of you who have never been to a family conference to make 2007 your first. The date for your diaries is 20th21st22nd April 2007 I look forward to seeing you there.

The LMBBS Family Conference 2005 began with a warm welcome from the Society's chairman, Phil Humphreys. Phil took pleasure in announcing that in October 2005; Dr Beales had been called to the 'Chair' and is now Professor Beales. Congratulations and applause was received from the hall. Phil extended his welcome to families new and old, particularly to Kathryn Murphy who travelled from Brisbane, Australia and to the Shrestha family from Geneva. The proceedings of the day were then handed over to Professor Beales

Welcome and Introduction to the day from:

Professor Philip Beales, Consultant Clinical Geneticist, Guys Hospital/Institute Child Health, Gt. Ormond Street Hospital, London & President of the Laurence-Moon-Bardet-Biedl Society

Good morning and welcome! I think this year; Chris has surpassed herself in terms of record attendance which I will certainly congratulate her on. There are 112 delegates here this year, 84 last year, certainly fantastic, Also I think we have the record for furthest a field from which we have a visitor, Kathryn Murphy, who we are going to listen to this afternoon. I look forward to that and thank her for coming all the way from Brisbane, Australia. In the usual time-honoured tradition, I will give you a run down of what has been happening on the medical/ scientific side over the last year or so, but just to remind you of the pretty packed programme we have for you today; We have the irrepressible Helen May-Simera [from my group at ICH] along with Tim Barret from Birmingham Children's Hospital, Peter Hards from Royal Society for the Blind, Joseph Evans, giving us a personal perspective which we always enjoy and some dietetic advice and insights from Chris Cheyette.

We have Professor Peter Hammond here again, you know him from years past, and who has been scanning your faces. We need more faces for our study. It is actually work in progress, but very interesting work whereby we scan several people with LMBBS and also those who do not have LMBBS and compare the faces to see whether there is a typical LMBBS face. So just two points; certainly if you are new [to the society] and haven't had your face scanned before, we would love to see you and secondly, if you are under 20 and you have had your face scanned before, we would also like to see you again, because obviously as you have grown, your face shape changes and it would be helpful to document that as well. Peter doesn't bite, so please have a word with him as he would love to see you.

Lastly I managed to cajole a new patron for the Society; Baroness Helena Kennedy of The Shaws QC - unfortunately she is unable to attend this year, but we will try and persuade her to attend next year.

I received excellent feedback from some of the speakers who were here last year. We had Professor Jim Lupski from the States, and Helene Dollfus from France, I will tell you a little more about her work later; Peter Francis, Ophthalmology, who spoke about gene therapy - sadly he has emigrated to Oregon, West coast of America, so Boo Hoo to him, but we are going to get him back anyway, and we are still working closely with him on various studies and finally, Mehul Dattani is still working with us at GOSH. They really enjoyed they conference and it stimulated a lot of conversation indeed.

I introduced to you last year what we felt were the underlying concepts or causes that give rise to LMBBS. This was the notion of cilia, the fact that these little hairs present on the surface of most cells in the body, come in two varieties, those that beat to get rid of the mucus from our lungs and those that sense the surroundings in the environment around that cell. We still have a lot of work to do but I believe some of the work that has been published in the last year now confirms our hypothesis - that in some way the dysfunction of cilia gives rise to LMBBS. Our suspicions came from several observations - first we know that with respect to vision, the generation of the photo-receptors in the eye are modified cilia. Second, some of the kidney problems are thought to be related to the abnormalities of cilia.

I am reminded to mention that Alison Ross, who is with us today, won a prestigious BA fellowship last year to join the BBC for 6 weeks and for her to get an appreciation of the workings of the media and science. She had a very hands on visit in which she produced lots of reports, which were published at BBC online. She was also a roaming reporter at the National Science Week in Dublin, but one of her notable successes was that she managed to convince the Radio 4 Frontiers Science programmers to make a programme about Cilia which involved myself, Alison and Dr Jo Hill and was broadcast on 11th November 2005 - it is still online if you are feeling bored one night and want something to help you go to sleep, have a listen to that - LMBBS is heavily featured [http://www.bbc.co.uk/radio4/science/frontiers_20051116.shtml].

Again in time-honoured fashion, I have dragged along everyone from my lab - we have two new members who I am now about to embarrass - Suzanne Rix and Jonathan Tobin. So any science questions, please direct it to them.

An Update into the Study & Research of LMBBS

Professor Philip Beales

Briefly I will tell you what's happened in the scientific world over the past year, usually with the regard to the number of BBS genes. The number keeps escalating and, as of two days ago, three more genes have been added to the list. These are *BBS9*, *BBS10* and *BBS11*. I now show these here on the pie chart which shows the proportion of people carrying mutations in any given gene. Many of you will remember that the *BBS1* gene, located on chromosome 11, is the biggest contributor - so if we are going to test a Caucasian from the UK for example, the first gene I would choose to look at would be *BBS1*, because that is going to give us the greatest chance of finding a gene mutation. Next it was BBS2 – now it is *BBS10*. We have now been fortunate enough to work with Helene Dollfus, who spoke here last year. Her group in Strasbourg, France discovered a new gene in a Lebanese family from the mountainous region of Lebanon. There is a village full of families with LMBBS - almost every family in the village is affected by LMBBS. The point I am trying to make is that LMBBS touches every population around the world. This is the very reason we are so interested in collecting blood from such families, as they can help us to find new genes. And so it was this family which helped to identify the new *BBS10* gene. *BBS10* seems to be a very important gene, as it accounts for about 25%of all the cases, so suddenly we have jumped from about 45% (this time last year) to almost 70% - a major advance and I will talk more about this later in testing. So what do we know about these genes? Very little. *BS10* is very similar to *BBS6,* the very first gene we identified in 2000 with Jim Lupski in Houston. It looks like it might be a chaperone protein and therefore we now have the challenge of linking this to cilia function. The other genes, *BBS9* and *BBS11* both discovered by Val Sheffield's group in Iowa is also completely new so we can only guess what they might do at this point in time.

You may remember, one of the other tests we did last year, was to test hearing. This was because the LMBBS mice were found to be deaf. We performed a test that we often do on babies, called otoacoustic emissions – the results of these tests were all normal. However, as I mentioned Helene Dollfus went back to France and tested about 30 or so people with LMBBS from all over France but did a different type of hearing test, called an audiogram. This essentially tests our ability to hear different frequencies at differing volumes. Her results were very surprising as the average age of those who took part was 25 yrs but they had the hearing of a 60 year old. So in conclusion - there appears to be a small amount of hearing loss, but not of a level that would affect you on a daily basis. What we don't know is what happens to hearing when you get to 60 or 70, when we are all going to lose a little bit of hearing anyway. We might need to consider warning Nephrylogists as they often use antibiotics that can harm hearing.

Helen May-Simera is going to tell us more about some of the work she is doing on the hearing aspects with Dan Jagger.

The last bit I want to share with you regards the issue of diagnosis - this has been a complete nightmare from a molecular point of view owing to the large number of genes involved and the cost. So I have been working with a company called Asper Ophthalmics to design a diagnostic 'chip'. This comprises of a glass slide onto which we place the patient's DNA. The slide contains all known mutations in 10 BBS genes. Briefly, DNA will stick to other bits of DNA according to how similar the gene sequences are. Therefore if there is a mismatch we can see this under the microscope a bit like a traffic light – green for a good match and so on. Despite being a limited test (only testing known mutations) it is very accurate and relatively cheap at 130 Euros a test. We just rolled this out a couple of weeks ago, so I believe that is will serve as a useful "first-pass" test.

So why test? In some instances we need to confirm diagnosis, particularly in young children where clinical diagnosis is often difficult. Other people I see want to use testing for family planning purposes and pre-natal diagnosis but we will need to test the effectiveness over the course of the coming year.

Finally, I hope you enjoy the day and hand you over to Helen May-Simera. Thank you.

An Update of the Laboratory Research of LMBBS

Miss Helen May-Simera, Great Ormond Street Hospital, London

Hello everybody powerpoint 1

I am going to talk to you a little bit about how scientists actually study LMBBS. As you most probably realise when Phil comes, he stands at the front at the beginning of the talks and tells of the latest genes that have been discovered and every year more and more genes seem to be coming and added to the list, I thought you might be interested to know what this means for us in the lab. Now of course it is very important to find more genes, because as Phil was just mentioning, diagnostically it is very useful and also if we didn't have the genes we wouldn't really know where to start and what we were looking at. You might be asking what actually is a gene? Well a gene is just part of your DNA and it gives the cell instructions of how to build the protein. It is the proteins that are the very important key aspects of this disease. Now you have to imagine that a protein is a small molecule that we are all made of.

Powerpoint picture 2

If you think of a house, a house is obviously made up of lots bricks. So you take lots and lots of bricks and you make a house, now if you take humans we are actually made of cells, as you know we have lots of different types of cells, we have skin cells, bone cells, hair cells and all different types of cells, and they all come together to make any living organism, now this is anything from animals to plants. We are all made out of cells, now the cells themselves are actually made out of other things called proteins, so again they are like little building blocks that come together, lots of different types of them all come together to make the cells.

Powerpoint picture 3

Now you can imagine then that genes are the recipes needed to make the proteins, so just like you have a recipe to make a cake, genes are just stretches of DNA that are on the chromosomes and give the cells the instructions (recipes) to make proteins. You have different genes that make different types of proteins. So all the BBS genes, 1 all the way through to 11, are all found on different places in the genome (on different chromosomes) and all make different proteins. Some of these proteins are more similar to each other, but by and large they are all very different.

Powerpoint picture 4

What we do in our lab. A lot of the time we are actually trying to look at these proteins and we can use special techniques that allow us to actually look at the proteins in the cells. The way we do this is a little bit like staining a shirt or tie-dying, first of all, obviously you take your cells; now these can be any cells that we take, we can take tissue from other organisms or we can take cells that are growing in a dish. We take our cells and we then have to place them on a glass slide and then you have to add a stain which is very much like a dye you would be using, but obviously this is a very special type of stain which attaches very tightly to these proteins. Now just as if you were tie-dying a shirt, you let the stain incubate for a couple of hours, so that it will soak in and not wash away, you then have to wash away all the rest of the unbound stain, so you do lots of washes. We actually just use normal water for this. You just wash off all the rest of the stain so that you are only leaving the stain marking the actual protein.

Then we use a really powerful microscope, the one we use actually fills up almost a whole room, it is huge, it is called a confocal microscope and it is particularly powerful because you can go very close up and you can see things that aren't really possible with the human eye. It is up to 100 times magnification sometimes, that's how powerful it is.

Powerpoint 5

Now on the next slide you are going to see an actual cell, obviously magnified many times, now I don't know if any of you recognise this cell at the top, this is actually a nose cell, my nose cell, I am quite proud of that. I know that last year some of you had the nasal scrapings done, well this was done to me as well, these are the cells that they get out, they are just the cells that line the back of your nose and these are very interesting cells because they have lots and lots of cilia at the top. Just as Phil was telling you, cilia are just hair-like appendages sticking up and some cells only have one cilia but these nose cells have lots sticking at the top so they look like a brush (here are the cilia sticking up at the top) also at the bottom of the cell we have the cell body which is the brain of the cell, which is where everything happens, its like the big computer of the cell, so what we done with this cell, we actually stained it, Jo did the work for this and in the first picture you can see she stained with a red dye and it marks the cilia so you can just see all the red cilia coming up. In the second picture she has also stained a protein called vangl2 which I am not going to go into because it is a complicated protein, they have all got funny names, but again you can see that it has stained the bottom and top parts of the cilia. We can label the cells with two stains at the same time and you can see you get pretty colourful pictures.

Powerpoint 6

So what does this actually mean, well it means we can actually locate the proteins and we can start answering some questions for e.g. where are all these Proteins in the cells, are they localising round the basal body (the anchor or the Cilium, that attaches it to the cell), are they localising around the cilia, I should mention that the basal body is the area just beneath the cilia where the cilia is anchored into the cell, so that's why that is important. This term often gets mentioned. We can also ask what are these proteins doing, are they moving around, because sometimes you can take live images of cells that are still alive and you can see proteins moving around in real time images and then of course we can also ask are there differences, if we take a cell from maybe a BBS patient and an unaffected individual will there be any differences as to where these proteins stick to.
And then there is the other aspect that cells grow and they divide and then they die and this is called the cell cycle and maybe these proteins are affected in the cell cycle, maybe they change during the lifetime of the cell.

Powerpoint 7

So the next picture I am going to show you looks a bit scary, but it was taken from one of Phil's recent papers, all it really is, is a cartoon of one of these cilia. It shows where most of the BBS proteins localise to along the cilia. In the last couple of years, we have found that most of the BBS proteins, now I don't know about the latest ones because we haven't done those yet, but we know that most of them localise round the basal body, which I was telling you is the anchor of where the cilia actually attached to the cell, and also some of them move up and down the cilium, this is very important, it is a process called intra flagellar transport, which is a long name, but it basically means moving things up and down the cilium.

Powerpoint 8

So obviously we need to know where these proteins localise in cells, but cells just don't work on their own, cells obviously work altogether? Because we are all organisms and we are all a mass of cells coming together, we need to look at other organisms to find out how mutations in these particular genes cause effects in whole systems, so not only do we look at cell lines working together, but we also have to use the help of our special friends, I know Phil introduced you to these last year, we have our very special mice that have BBS and they also help us a lot in studying the disease. In the last year or so we have also been looking at other model organisms and I am going to talk a little bit about these now.

One of them you will actually be surprised is yeast, this is just normal yeast you can buy in a supermarket, that grows and you put in your bread, I will talk about that later, the other thing we also have is zebra fish.

Powerpoint 9

I will just go back and talk about the mice. As you know from last year we were looking at the mice. Some of them couldn't hear, the way that we could test this was that we held one of our mice on our hands so you could administer a really sharp sound just above their head and if they could hear the sound they would just flap their ears back and you could tell they didn't like the sound. A lot of the mice couldn't hear the sound at all, so they weren't bothered and didn't flap their ears back. Because of this, we looked at the patients and as you know last year I did some OAE readings at the conference and they were all absolutely fine, we didn't see any abnormalities whatsoever, however as Phil was saying in France they have done other tests which showed that some patients had sub optimal hearing.

That's all I am going to talk about the patient side of things. Because we were looking at the mice and we thought that there was something going on we thought we would have a look a bit more closely to see if we could find out what was going wrong in more detail.

Powerpoint 10

Now I don't know if people recognise this – it's an ear, it might look complicated, on the outside we have the pinna, which is the part of the ear that we all really know, the bit on the outside, then you have the ear canal which is where all the wax and the gunk get caught and then you have all the actual important parts of the ear where all the aspects of hearing take place. I am actually looking at a very small organ inside the ear called the cochlea which looks a little bit like a snail shell, a very beautiful organ and it looks a little bit like this (slide) but it is not just the cochlea I am looking at, I am looking in more detail at an organ in the cochlea called the organ of corti. Now this might look very complicated. But don't worry, the only thing I want you take from this picture is the fact that we have these two cells here. These two cells are called pillar cells and they are called that because they are very strong, they are holding up the whole organ of corti. They are like 2 pillars leaning against each other and they are very strong and important because they hold up the whole of the cochlea, so I have been looking at these particular cells called the pillar cells.

What keeps them so rigid and strong is the microtubules inside them which is like scaffolding, these are cells full of scaffolding,

Powerpoint 11

We now have a psychedelic looking picture, this is actually a section of a mouse and you can see the cells. You have the two pillar cells which are like the upside down triangle, in green I have used a stain called acetylated tubulin which marks these microtubules which I was telling you about earlier, like the scaffolding of these cells. What is also interesting is that I labelled it with the stain for the BBS4 protein. It seems that BBS4 is all the way in the middle of these cells, it doesn't seem to be anywhere else, it just seems to be in the centre of these pillar cells which of course was quite surprising because we weren't expecting to see it there. And the other interesting thing is that the top picture is a developing ear, and when we look in the adult a couple of months later, you can see that even using the same conditions, the red BBS staining isn't there anymore which means the BBS4 protein is only important for development and after development of the ears when it has all been set up and done, the BBS4 is not actually needed anymore so you don't see it.

It is things like this that have really helped us get an understanding of not just what the genes are for but what the proteins actually do and it is the proteins that actually make the cells and the proteins that actually do the work. The clues we are gaining from these studies help us not just to find out what is happening in the ear, but also affecting processes in other parts of the body, so we are learning more about microtubules and cilia in all these other cell types, which will hopefully give us more insight as to how these BBS proteins are affecting parts of the body such as the photo-receptors in the eye and the kidney cells.

Powerpoint 12

I am going to very quickly talk to you about zebra fish, why is it called a zebra fish? Because they are very easy to look at and you can get very large numbers of them quite easily, so we have jumped on the bandwagon and decided to look at them too and what we have found is that we have got some LMBBS zebra fish, so we can look at what happens to them.

We have a normal zebra fish and a BBS one at 24hrs old, and just from looking at these you can really tell the difference. The LMBBS ones have problems with their eyesight, they can't see as well, because they are still babies there is not that much we can tell about them, this is very much ongoing work and maybe next year I will be able to tell you more

Powerpoint 13

One last thing I want to talk about and that's yeast, which is another model organism we use, we do an experiment that has a long name, it is called the yeast 2 hybrid. Using the yeast as a vessel to do the experiment, like using the yeast as a cooking pot, hijacking the cells mechanism and machinery for making proteins. This is just normal yeast you use in cooking; we just grow it in the lab. As I was telling you before, proteins are the building blocks of cells so we are all made up of lots and lots of proteins so as you can imagine proteins don't just clump on their own they have to stick together to other proteins, so it is a bit like Lego bricks, so if you can imagine red bricks can stick next to a blue brick and they are held together by a green brick, that's how they all come together. So we know that proteins stick together, proteins have binding partners and what we really want to find out is what other proteins stick to BBS proteins. This is important for us to work out how these proteins work, and also it might help us to identify some other genes. Because as Phil was telling you there are still some that are unknown and this is maybe a way of finding out and discovering more genes involved in the disease.

Powerpoint 14

So the way we do this, you will be surprised to know is almost like fishing, so what you do is take your yeast cell, we give the cell information to make a BBS protein (so the BBS protein is the bait) and then we put inside lots and lots of other proteins, so they are all swimming round in the yeast cooking pot, and we want to see which ones will actually stick to the BBS6 protein.
We use the BBS protein as bait and see which one will grab the bait and stick to it.

Powerpoint 15

Now the other way you can see this is like a jig saw puzzle, so if you imagine if BBS protein is a round oval and you have lots of different pieces of this puzzle, one is a triangle, then a round, a cross and a crescent moon, the only ones that will stick to the BBS protein, is the crescent moon because that fits in a nice shape with the oval, so that's a little bit of how this particular experiment works and this has helped us realise that some of the proteins we work with for e.g. BBS2 and BBS6 are actually very strong partners and they work together. Also 2 and 9 work together. We are hoping to get a better picture of these cells to know exactly what is going on, even though we know that it is cilia that is causing the disease, there is 'More to it than Meets the Eye'

Powerpoint 16, 17, 18, 19
Helen showed pictures of the workings of the lab

Thank you everybody I hope it has helped you

Professor Beales thanked Helen for a concise and clear talk and invited questions from the floor.

Question

I was interested how you actually gave the fish and the mice LMBBS

Answer

Different techniques can be used, with the fish you can inject something called a morpholino which is very technical and complicated, but it basically cancels out the gene that we are looking at, so you are selecting for it, which is why the zebra fish are so good to look at because they are one cell zebra fish. You can see on your microscope and you can actually inject it, whereas most animals you can't. So with the zebra fish the small embryos can be injected at the one stage, so you

knock out the gene right at the beginning.

Question

The proteins that you are talking about that are in the cells and obviously there can be problems with the proteins, can that also be connected with the obesity and the proteins that people actually take into their body when they are eating.

Helen asked Professor Beales to answer this

Answer – Professor Beales

What we have to differentiate between, is the protein that the body makes and the proteins that you actually take into the body itself, in terms of food and I would ignore the latter part, because that really gets broken down and assimilated and used accordingly as part of nourishment, but the cells themselves make proteins in their own right. This is work ongoing in the lab to try and find out what role they have in causing the weight gain aspects, but still very early days so we just need to differentiate between the 2, so there is no relationship between what we actually take in.

Question

When you said that giving the zebra fish and stuff that you give to knock out the gene, we have a mutation in the gene so how do they get the condition, just by knocking out the gene.

Answer

When you have a mutation effectively, most mutations are knocking out the gene, it means because of the mutation you can't make the proteins, so with the zebra fish you are taking out the whole gene just to start with, so it is effectively like giving them a mutation.

Question

You said that when you go fishing in this yeast culture and proteins come along and stick to the BBS gene, how do you identify what is stuck to the gene

Answer

Very good question, what we then have to do is we find the ones that are stuck,

we then have to unglue them so to speak and then we sequence the little piece of DNA, we go back to the DNA stage, we sequence that, we look on the internet and find other sequences, compare them and find out which ones they are

"Setting up dedicated clinical services for children with obesity syndromes"

Dr Tim Barrett –Reader of Paediatrics, Birmingham Children's Hospital

It is a real pleasure to be invited here today; it's also nice to see Phil's team, such a dedicated team of scientists working with you on this syndrome. I work at Birmingham Children's Hospital. I would like to talk to you about the background to Obesity in Childhood as we understand it. I will use another rare form of obesity that I have been involved in called Alstrom Syndrome, which has some connections with Bardet Biedl.

I will also describe how we set up medical clinics for people with Alstrom Syndrome connected to Alstrom Family Support Group weekends.

I will then describe how we have applied for government money to fund those and where we have got to. I hope that this will be useful to you if you have plans yourself applying for government support to set up your own clinics.

I work at Birmingham Children's Hospital, which is not dissimilar to Great Ormond Street, but instead of just specialist services it has lots of ordinary ones as well. We have both routine and specialist diabetes services, and we are now developing one for Alstrom Syndrome. In our hospital, patients with BBS are seen mainly by the kidney teams there. We also have set up specialist services for children with obesity and the different complications associated with that. We are also involved in research particularly with different syndromes related to children, not so much BBS but other ones like Alstrom and Wolfram syndrome. I came into this because amongst other things I look after children with diabetes and children with obesity and all of my Hormone clinics were being flooded out with people coming along saying 'oh there's a problem with my child Doc, there's something wrong with his hormones, can you sort out what's the matter with him? ' Usually it is because the child is eating too much and not doing enough exercise; however occasionally we see children who actually have an obesity syndrome. We have been struggling to sort out those people with a rare syndrome such as BBS or Alstrom from those who are overweight because they eat too much.

To tell you a little bit about Alstrom Syndrome I will tell you a little about a young lad called Sean, not his real name. He was a healthy baby, but at 6 weeks of age, developed heart failure and was in the intensive care unit of Leeds Hospital. Fortunately he recovered from that and did well with medicines, but his Mum began to notice that the light was hurting his eyes, particularly in the first 6 months of life and

when he was seen by an Ophthalmologist he was diagnosed with retinal dystrophy. He developed obesity from about the first year of life and at first the doctors who saw him thought his Mum was feeding him too much. When he started school, they recognised he was developing some deafness as well and as a result he struggled educationally. With all of these things, Doctors began to think they were related but he only got a diagnosis of Alstrom syndrome when he was 7 years of age. This distressed his Mum a lot, and I suspect that this has also happened with some BBS families. Since then he has been a patient at the Children's Hospital, because he has developed some complications, including high cholesterol and diabetes. I met him because of the diabetes.

I have tried to compare Alstrom Syndrome with what I know about BBS. In Alstrom Syndrome there are about 35 people known across the country and there may be about the same again that have not yet been diagnosed, so it is pretty rare. BBS as far as I understand is a little commoner and there are perhaps over 500 in this country, but still very rare when the population in the country is 60 million. Alstrom Syndrome children and BBS children are all at risk of obesity; it comes on very quickly and doctors can confuse it with common obesity when it is clearly not the same. Alstrom children also get poor vision and indeed, the lights hurt their eyes very early on, as with Bardet Biedl. Unlike BBS, Alstrom patients get heart failure infancy and those who don't get it in infancy can go on to get it in their teenage years. Some people with BBS have extra digits; the 3 BBS patients I have seen have all had kidney problems, which is not usually a feature of Alstrom. Doctors have often struggled to make the diagnosis, and a lot of families with Alstrom complain about this. In both Alstrom and BBS children get high lipids in the blood and diabetes; both conditions are also genetic and the inheritance is called recessive. In recessive inheritance, you can have a healthy mum and dad, giving rise to a child with Alstrom Syndrome or BBS; the Mum and Dad are not affected at all. However there is still the risk of having another affected child; in the case of Alstrom the risk is about a quarter. In Bardet Biedl there are 11 known genes so far; with Alstrom we only have 1 known gene that seems to account for at least 80% of all people affected with it. I have mentioned that there are 35 families. We have done a study working with some UK Alstrom families, and found that most of them have mutations in the Alstrom gene. There is also an Alstrom Mouse, which gets fat and gets the eye and ear problems; work is being done in a laboratory in North America to find how the gene works. Scientist are using very similar techniques to understand Alstrom syndrome just as they are with BBS. The Alstrom protein also localises to part of the cilia structure just as one of the BBS proteins does. I got involved with this because the Alstrom Society has a family support group just like yourselves, although on a smaller scale, set up by a lady who had 2 affected children in Devon; she persuaded the local doctors to take and interest because it had taken so long for her children to be diagnosed. One of these local doctors, Dr Richard Paisey arranged for family weekends to occur Devon. They would run a medical clinic on the Monday after the family weekend to see people with the condition and try and pick up conditions that had not been spotted by their local doctors. This was for adults and children, and at the clinic they had a dietician to help with the food aspects, an adult doctor, a heart doctor, because heart failure was a

feature and a nurse and a kidney doctor. They detected complications and advised on this and also arranged for research studies, it was great for families living in the South West of England, but as you can imagine it was not easy to get to Devon if you lived in Scotland or the North of England, which made it difficult for families to attend. I was lucky enough to get invited down to a family conference in Devon in 2002. The clinics were held on a good will basis, so all the medical staff were giving their own time and using the hospital facilities without it being paid for which was fine, but is wasn't sustainable to continue. It was linked to a family weekend, so lots of families were able to get together to socialise and then have a medical clinic at the end of a weekend. Consequently we set up one in Birmingham in 2003, which was popular with the families because it was more central. We took over the Heart Investigation Unit, because that was the relevant complication of the Alstrom Syndrome, on a Monday morning when the unit was not being used. It has a nice waiting area and we could organise refreshments for all the families. The families would attend the conference at the Novotel in Birmingham and visit us on Monday. At the clinic we would have children's specialists as well as adult specialists all working side by side, dieticians, psychologists, doctors and nurses all working side by side and we have 16 families + their guide dogs all up on the Heart unit. The aims of the clinic were to try and develop expertise about Alstrom Syndrome. Families had complained that if they were diagnosed in Essex for example, the local doctors had never heard of the condition, they did not know what was involved in treating them and did know what were the complications to look out for and they also felt a great sense of isolation, because they didn't know anyone else with the syndrome either. So by coming together once a year at the family weekend like yourselves they met others and were able to support each other, but also the doctors who were involved were able to build up a clinical picture about the syndrome and work out what complications to look for, but then by developing that experience they could then contact the local doctors and advise them on how to care for the individuals locally.

The first aim was to build up some clinical expertise about the condition; secondly to offer an annual check, especially for children, to help their growth and development, to avoid complications and actually screen for them. Sometimes it involved a blood test, but more importantly a heart check, this was very popular, because the families felt they could get it all together at a 'one stop shop'.

Thirdly it was to advocate for affected children with the local doctors to make sure they were getting all the services needed and the correct help and benefits. Often they were struggling in different parts of the country, working through the NHS minefield to get their entitlements and services.

Finally as well as family support to try and get clinical studies that would enable families to be identified and hopefully get the treatments and support.

I will give you an example: one of the first studies did in Birmingham was to develop a genetic test. Lots of families had been approached in the early days by doctors in North America asking for a blood test to try and find the gene for Alstrom Syndrome;

the gene was eventually found by 2 groups, one in Newcastle and one in America about 3 years ago. Families then wanted to know if they had mutations in their Alstrom gene but found it very difficult to get results back from those places, so one of the first things we wanted to do was try and identify the mistakes in the Alstrom gene and feed the results back to the families, I am pleased to say it was easier in Alstrom than it is in BBS, because there is only one gene, despite the fact that it is a very big gene. By looking at the different families we could actually find the mistakes and feed that back at the family clinic. We found that there was one hot spot in the gene and if you screened that, not the whole gene, you could find most of the mistakes. Another study was what happened in the childhood obesity and by measuring people's height and weight and also body compositions by a special scan in the x-ray department we could have a look at the amount of fat in the body from it. We found that as you went from childhood to adulthood, the obesity gets better. We were surprised to find this, as we knew that diabetes became commoner as Alstrom people became older. We know that diabetes is related to being overweight, but we do not understand why Alstrom patients get thinner yet still get diabetes.

So as a result of teaming up with the Alstrom families at these clinics we have been able to get some clinical studies published in different journals and get advice on how to manage Alstrom. That has been fantastic because it has been a real team effort, the families have all been part of this and the Family Support Group of Alstrom UK has been a real support and acknowledged on the papers we have published.

I mentioned that the problem with these clinics is that we did not have any funding for them and it was all done on good will. The families had difficulty reaching Devon and families were still being diagnosed late, because the local doctors, not unreasonably, were slow to make the diagnosis because they hadn't seen Alstrom before. I was using up an awful lot of good will on the Cardiac Unit of the children's hospital by running these clinics, so had to give lots of boxes of chocolates to the nurses to let me carry on using these facilities. A lady called Kay Parkinson runs the Alstrom Group persuaded me and my colleagues at Torbay hospital to apply for government support to run these clinics. We applied to an organisation called NSCAG: the National Specialist Commissioning Agency Group. They provide specialist advice and help to every local hospital providing the service, so they fund small numbers of centres for very rare conditions. They do this by cutting off little bits off the budget off every hospital in the country and then give it specifically to the specialist services. So we obtained a dedicated fund. The budget of NSCAG is significant, about £185 million and that is what they spent in 2005 for about 20 different rare conditions. Two examples are Children's liver disease, funded through NSCAG in 3 centres; a rare skin disease called Epidermolysis Bullosa; and another metabolic disease called Gaucher's. The NSCAG commissioners told us that they wanted to fund an already established service; and they wanted to see clinics that have actually worked in practice before funding, so it is a bit like the chicken and the egg. If you have no money to set up the service and they wont give you any money until you set it up, you are in a bit of a difficult situation They also fund clearly defined groups of patients and they are happy to fund a small number of centres, but they like to do it in partnership,

usually with more than one centre. It is vitally important to justify the costs; it has to relate to a rare condition that it would be impractical for each primary care trust to commission separately. The NSCAG website suggests conditions which affect less than 1,000 people in the UK.

So in May 2005 we then had site visits from NSCAG Commissioners who came both to Torbay and Birmingham and met the business managers of the hospitals, the patient support group and the clinical teams. The NSCAG Commissioner was particularly interested, because of the late diagnosis: some children were not being diagnosed until 18/19years of age. Potentially from the government point of view this could have medical legal implications, because sooner or later someone is going to die before they are diagnosed. In addition, Alstrom Syndrome progresses and needs expensive treatment such as heart/lung transplants. From NSCAG' s point of view, if we could set up a clinic to screen for those and hopefully try and pre-empt or delay the progression, it would be cost effective.

Another reason for NSCAG's interest was consumer pressure. As I have said the patient support group for Alstrom was led by a very determined lady, Kay Parkinson and I believe a lot of the families wrote to NSCAG to get these clinics set up.

Finally NSCAG were aware that there were existing clinics, both in Torbay and Birmingham and they liked the idea of a joint application from 2 hospital trusts and if they could see two Trusts working together in a sensible way they thought that perhaps that would be good thing to fund. So we got agreement in principle last September to actually fund the service. We then had to get the business managers to get full costings for the staff, the rooms, the equipment etc and they gave us a joint budget in the regional area of about £300,000. It may not sound a lot, but on the other hand for a condition like Alstrom which has never been funded before is absolutely fantastic and we were really pleased. This then led to face to face negotiations, we all had to go down. In the discussions they explained they would only fund NHS services that were relevant, because we were working very closely with the Patient Support Group.

We arranged to have four clinics a year at Birmingham Children's, 8 -10 children coming to each clinic; a transitional clinic for teenagers, so moving from the children's to the adults service, leaving 2 clinics a year at TorBay for the adult services and one of these clinics was linked to the family weekend. The next family weekend for Alstrom is Birmingham in October and at the end of the family weekend there is a specialist clinic on Monday, so people can come along to the family weekend, stay an extra night and visit the clinic the following day. Because a lot of people with Alstrom seem to be in the Leeds community and haven't been identified before and clearly there is a real need, we are going to arrange to have an Outreach Clinic. When we visit Leeds once a year, we can see families there who cannot make it to Birmingham and clearly there is a need for families who want genetic testing, so we are sub-contracting that out, partly to Leeds who have already got an NHS services and also to provide it locally which will make it even faster and also these two centres will share in supporting and sharing a Family Liaison Officer, which will probably be someone from

the Alstrom Family Support Group. For NSCAG we had to demonstrate some measurable outputs so that we can show that we have actually helped families.

So where are we at the moment? The service was commissioned on the 1st April 2006. The first clinics are going to go ahead at Birmingham Children's Hospital, and we already have 11 families coming on that day.

So in summary. We have a network of specialist clinics which we think are going to provide practical help for families with Alstrom syndrome. We have built up a team of doctors, nurses, dieticians, psychologists, audiologists and cardiologists who are all becoming experts in Alstrom syndrome.

We desperately need and are very proud of our relationship with the family support group, because without them we wouldn't have got this started in the first place and it is only through that partnership that this is going to work. If there is anything that we can do to help your society I would love to try and do so. I hope that some of our experiences will benefit you in the future if Bardet Biedl goes along this route.

Thank you very much

"Then & Now"

Peter Hards, Royal Society for the Blind

Hello and Good morning my name is Peter and I work for an organisation called The Royal Blind Society, first of all I would like to thank Chris for the invite, I do feel rather humbled being in such exalted company, basically I am the only person speaking today who I have never heard of. Old joke but thank you for laughing anyway

I don't use any visual aids, I am used to speaking to groups of blind and partially sighted people. The title of the talk is Then and Now, basically what I am going to do is to tell you about what the Royal Blind Society does these days and I am going to start with a little bit of history.

The Royal Blind Society started in 1863, 143 years ago as the brainchild of a Victorian businessman, Thomas Pocock Snr and the Charity began life as the Protestant Blind Society, I suppose you could call Thomas Pocock a typical Victorian philanthropist, it changed its name in 1884 to the Blind Pension Society

and basically did what the name on the tin and offered small pensions to those who if they lost their sight also lost all means of earning a living. The royal bit came in 1887 when Queen Victoria celebrated her Golden Jubilee, she took on patronage of several charities, including the Society and that royal link has remained unbroken with Queen Elizabeth 11 our current patron. We were talking earlier about fund raising and budgets etc, well Her Majesty sends us a Christmas card every year and a very kind cheque as our patron of £190.00.

The Charity continued along those lines as a pension awarding charity, right up to the advent of the Welfare State in 1948, when obviously the State began to take over a lot of the responsibilities for that kind of work. So from that time the organisation began to reinvent itself into a grant giving organisation, which we still are today, continued along those lines until the end of the 1990's until we received a letter from a little charity working in West Sussex who had recently closed a residential home called Honeywood House, the letter basically described the closure of this lovely home, beautiful gardens and backing on to the sea.' We think we should be doing something positive with this home, but don't think we can do it on our own, would you like to come and talk to us about any future usage'. Now this letter was sent to about 30 other charities all working with blind and partially sighted people, perhaps they didn't put stamps on all the letters, because the only reply this little charity received, was from The Royal Blind Pension Society. The two charities got together and decided that Honeywood House would make a marvellous holiday hotel for guests with sight problems. They employed me for some reason in August 2000, for which I was very grateful and I had a wonderful time, because as I am sure most of you know, it is much more fun when you are spending someone else's money and I had a great time spending £35000.00 of the two charities money in totally renovating, refurbishing and in some parts rebuilding Honeywood House to make it the hotel it is today, looking back now and hindsight is a great thing, it was probably one of the most exciting times of my working life, but also, the most terrifying, because when I turned up on day 1 all I had with me was a pad and a pen, no customers or guests, no staff or colleagues, no brochures, no policies or procedures, but, somehow we muddled our way through and opened our doors as a hotel. Specifically for guests with sight problems for the first time 10th February 2001. I am very pleased to say that after spending that huge amount of money, Honeywood House has gone on from strength to strength since. I had a very pleasant 3 years there until my wife and I decided to relocate back to our spiritual home which is in the Midlands, but then I received a telephone call from a Charity called Henshaws Charity for the Blind, working in the North of England, Manchester, Newcastle, Harrogate Liverpool, asking me if I would consider taking on another hotel, so literally myself and my boss a chap called Graham hotfooted up to Manchester for some very amicable and constructive discussions with the people at Henshaws, with the result being that in December 2002 we took over their Belmont Hotel on the seafront in Llandudno, that concludes the history of the present day Society.

These days we work in two main areas, I will give you a brief resume of the holidays that we offer and hopefully you will come and see me this afternoon in the workshop otherwise I shall get very lonely. I will explain a little about our own hotel, Honeywood House which has a special place in my heart, it is still my baby in many ways even though someone else is in charge. Honeywood is a small hotel, deliberately small, situated 2 miles from Little Hampton and it is filled with amenities and facilities specifically for people with sight problems, for instance, there are talking notice boards situated around the building, to tell you where you are, which is a bonus, but also how to get to other parts of the hotel, but probably the best example I can give you, is to describe what is in each of the on-suite bedrooms, all the rooms have emergency alarms, so if guests have a problems they can press a button, pull a cord or have a pendant alarm if they prefer, which brings a member of staff to them immediately, there are touch lamps, avoiding having to press buttons and switches and burning yourself because invariably the switches are near the hot bulb and you say ‘oh crikey what a pity’ when you have burned yourself. Talking clocks with a range of alarms from ‘cock-a-doodle to jersey cows’, guaranteed to wake you up with a start first thing in the morning, British wireless with cassette in Braille and large print and in the hospitality trays where you make your teas/coffees there are liquid level indicators, which cuts down the possibility of anyone scalding themselves, especially when making that first bleary eyed cuppa at 7am, these facilities are right the way through the hotel, 2 comfortable sun lounges, dining room, which offers half board accommodation and like most hotels, provides evening entertainment, trips out in their own mini bus, and of course the all important “Bar”. One of the success points of Honeywood is its size, maximum of 18 guests at any one time with ground floor and first floor bedrooms, connected by a talking lift, it means that guests who may be coming for the first time with significant sight problems very quickly get to know each and the staff and I think that is of particular benefit to them. Honeywood offers very much a one to one personalised service, which guests who return time after time very much appreciate indeed.

Having said that there are plans to increase the size in the near future to provide 4 single rooms and one family room on the ground floor and to build a huge conservatory on the back of the building, so that is something quite exciting for us.

The other hotel The Belmont, I think is on the best position on the seafront in Llandudno, underneath the great orb, near the pier on a safe, clutter free promenade has all the same facilities, so I wont go into detail because of the time. Because of its size it offers a very different service to Honeywood House, which is small and intimate, The Belmont because of its seafront position a friendly seaside, family atmosphere hotel which is particularly valued by the very many large groups who visit along with individuals, both hotels are doing extremely well.

Speaking to a couple a few years ago, who loved visiting both hotels, said that

just occasionally, they would like to go on a self catering holiday where they could do what they wanted, when they wanted or not as the case maybe, this struck a chord with me and we started to look around for something to fit the build, we found a holiday home on a Hoseasnons site on the seafront in Burnham–on- sea, which had all the facilities indoors and out, the holiday home has been especially adapted for people with sight problems but also for people with limited mobility, there is a ramp up to the front door, full wheelchair turning circles in lounge and bedrooms, the lounge also has a large pull out bed, we recommend that 5 people use it at any one time although it will accommodate 8, we think 5 is comfortable, this offers a good low cost alternative to a hotel holiday and by that, for instance in the height of the summer this year say between July, August and early September the holiday home costs £250.00 that's not per person, that's for the holiday home, so for 5 people going on holiday it is only £50.00. and I am sure that not even in Northampton in the height of the holiday season, can you get a holiday for £50.00 each, unless of course you stay at home, I think this has been proven by the fact that all I have left this year are cancellations

Now we are always looking to prove the number of quality holiday options for people with sight problems and about 18 months ago, we looked at re-activating holidays for people at recommended hotels on those wonderful holiday islands of Jersey and Guernsey and it is a terrible job sometimes, but someone has to go over for about 10 days with the objective of inspecting as many hotels as possible on both islands, looking at them from the experience of Honeywood House on all sorts of levels, but basically accessing the buildings, good level steps, a ramp perhaps, ground floor bedrooms etc, staffing levels and level of assistance, attitudes of owners and managers which became apparent to me, was terribly important while I was there, partly because the Disability Discrimination Act, basically governed on the mainland, doesn't apply in Jersey and Guernsey, they are part of the British Isles but not the UK, a good example of that is that 2 of the hotels I visited didn't accept guide dogs!!!, they didn't make it into the final selection, but after this arduous 10 days which I thoroughly enjoyed, purely by accident it came down to 6 hotels on each of the islands, these have been put into a brochure offering a selection of travelling choices by sea from Weymouth and Poole or Portsmouth, by air from 28 regional airports throughout the UK, including some I had never heard of!!! We had an amazing year last year and this year is proving to be very popular

We are always looking for new options and as I travel around the country, often by invitation (I don't impose myself) when I am talking to people and we have a Q&A session at the end and very often it is the same statements and comments come up. 'I'd love a holiday, but I worry about getting there and back safely' 'I'd love a holiday, but insurance can be a problem, because I have medical problems, that many insurance companies wont cover' etc. We have looked at these comments to try and ratify the problems, and are currently working together with another holiday mainstream provider. Later this year we will be

publishing a brochure which will advertise a further 6 hotels on the UK mainland all with a good range of facilities and special offers, but including, local pickups, insurance, the service of a tour manager etc, which is something for me to look forward to.

That's our holidays for the moment and I wont expand on that because of the time, but the other main body of work we do is in grants, these are not insignificant amounts of money, last year we awarded about £105,000.00p in total, this year will be nearer £110.000.00, these grants will be specifically for visually impaired people on low incomes, there is a little means test involved, I wouldn't kid anyone there isn't, but there has to be, but if you have ever applied for a statutory benefit where you have to fill out 3 full size books and you need a sleeping bag, flask of coffee and 3 or 4 blankets and pillows just to keep you awake, nothing like that, just a couple of sheets of A4 paper, to provide benefits in whichever shape or form they might be, it could be for transport to get to their Social Club or group every week, it might be for some technical equipment, perhaps a talking microwave, which has reintroduced so many people to the benefits of cooking again, which for about £200.00 they are a real benefit. The thing I am particularly pleased about with our grant scheme is that not only does it provide them an award, but it provides it quickly, in some cases same day or the next at the latest.

Peter thanked us for the invitation and ended his talk with some true stories from Honeywood house which caused great hilarity from the audience

Professor Beales thanked Peter for a wonderful rendition; asking delegates to visit him in his workshop this afternoon for more information

"An Insight into No Sight"

Personal perspective – Joseph Evans

Hello everybody, my notes are no good to me because I can't read them, so forgive me if I lose track of what I am saying

My name is Joseph; I will be 21 this year. I live in Church Stretton, Shropshire.

Statistics say that parents carrying the LMBBS gene have a 1 in 4 chance of having the syndrome, true to statistics I am that 1 in 4. I have an older sister and 2 older brothers. According to Mum I was well behaved prior to birth. The first surprise in store was that I had the extra digits on hands and feet. I never actually realised this as they were removed in the first few weeks of my life. There was no hint of the syndrome being present in

my pre-school days. I was diagnosed with LMBBS when I was 8 years old. It took a long period of time and many hospital visits to arrive at the final diagnosis. It started to present itself when I was unable to read from the blackboard and amused myself by aggravating those around me, those who know me, would say that nothing has changed. As well as my poor vision my weight gain was also causing concern, that hasn't changed either.

We were referred to Paediatrician in Liverpool who had his suspicions and directed us to Moorfields Hospital, soon after this LMBBS became a household name

The local education authorities were very supportive and I soon had my own specialist support teacher, I was taught touch typing skills which have held me in good stead. Eleven years old and I became a student at RNIB Worcester, when I found the help of a specialist school a great help. My mobility improved, although it took me a very long time to get used to a cane. This is no longer the case and I have found that my cane has worked like magic attracting well meaning people in all directions, sometimes far too many. It is quite amazing how the street crowds fall away to let me through, especially beneficial in battling through the London underground in rush hour.

I am currently studying at RNC Hereford where I have attained A Level History and am hoping to achieve the same in Sociology and Critical Thinking.

Some people I meet think that being visually impaired means that you can't do very much. I have never really found this to be the case, although I can be selective and it is strange how, it is always the boring things that I can't do. I have no difficulty in catching a train to Manchester, when meeting up with my brother to watch Manchester United, followed by a few pints in the pub. I am also quick to accept an invitation to visit my brother in Brighton, a four hour journey with two changes, but so far they have not lost me!!

I have also mastered the art of taxi drivers doubling up as "meals on wheels". Our local KFC is a little too far to walk. "Oh Dear, I don't think I should have mentioned that as Dr Beales, sorry I mean Professor Beales will not be too happy " my diet is an ongoing problem.

Supermarket shopping can hold a few surprises; strangely the contents of jars and tins are not always what I thought they were. I found that conditioner doesn't have quite the same effect as shampoo.

I have enjoyed my years of study. I have found modern technology a big

help in this area. I have Feature Magnification software both at home and college. Last year I purchased an amazing piece of machinery known as the scanner Humanware. This enables me to have any typewritten word translated into speech. I have been a keen collector of first edition books over the years and I can now appreciate their contents; it also comes in handy for reading my post and typewritten instructions. I can also access college notes, although the majority of these are now on disc.

Three years ago I began learning Braille and am now busy reading Harry Potter and the Philosophers Stone.

I enjoy travel and have a keen interest in World War 2, so I have been able to combine the two by going on coach holidays through Europe for the World War 2 theme with a tour guide, the hours travelling were spent listening to the information provided.

We visited such places as Colditz, the three dams in Germany which were the sight of the Dambusters and Dachau Concentration Camp which was pretty horrendous. I found East and West Berlin very interesting and was able touch the remains of the Berlin Wall. Headsets were provided in English, making it easy to access information. I have had two such holidays and hoping to go again in the autumn.

Last year I was fortunate to join the International Computer Camp, held in Prague. Students from 15 different countries around the world meet up every year. Seven of us travelled from the UK and had a brilliant time.

My other hobby is Manchester United. I belong to MUDSA which stands for Manchester United Disabled Supporters Association, an annual cost of £25.00 for me and one other. We have free entry to games, a special area where headsets are supplied, a Christmas lunch when we meet the players and have a photo to prove it and then an invite to the Annual Dinner, this year Sir Bobby Charlton was the guest speaker. Anyone who has a visual impairment or disability can join MUDSA, so if there are any fans out there, please contact me.

“It is not an easy ride having LMBBS, it poses a lot of challenges, not problems, challenges, but if at first you don’t succeed, try, try and try again”

Professor Beales thanked Joseph for his inspiring perspective and wished him well for the future

“Obesity & Diabetes -Can Food Choices make a Difference”

Chris Cheyette, Diabetes Specialist Dietician, Mid Essex Hospitals

NHSTrust, Broomfield Hospital, Chelmsford,

0pen with first power point

I would like to thank Chris for asking me to come along to chat with you today,

What I am going to do today is try and give you a few practical and helpful tips, talk about a few controversial areas such as low carbohydrate diets I am sure lots of people have got ideas on what works and what doesn't in terms of diet and weight loss.

Powerpoint 2 outline of talk

I would like to spend a couple of minutes talking about what diabetes is, to make people more aware of the condition and a then talk about the symptoms as well, and why there is a link between Type 2 diabetes and Obesity. I'll then talk about whether food choices really can make a difference or not, what the other causes of obesity maybe in terms of genetics, a little about physical activity and a few ideas around that.

Powerpoint 3 & 4 Causes & Symptoms

So in terms of diabetes, there are two types, there is Type 1 and Type 2 and type 2 diabetes is the one that is mainly linked to obesity, it used only to be present in adults, but it is becoming a lot more prevalent now in younger people. It is basically where the pancreas, an organ in the body, doesn't produce enough insulin or sometimes it is more that it overproduces insulin. This is due to the weight/fat that is being carried around the middle and the waist.
There is also a certain a genetic link as well but also environmental factors, because of what people eat and how big they get, but also people seem to think that having diabetes is caused from eating too much sugar, but that is not the case. There are a lot of misconceptions about diabetes in terms of if you have diabetes, you have to cut out sugar from your diet, but that is not the case.
So here are just a few of the symptoms to make you aware of them. People normally have impaired glucose intolerance (pre diabetes) for a number of years and this then usually moves on to Type 2 diabetes, the symptoms people normally get with type 2 diabetes when it is fairly well established, is feeling thirsty a lot of the time, going to the toilet very frequently and may start to go in the middle of the night as well, being quite tired, irritable, blurred vision in some people, some people can get weight loss. The diagnosis in type 2 diabetes are usually 80/90% of people are overweight, If people have one or two parents with type 2 diabetes they are much more at risk of getting it as well.

Powerpoint 5 Diagnosis & 6 Uncontrolled diabetes

As I say it is becoming a lot more common in younger people and certainly I have been working in diabetes for 3 ½ years, and a dietician for over 5 years and when I started those were very few and far between of diabetes type 2 in young people, but I now have 6/7 adolescents I look after with type 2 diabetes, so it is becoming a lot more common. You only have to look over to America to see the prevalence there; it is actually taking over the more common type which is type 1 diabetes at the moment over there. In terms of diagnosis, it can usually be diagnosed with a urine test followed by a blood test and pre-diabetes is usually diagnosed with a blood test as well. The reason why diabetes itself is a problem is that it can lead on to other complications. The reason for that is if you are carrying too much sugar in your blood stream, and your sugars are out of control it can start to cause damage to lots of different organs in the body and to the blood vessels as well, so it can cause problems to the eyesight, the heart, giving an increased risk of heart disease, kidney and nerve damage, especially down in the feet because that's where the blood fails to get too.

Powerpoint 7 Can it be stopped

In terms of whether it can be stopped, well yes it can, there is good evidence to show that type 2 diabetes can be prevented, 2 the main studies that have been done, and they looked at lifestyles and exercise and showed that by paying a lot of attention to the foods we eat and also to the amount of exercise we do, you can actually reduce the incidence of diabetes by about 60%. The big challenge for us all is how we can get people to look at their lifestyles and how we can get them to look at the types of food they eat that are going to help and increase their exercise, over the last 50/60 years the amount of exercise we do has taken a nose-dive, there are a lot more things now in our everyday life, we have television with remote control, also every room in the house seems to have television these days, people are using their cars a lot more often and people have a concern about being safe on the street as well, so people do tend to do a lot less exercise. Schools are tending to get rid of their playing fields, there is not as much physical activity on the curriculum as there used to be and the government is recognising this, but there is still a long way to go before we start to reverse the problems. So the reasons why weight really matters I because it's what you carry around the waist here that really matters and increases your risk of type 2 diabetes. It is not just diabetes it also increase the risk of the other conditions, which lumped together, are called the metabolic syndrome. This is something that can affect your cholesterol levels, increase the risk of blood pressure and it is all linked with carrying too much weight around the middle.

Powerpoint 8 why waist and measurement matter & 9 body index

Ok so when we say obesity what do we actually mean, well in children it's really done on what we call a percentile chart and I don't know whether any of you have had any experience of going along to see dieticians or nurses and their weight is being plotted on a chart, the way this is done is to look at the weight

and the height to work out the body mass index, which is just a measure for weight and height, to plot it on a chart to see if it fits into the normal range for that age group or whether it fits into a higher range. What we really need to be doing is picking up children who are at a higher risk of being overweight for their age and try and pick up early so that can do things with the children and their families as well from a very early age to try and introduce things that are going to slow down the progression and the increase in weight. A child doesn't necessarily have to lose weight for it to be of benefit to their health and they can actually lose inches off their waist by doing physical activity or change in diet, without losing weight if they actually lose some of their waist size, it can increase and benefit their health and decrease the risk of getting type 2 diabetes as well. In adults as well, what we use is the body mass index, as I say that is the measure of weight and height, we define being obese as a body mass of over 30 which puts you at an increased risk of many different diseases including type 2 diabetes and again we can look at waist circumference as well as a cut off point to a high risk so in women it would be 88cms and men 102 cms.

Powerpoint 10 Causes & 11 cartoon I hate diets

So in terms of cause of obesity there is quite a lot of different reasons why people might become obese, certainly for LMBBS there is definitely a genetic link there, there is another syndrome called Prader-Willi Syndrome, which people may have heard of which is also linked significantly with obesity. People with Prader-Willi tend to overeat and they don't tend to feel full, it is my understanding as I say I don't know a huge amount about LMBBS that there is certainly a link there and someone was saying earlier on that that's the sort of thing they tend to find that they do overeat. But also with obesity as well, there is a genetic link, it is a bit like a jigsaw puzzle, there are environmental factors as well, the kind of lifestyle you lead and you lead as a family that can certainly affect the amount of weight you or your children may put on. There are also some triggers to eating as well, so the habits that we tend to learn from a young age can go through all the way into adulthood. I have patients now who say that when they get home from work at 5pm which is the time that they used to get in from school, they always used to raid the fridge, they still carry on with those habits and these are the things they need to try and break. Medication can cause weight gain as well people often say that because of their weight they have a slow metabolism, for the majority of people this really is not the case in fact people that are overweight normally have a higher metabolism, they tend to burn up more calories and need more calories to stay at that weight.
It is ok knowing what the causes are, but what are we going to do about it in a practical sense, I think anyone who has tried to lose weight can relate to this (see slide) hating diets. Know one likes being told what to do and put on diets, you only have to pick up any magazine or look at the internet and you will come up with thousands of different ideas e.g. the Atkins diet, GI, south beach, blood type There is a lot of conflicting information and confusion and this is why as a nation we really don't know what the right thing is to do for our family and children.

Powerpoint 12 Energy Balance

We were talking earlier about low carbohydrate diets, there has been a lot of work done on this, to see what the effects are over the short and long term and there is some encouraging data to show that low carbohydrate diets do give a good amount of weight loss, but the only problem with that data is that they have done it for 6 months and compared it with a classic low fat diet, showing that with the low fat carbohydrate diet you got more of a weight loss within the first 6 months but then they found that people falter that and they looked at a year's time, that people following the same 2 diets, was that there was no difference at all in the weight loss, this has been repeated in a couple of studies now, so at the moment in terms of dieticians and the advice we give out people, we certainly do not recommend that they follow low carbohydrate diets, because there are also other problems associated with that, one of them was picked up on earlier, that the amount of fibre can average at around 3 to 4 grams a day and we should be eating around 18 grams of fibre daily, the other thing is that carbohydrate gives us energy. I don't know if anyone has tired to do a low carbohydrate diet, well some find their energy increases but after following it for some time, they find their energy levels can start to dip quite a lot, people can get quite bad breath, constipation and also the risk in the long term of the very high fat concentration, so at the moment there really is not enough evidence to say that it is going to work, but we really can't discount low carbohydrate diets because they might be a good thing for some people and different syndromes as well, so work needs to be done in that area, but it is not something we can really recommend to people at this moment.

The other diet that people might have heard of is the low glycaemic diet of the GI diet that seems to have been quite popular, that one maybe a bit more promise for people and that certainly for us working in diabetes is something we advocate to our patients. It is quite a complicated diet in terms of understanding how it all works, but it is basically linked to certain carbohydrates in the diet that we eat and how quickly they get turned into sugar, you will have probably seen the balance of good health there, now carbohydrate is in your cereals and potatoes, pasta, rice pulses, the thinking is that if we eat carbohydrate that gets turned into sugar slowly then what's going to happen is that our bodies wont need to release as much insulin, because insulin is the stuff that clears those foods away and too much insulin in the body is thought to be linked in some people to increase the amount of obesity they have, so if you can have more foods which contain slow releasing sugar then it s thought that might benefit you. So there is some evidence there that it might help.

Powerpoint 13 & 14 getting the balance right

When someone is trying to lose weight, one of the things we look at is energy balance, basically the scales really, it is how much food is going, so how much

you are eating and how much energy is going out, how much energy you are expending, in order to get weight loss what we have to do is to tip the scales when you have an imbalance, so in order to put food in for the less energy you need or do we need a bit more energy going out, ideally we want the best of both worlds, so a bit less energy going in and a bit more going out and in that way hopefully over time the weight will start to come down. The thing you have to remember with weight gain and weight loss, is that it takes a number of years for that weight to start going up and it can be a continuous process for people to put weight on over a number of years and for people that put on weight in very early years, when they are children, we need to slow down that weight increase if we can, it doesn't have to be a lot to slow or stop the weight gain. It may only need to be a 100 calories a day difference, so that's not really a great deal, but it is keeping it stable over a number of years, there is no point ingoing on a diet for a few weeks, saying well that didn't work, you really need to be looking a whole family approach, maybe instigating things that you are going to change the family over a number of years, looking at what foods you eat and change little by little and hopefully that will start to have an effect This plate model is quite good if you can imagine your whole days food intake, everything you eat on one plate this is the kind of proportions of food you eat where your food should come form and you can see the 2 biggest areas we advocate now is from lots of fruit and vegetables and quite a lot of carbohydrates foods, but the right type of carbohydrate foods, so we are talking more wholegrain and granary types of bread, things like pasta, the smaller portions should really be coming from the protein foods, that is meat, fish eggs and cheese and dairy foods are also very important for calcium foods like milk, yoghurt and then the smallest portion are the really high density fatty foods, like crisps, chocolate, KFC, McDonalds, burgers etc, we are not saying that people shouldn't eat these foods, just in moderation and the smallest component of the diet. It's also difficult with children to try and marry up something like this and making sure that you have things in the house that everyone likes as well, rather than feeling that you are putting people on really strict regimes, so there is some work being done on a kind of traffic light system which is a good way of teaching children how to see that red is almost like the foods that they shouldn't eat very often, but they can have occasionally, it is quite useful, they have done this at the clinic with younger teenagers where we have worked with their parents to build up a list with their children and we have put a traffic light system on their wall at home and they have a red list, which contains the foods that they are only allowed to eat occasionally, they have an amber list which I foods they might be allowed to eat every other day or once every 2 or 3 days a week and then you have the green foods that they are allowed to eat quite freely and it is quite good to teach the children that so that they understand, why they should not have the other ones to often in their diet.

Powerpoint 15 why not swap it & 16 portion sizes count

So it is really trying to get the balance right and trying to look at the different

foods, try to aim for lots of fruit and veg, complex carbohydrates. Slow releasing ones potentially, smaller portions of meat, watching out for fat intake and having occasional treats. It doesn't really take a lot to make a few swaps within your everyday food patterns, that add up to quite a huge amount of calories, there are just a few examples if you think about a normal bacon, lettuce and tomato sandwich, if you were to swap it for a more healthy one where you have low fat mayo, cut the rind off the bacon, you can save a whopping 326 calories a day about 25 gms of fat, something like shepherds pie and vegetables instead of steak pie and chips you can save about 250 calories an apple instead of a chocolate bar, saving up to 200 calories a day, you only need to save about 100 calories a day for it to start to have an effect on your weight. Portion sizes count as well so if you think about the size of your plate and fill up more on the fruit and veg and cut down on the other portions it will make quite a big difference to your calorie intake, change the size of your plate to a smaller one, People that sit down and eat at the table together tend to eat less calories, compared to people that are on the go all the time. Eat regular meals, people who skip breakfast, put a lot more weight on than people who do eat it, eating slowly can also make a big difference

Powerpoint 17 something to think about & 18 ways to save 100 calories
19 shopping tips & 20 Food Labelling

Just a few simple tips for you which can make a big difference to families when they are looking at their eating patterns and what they do, one of the things people don't tend to do is they don't write out a shopping list and plan the menus for the week ahead, I am not saying you should plan every single meal, but what meal you are going to have at night time, you don't even have to say what day of the week you are going to have them, but say what things you need to make, so if you write a shopping list you are going save quite a bit of money when you go shopping, but you will also be able to plan what you are going to have, your portion sizes and buy what you actually need and not just impulse buying. Don't go shopping on an empty stomach, we have all done that I am sure, but then you get tempted by all the special offers, be careful with these and try not to buy more than you need, because the more food there is in the house the easier it is to give in to temptation and overeat. Food labels can be very confusing and people don't always know what foods to go for, if it says its low fat, is it good for you? Low fat products can often have a lot more sugar in.

Powerpoint 20 & 21

It doesn't take much to burn up to 100 calories, Eating a 100 calories a day more than you need can add up to 29000 calories in a year which can lead to people gaining up to 12 lbs in weight over a year. To burn off 100 calories you could jump on a skipping rope for 9 minutes, do a bit of gardening, a 15 minute walk, walk the dog for 20 minutes, wash the windows and so on. Has anyone tried using a pedometer, why not try and see what your normal everyday exercise is

and if you only do 2000 steps and then build up to doing an extra 1000 and try and build up from there it can actually make a big difference over a period of time. Why not park the car a bit further away and walk, use the stairs more than the lift, just trying to build more into your everyday life can make a big difference to your actual energy and expenditure

ANNUAL GENERAL MEETING

The Hilton Hotel, Northampton
Saturday 21st April 2006

1. Apologies for Absence
Lucy Jaques, Jill Jaques, Mr Ullman

2. Minutes of previous Annual General Meeting
The Minutes of the AGM on Saturday 9th April 2005 previously circulated, were agreed and signed.

3. Election of Officers
The current Officers, Phil Humphreys (Chairman), Julie Sales (Secretary), and Kevin Sales (Treasurer) were all eligible and willing to stand for re-election, and in the absence of any further nominations, were duly elected unopposed.

4. Election of Committee
The current committee members, Steve Burge, Graham Longly, Chris Humphreys, Tonia Hymers, Anne Crotty, Craig Barrass and Jackie Farrington were all eligible and willing to stand for re-election, and in the absence of any further nominations, were duly elected unopposed.

5. Chairman's Report.

The Chairman said "it had been an excellent year for the Society".

The Chairman announced that at the end of the 2005 AGM, Dr Philip Beales accepted our invitation to become President of the society. He then congratulated Dr Beales on his latest accolade when he was called to the 'Chair' in October 2005 and is now Professor Beales

The Chairman said we were honoured when in July 2005 Baroness Helena Kennedy of The Shaws QC accepted our invitation to become Patron of the Society. Baroness Kennedy is a distinguished Barrister and Life Per; she is also an acclaimed Public Speaker, Broadcaster, Television Presenter and Author. At present she is chairing the Human Genetics Commission which advises the Government on issues related to developments in genetic science.

The Chairman said that it was with regret that we had received the resignation of Robbie Hymers our Vice-Chairman. He thanked Robbie for his support as Chairman and Vice Chairman over the past 6 years.

The Chairman said that a nomination for this post had been received nominating Terry Crotty, who at present is a co-opted committee member, nominations were invited from the floor, there were none and Terry agreed to accept the post.

The Chairman said that it had been an extremely successful year for the Society thanks to the efforts of our Fund Raising Co-ordinator Anne Crotty, Members and Friends who have excelled themselves raising a total of £17,000.00. We have 4 new Friends and others have increased their giving and for this he thanked them.

The Chairman thanked Anne and Terry Crotty for their continued support attending Sight Village, to Adrian Stares for his support maintaining the website, although Adrian is unable to continue, we have been fortunate that Lance French has taken on this role and like Lance, with no charge to the society. As a result of this vital link we are now able to find out how many people access our site. To date we have over 500 visits. USA and UK are the top visitors, but people all over the world also access us.

The Chairman thanked Julie for maintaining this vital link along with her Secretarial, Childcare and Merchandise duties. He reminded the delegates that information can be downloaded, saving the Society a considerable amount of potage. The website is well worth a visit on www.lmbbs.org.uk

The Chairman thanked Chris Humphreys for managing the 'helpline' and the day to day e-mails, an increase in all areas from families and professionals in the UK and abroad, largely due to our successful website. The Chairman thanked Chris for her continued role of Conference Co-ordinator.

The Chairman thanked Tonia Hymers for the twice-yearly newsletters and Conference Report, which are available to read or download from our website, also available in audio cd. The Chairman reminded the members that without their snippets of information, stories and photographs, there would be no newsletter and asked that they contact Tonia direct

The Chairman referred to the figures in the Treasurer's report, which he stressed did not include the costs of this year's conference, and thanked the Treasurer for his continued tight rein on the spending of the Society.

The Chairman reiterated the comments of the Treasurer in his thanks to all those responsible for the fantastic efforts in fundraising and grants, enabling the Society to continue funding all those with the syndrome and making a generous subsidy for members attending conference 2006

To conclude, the Chairman reported that Conference2005 had seen an increase in delegates attending. The continued support of Professor Beales and his team, the sterling work of the committee and the attendance of a host of eminent speakers had once again made this event a tremendous success. He finished by saying that if anyone wished to have any involvement, or had any suggestions that would benefit the aims of the Society; he would be only too pleased to hear from them.

6. Treasurer's Report

The Treasurer reported that the accounts had been balanced for 2005 and audited and checked by Mr Thompson, the Accountant, who agreed with the figures. He said that the committee had again been very disappointed with the amount of interest paid by the Halifax account, this account has now been cancelled and the balance transferred to the Barclays Account. The two accounts are running smoothly, with the Friends Account increasing monthly balance.

The Treasurer said that in 2005 with the help of the Friends Account, we were able to transfer £1,300.00 to the Barclays Account which was used for the trip to Drayton Manor, so a big 'thank you' to our Friends. . He then reminded the members that Anne Crotty would be available all weekend to assist anyone who wished to become a 'Friend'.

The Treasurer gave thanks to Anne Crotty who managed to secure 4 grants totalling £7,500.00 of which £2,000.00 was received thanks to Jay Halligan. The grants were as follows

The Hilton Foundation	£2,000.00
Yapp Charitable Trust	£1,500.00
London Law Trust	£2,000.00
Hope 4 Children	£2,000.00

The Treasurer thanked all those who had worked tirelessly on fundraising. Donations and fundraising increased to just under £10,000.00 on last year. Making a financial gain of over £9,000.00 on last year.

The Treasurer said that as a result of the increase we have been able to substantially subsidise members for Conference 2006

7. Appointment of Auditor

The Chairman confirmed that Michael Thompson had kindly agreed to continue as Auditor for another year and he was duly re-appointed.

8. Any other business

The Chairman had received no written notification of any other business, but invited those present to raise any questions. In the absence of any such questions, the meeting was closed.

“More than Meets the Eye”

Personal perspective by Kathryn Murphy, Brisbane, Australia

More than Meets the Eye
Personal perspective by Kathryn Murphy

Bardet-Biedl Syndrome. I think the only way to describe it is life changing. When I first thought of talking about my experiences of Bardet-Biedl I wondered if you had plans of extending the conference, as so much has happened I could talk for hours, but I won’t. So far it has been 10 years of a never-ending roller coaster ride. On one of the society’s brochures it says “More than meets the Eye”’ there is possibly not one week goes by when I do not think of that saying. I feel it explains me so well in many ways. I use “More than meets the Eye” in two respects, which I think will give a good insight into my experiences of BBS

In the first instance “More than meets the Eye” is so important to the fact that although vision is a huge impact to the syndrome, it is not the only problem. My journey on the BBS road started out in August 1996 when I first heard the words “ Retinitis Pigmentosa”.

I think everyone here, who has been or who has had a child that has been diagnosed with RP and BBS would agree, that it is a harrowing experience. My experience was no different. From the day I heard about the possibility of RP, it took over 2 months to get a final diagnosis. My problem here, which has continued throughout the years, was that I didn’t fit the standard usual diagnosis for RP. It didn’t help that I had a Retina Specialist who thought he knew absolutely everything. To put it bluntly I challenged that and he didn’t like it. Part of the delay in diagnosis was the normal ERG result I had. This is the test that measures how the cells of the retina are responding to light and dark. Apart from this no true night blindness, I had the pigmentation on my retina that strongly suggested RP. I had the ERG test repeated and over a stressful 2 weeks wait, was to learn that thankfully it was still normal. With RP this result is abnormal. Eventually, however, I was given a diagnosis of Sector RP and Macular Dystrophy, like Macular Degeneration

For those of you who have not heard of the Sector form of RP it usually only affects one area of the retina. In my eyes I was extremely fortunate that the RP is in the inner most part of the retina, which corresponds to a loss of sight in the upper, outermost area of my sight. The remaining part of my retina is free of RP. My understanding of why my ERG is normal is that the area is so small in comparison to the normal area of retina that it

does not register. However if it were possible to do a test on just the area affected by RP, then it would be greatly abnormal

It took me a long time to understand how I could have RP with a normal ERG result, battling my Retina Specialist for some information on the Sector form of RP. Ten years ago all that was available was information on general RP. Finally to shut me up my specialist gave me some information. 150 photocopied pages from one of his Ophthalmology Medical Text Book on the chapter of RP, really appropriate. What he didn't expect was that I would read every word with the help of my Mosby's Medical Dictionary and from this it would lead me to self-diagnosing myself with Bardet-Biedl Syndrome.

While I was reading the information I came across the word "polydactyly" and was astounded to find out that it meant extra digits, just like the extra fingers I had been born with. I had one on each hand, but they had no bones and they were removed in the days after my birth. To this day I will never forget telling another doctor about my extra fingers saying "I know I don't have this condition "Beede Bardete" but I was born with an extra finger on each hand" From there it all snowballed and in October 1997 a Geneticist diagnosed me with Bardet-Biedl Syndrome

To this day I don't know why I asked so many questions, or needed so much information. At the time I think part of it was a coping mechanism, also I was in my first year of University studying Nursing, where we were constantly being taught the importance of the patient's right to be fully informed and I wasn't. Maybe it was the fact that the information I was finding just didn't fit with what I had been told by my specialist. Normally I am a person that rarely questions things, but I seem to question everything about BBS and what I am told. Sometimes I desperately wished I hadn't, but I know I wouldn't be where I am today if I hadn't. These days when a certain condition is in question I definitely research it, because I know how important it is to be aware of what the doctors are saying or not saying. Over the years I have seen an excessive number of doctors. I normally take along information on Bardet-Biedl and have to tell them what the syndrome is and then pay the doctor. I've always wondered if that was the right way round. Out of probably close to 30 doctors only 3 have known what BBS is, except for the Eye Specialists.

I don't know if it is different here in the UK, but in Australia BBS seems to be primarily seen as a serious genetic eye disorder with some other complications. Initially only Eye Specialist had any knowledge of the condition at all. I would have to say I have now educated many more doctors. I appreciate the emphasis that BBS attracts to the concern of the loss of sight. Because of the way I was diagnosed I had to deal with this for years, as the medical literature is alarming. No one explained to me until

some 3 years on that there can be great variations in the way individuals display conditions within the same genetic disorder. I had spent 3 years reading literature on the alarming and fast rates of blindness in BBS with the 3rd decade of life, from when I was the age of 19 to 21 I had read some BBS profiles and felt extremely fortunate and could tell I was a bit different, but at the back of my mind I was still scared stiff that I would go blind. It took another 2 years when I next had my eyes checked in 2002 to really relax about my vision, as once again there had been no deterioration from 1996. My vision component of BBS will always be there, but it would have to be the condition that now affects me the least. This year it is 10 years since I was diagnosed with RP. I still have no night blindness and continue to drive my car day and night. Every day I am thankful that there has been no change in 10 years. RP is a major condition and characteristic of BBS but what about all the other features?

No one is diagnosed with BBS just because of RP. When I was initially diagnosed it was on the basis of atypical RP, Macular dystrophy, Polydactly, Kidney disease that of reflux as a young child requiring surgery, slight webbing of 2 & 3 toes and weight difficulties. My poor Mum always got into trouble with the health nurse when I was a baby, because I had doubled my expected weight. Even by early 2002 I had encountered more troubles with my kidneys, experiencing sleep deprivation due to excessive nocturia, getting up to go to the toilet at night. Back in 1998 my kidneys suddenly one weekend had a significant concentration problem. This did not improve however since I have had a urinary concentrating deficit, as is talked of the BBS literature. As my Kidney Specialist of the time said, my kidneys work fine, they just leak a little too much. If you had met me in 2002 I probably would not have mentioned BBS, as once another doctor managed my nocturia BBS had no affect on my day-to-day life. I think I had to take 3 tablets.

Today 2006 Bardet-Biedl affects my day-to-day life everyday. Firstly I now swallow at least 23 tablets a day, but generally this does keep me feeling pretty good, and has improved my quality of life. This however not only introduces the merry-go-round of complex interactions between medications but what also of their long-term side effects, as I am sometimes reminded by some well meaning people. Without most of them I know I wouldn't get to a long term future because of the effect of complications such as high blood pressure, which remained uncontrolled until a year ago. My kidneys are now a constant daily problem as although they function normally, they are unable to regulate my water and salt levels. I regulate this by symptoms I feel such a headaches and leg cramps, indicating salt cravings and my weight, which indicates the degree of fluid I am retaining. I have just found out that apparently I actually have "mild diabetes insipidus". Confusingly I also require a diuretic to control my blood pressure as I retain fluid. This means a fine line in regards to salt

intake as it is salt intake that causes the fluid retention, yet it is necessary to have salt because as my doctor terms my kidneys are salt and water wasters. As I also put out more water fluid than I can usually intake. I am prone to migraines and the dehydration contributes to migraines. The migraines and any excess fluid are the primary cause to an increase in my blood pressure. Due to a lot of teasing as a child I am very sensitive about my weight, but I have to allow more salt into my diet now and accept being heavier for better health. This is something I am not doing very well with. Not only is it important to my kidneys and vision to keep my blood pressure controlled, but is also important for a condition I developed late 2004 called Coronary Artery Spasms. This is where the main artery supplying the heart with blood contracts to a degree causing chest pain, Angina. This is now well controlled with medication. After talking to Phil Beales, whether or not this is actually a condition from BBS is more unknown, but I do know that the physical strain of the dehydration, increases in blood pressure and fluid retention which goes hand in hand with the blood pressure which cause the coronary artery spasms. The effects of BBS definitely trigger the spasms and angina.

Just before leaving Australia I realised once again that I was having a problems retaining large amounts of fluid. Unfortunately it was too late to alter any medications and so I am carrying extra fluid mainly on my stomach and some in my face, hands and feet. I am also having increasing amounts of headaches and increasing intensity which feels exactly like blood pressure headaches. Somehow through the last few years I seem to have developed a fairly accurate sense of what is happening with my body and health. The hardest part has been getting the doctors to listen, especially when the symptoms haven't been of the normal description. Fortunately I now have 2 doctors who realise I have a very good in-depth ability of knowing my body which is helping all of us in my treatment.

Fatigue is the other huge problem that affects me daily. I have been diagnosed with Chronic Fatigue Syndrome, but that was even before most of these medical problems began. It is possibly now just chronic fatigue due to chronic illness. I also think that the way my kidneys work is also contributing to this, as everyday they lose excess salts and water which as I acknowledge through symptoms I have to replace through my diet and fluids. It is a constant never ending cycle. I also have another condition call Fibromyalgia, which is also a fatigue condition involving joints and muscles.

Aside from the medical conditions of BBS there are the emotional and social complications. I think this is where I tend to think of the phrase "More than meets the Eye" the most. I know how fortunate I am in the way I am affected by BBS but sometimes it can be a double edge sword. People

have no idea that I have a genetic syndrome with all the medical consequences that I experience every day. Don't think that this doesn't include the medical professions. When I see a new Doctor for the first time it is like opening up a can of worms when they say so "have you got any medical history"? Would you believe I've then had some doctors basically saying I wanted to have BBS as they strongly doubted the diagnosis? I wish. Thanks to Phil Beales this is over as I now have genetic proof from DNA testing. I know that I have the BBS 1 gene. For myself to have the results of DNA testing is great because now no doctor can doubt my condition, not that I can even believe there would be anyone wanting to have such a condition. But even when I received the results I was bewildered for a few days. Prior to receiving the news I had been saying to family, that if nothing showed up, it probably just meant I had one of the other genes for BBS which in 2003 had not been found, since I have a somewhat milder form of BBS than most. That is how sure I was I had BBS. There was no doubt. However when those results came it was there in black and white. BBS1 gene mutations, there was no going back. I think more than anything what bewildered me the most was that I had no idea in my subconscious that I had the tiniest hope that I didn't have BBS. I think it is important for anyone considering DNA testing to realise that it doesn't change any of the characteristics of the condition. It will only confirm if they can find the relevant genes causing the condition. I think if you or your child has the relevant number of characteristics to be initially diagnosed with BBS it is likely that in time one of the genes will be able to be found. Once the genes are found there is no turning back, you can no longer hope that maybe the Doctors have got it wrong because the diagnosis will never change and you have to be prepared for this.

Despite the diagnosis, even way back in 1997 I never considered changing my career decision to be a nurse. It was the only career I had wanted to do since I was about 15. I was warned never to disclose my diagnosis of BBS more importantly the RP due to the risk of discrimination. I was always scared someone would find out and then there was always part of me that wanted to let people know the real me, what I was trying to deal with. Instead I kept it all to myself and never really fitted in at the hospital I worked at. During the years when I thought RP was going to cause major deterioration to my sight, there were also the worries and tears about what I would be able to do when I couldn't see well enough to nurse. After many changes to my hours and positions I finally had to leave nursing late 2002 due to my physical health. I then worked for 2 years, 12 hours a week as a medical receptionist but in the last year could not be relied upon as my health was extremely unstable in 2004. For the last 18 months I have not worked and have had the best health I've had in years. Recently I completed some work experience working as a nurse in a doctor's surgery for a few hours two or three days a week. I enjoyed this and managed the work as it is much less physical than a hospital. Hopefully when I return to

Australia if someone will employ me for 2, 4 hour days, in this capacity I will be able to return to nursing.

Although because I receive a disability pension after much red tape and endless paperwork, I am not obliged to find work, I think it is important for me. The hardest non-medical aspect I find about BBS is how socially isolating it is. It would really great to have weekly contact with other people and to be using my knowledge and skills which would also be beneficial to my confidence. This is something that is lacking in my nursing since it is close to 4 years that I stopped. As there is not much financial benefit to me working it is important that I do not work more than is comfortable to maintain a good balance between work and some social life. That usually does not leave much time when you also factor in doctor appointments as well, plus an afternoon sleep which is needed most days. This generally does not leave a great amount of time for social occasions and generally by night time I am exhausted and too tired to go out or if I do I am home by 10pm. This generally I not the life of a 24 to a now 28 year old. Generally my social circle rest heavily on my parents and some of their friends. I have one good friend in Brisbane who I see most weeks but unfortunately my very small social circle is made even smaller as I now live 2 ½ hours from one good friend and 1 ½ hours from another. Also my sister and her husband live in Melbourne which is a 2 hour plane trip. She fortunately is not affected by BBS. BBS truly shows you the people who are your true friends and care. Even then I know at times they don't want to ask the words "How have you been" because normally something else has happened, some new medical drama.

Meeting new people and getting to know people when you have a condition like BBS definitely provides many challenges. To this day I never know how or when I should tell someone about my condition, as in my case it can go unknown. As I want everyone to know about BBS I find hiding my condition conflicts this however, at the same time I want people to get to know Kathryn first. So I usually take this option. I have found that if I introduce BBS from the beginning it becomes, "oh poor Kathryn with BBS;" or in most cases this is too overwhelming. "I don't want to know" and people then generally take the easy option and shy away. Initially this was hard to understand, but if I hadn't been through what I have, I would be intimidated by someone telling that they have all this medical stuff. What you do say or what should you do? Just be a friend, be there for when you need to talk, a shoulder to cry on when needed, learn to work around any medical needs and just be there. I know that's all I've ever wanted, not wonderful well meaning words that do nothing. I know people get overwhelmed by the condition and it is not surprising because it is. I no longer blame people running from knowing about BBS because truly if I had the chance I would have run many times over. I sure most of you would, we just can't.

Although I am fortunate that people do not realise that I have any sort of genetic or medical condition, I often feel out of place in situations and misunderstood. People don't realise that there is "More than meets the Eye". Because of this as I described earlier I have never had the normal social life of a young adult. I know that I am quite a serious person and know myself well and that is because of my BBS and its increasing effect on my life. I tend to intimidate people of my own age which I don't realise because up until now they have tended not to have the same assurance of themselves because they have not been through the roller coaster ride that I have. Sometimes my Mum describes me as "an old head on young shoulders" For many years I have felt older than my age and in general, normally find it easier to socialise and communicate with older people than that of my own age.

I also hope that one day I will have that "someone special" in my life. Unfortunately I have already had my worst fears realised having BBS used as an excuse by a guy. My other dream for the future, something I don't look too far ahead is to have a child. This is something I have no idea if it will ever be possible. This is because some days this body is just so tired and so many of the medications that keep me fairly well would need to be stopped or changed. That is just for starters. What of the genetic risks? Even in the best possible scenario the child would carry one BBS affected gene. Every year this dream seems to be slipping further and further way. Every year for the past 3 years I have wanted somehow to be a voice for BBS in Australia, start to at least get the government and the medical profession aware of the condition. 2006 is here and still this is yet to occur partly because I don't know where or how to start and also I have been unwell. I read the publications from the Society and wish Australia could be doing something like this as. My dream is to have an Australian society for BBS sufferers. From what I can see of the LMBBS Society most of the input here is being done by the parents who have children affected by BBS. However I feel that with all my knowledge from my nursing I should be doing this. I not only feel I should, but I also want to because I strongly believe that every person has a purpose for their life. I do not believe my nursing degree was completed in vain as because of that training I have gained a much greater understanding of the working of the body and its systems, which constantly assists me with my health and Bardet-Biedl. It is because of this understanding that I feel I am an ideal candidate to educate people on the condition. Unfortunately because I have BBS myself and my consequent health. I am not even the best e-mail respondent

Looking to the future is hard and something as I said earlier I try not to do too much as that is when I can quickly become overwhelmed. Generally I try to live my life day to day and I am getting much better about not worrying so much about the day after. I do have hopes for the future

though and that is not just because of the wonderful research and developments that are happening on this side of the world with BBS. It is because I am a Christian and believe in God. Some of you may not believe and some of you may. But I do and by being a Christian, God does not promise that everything will be wonderful in life, there will be times of trials, but with him you can overcome them. It is going through these times that you develop your perseverance, character and hope. It is said in the Bible "For I know the plans I have for you" declares the Lord "plans to prosper you and not to harm you, plans to give you hope and a future" Many times I haven't wanted the situations that I have been through, especially with BBS, but each time I got through and now I am grateful for the person that I have become because of the experience, for how much I have grown and no longer take life for granted. There is another particular verse from the Bible which helped me to come to terms with my BBS diagnosis which I will share as hopefully, maybe, it can also be of some benefit to those of you who do not know God. This is in Psalms

"For you created my inmost being, you knit me in my mother's womb. I praise you because I am fearfully and wonderfully made; your works are wonderful,…. When I was woven together in the depths of the earth your eyes saw my unformed body. All the days ordained for me were written in your book before one of them came to be"

For the first time in years I felt special. I had been made this way for a reason and who knows why. That I certainly don't, but I am wonderfully made. My body might be breaking down but it is wonderfully made and it is worthwhile, as to I am worthwhile.

In closing I just wanted to read some words of a song that just touched my heart when I heard it. After this please feel free to ask my any questions. This song is written by an artist called Roma Waterman. It so fittingly describes my BBS roller coaster ride

If you asked me how I'm feeling, I'm broke to pieces, now I'm healing
If you wonder how I get through, there's something stronger than me and you
As I look back I keep smiling, coz through the heartache I kept fighting
But I am not perfect, someday I fall, but I don't give up through it all
There has been a kiss of grace on my weary teary stained face……

The Chairman responded to Kathryn –"Kathryn, I think every mile you

have travelled has been worth it just to listen to you this afternoon. You have been an inspiration and I am sure that everyone here will agree. We all hope that you will realise your dream and we are here for you. If we can assist you in any way, please let us know. Thank you

Closing Remarks

The Chairman said he hoped that everyone had enjoyed the conference and had found it not only informative but interesting as well. Without mentioning individuals for fear of missing someone out, the Chairman thanked Phil Beales and his team, each and every speaker, the hardworking committee, but most of all the delegates for attending and making this such a successful conference. The Chairman assured members that their committee would be working hard in the coming months to ensure another successful conference in 2007

Thank you all for attending

Dates for your diary 20/21/22nd April 2007

Feedback

The delegates were asked for their comments regarding…

The Saturday morning programme

"Excellent – "Good balance of interesting and informative talks" "The ability of the speakers to communicate with the layperson is excellent" "Helen's presentation made a picture that was easy to understand" "Amazing how much I learned" "Good variety of speakers"

The Saturday afternoon programme – Workshops

“Kathryn’s perspective was brilliant, so moving” very emotive perspective from Kathryn” “what a brave lady” “All the workshops were brilliant” “So much information from the workshops” “Excellent – it gave us a chance to find out more about each subject in face to face talk” “All of them, they were excellent and very helpful” “Not enough time, I would like to have gone to all of them”

The AGM

“Short and to the point” Could benefit by being longer” “what a short one, well impressed” “efficient” “Short and sweet” “Fantastic, wish all AGM’s were as speedy”.

Volunteers

“Where do you find them, they really are the unsung heroes of the conference” “Cannot appreciate enough, the time and care the carers gave to look after our children” “Wonderful, had a fab day out, must make carers go on higher, faster and wetter rides”

Meals

“Tremendous”” “Can’t fault the food at all” “Friday evening and Saturday lunch excellent. “Chef is a scream” “Breakfast could be better if not cooked in such large quantities and left on hot plates” Excellent, look forward to next year” “Can’t fault them”

Accommodation

“Fantastic” “Excellent” “Staff excellent, always more than pleased to help” “Always good clean and comfortable” “All you need in comfort” “Very good, staff very pleasant and courteous” “ A good standard with family environment”

Overall organisation

“Excellent, everything very well done and precision timing” “Thanks for taking the time to meet and greet personally”. Congratulations to the committee members for a very successful programme” “Fantastic and well executed” “Well organized and executed”

Suggestions for next year

“Make it a week long conference” “More of the same” “Workshops running for the weekend on Diet, encouraging/supporting weight self management in adolescent/post adolescent LMBBS,” “Coping with visual impairment”

"Benefits" " Visual Aids", "Tips on day to day coping with the syndrome" "Renal" "Orthopaedic"

Comments received from DVD

"Great" "Brilliant" "Brought back memories of a brilliant weekend" "Great, the Grandparents loved it" "Having watched the bits with us in it, delight and embarrassment, depending on age" "Memories for the year of a fab weekend. Thanks"

Young Members Report

Hollie Sales + Chloe Maclean - Cub Conference Reporters

Saturday Morning

Chloe and I woke up at 6: 20 am; we talked about Drayton Manor for 10 minutes. We decided that we couldn't get back to sleep so we got dressed and went downstairs and waited for breakfast .For breakfast we had: a bowl of cornflakes, choc-chip muffin and bacon and egg .After breakfast everyone started to arrive for the coaches to go to Drayton Manor, they were all very excited. We went on the water rides, sombreros, roller coaster and many more .We got back to the Hilton hotel at 5:20 pm. After our dinner we watched Nanny Mcphee with the carers and other children and then at 9 o clock we did our Fashion Show, Alex sang a song at the end, it was great and more than 80 people came to watch us. When we did this at home last year we raised £395.00 for LMBBS

I found Drayton Manor really exciting. I liked the people I went round with .The rides were so cool and wicked; I can't wait until next year. Maybe Chloe and I will arrange another show as everyone said it just was so fantastic and well done. We both felt very nervous about doing our show.

So thank you to every one for watching and supporting us.

A Carer's Personal perspective
Sarah Butler

LMBBS, what was that? My husband and I didn't have a clue. IVF was all we

knew about, and the long 2 years we had struggling to get our Florence. She came naturally in the end, all that money spent!! It's only money! During my journey through IVF I met this lady called Julie Sales in hospital. She was so nice, we made friends real quickly. Same silly sense of humour. She had two lovely girls, Hollie and Danielle. After we both came out of hospital we spent time together and grew into great friends.

After Craig and I learned more about LMBBS, we felt that we would like to become carers, even though it was a challenge for us. So, 4 years ago we came to our first LMBBS conference as carers. Craig felt very nervous about it all. Me!! Well, I just took it all in my stride.

We arrived to a beautiful hotel, nice room and very friendly people. I was speechless, here we were, amongst these lovely people and every one of them a different story to tell. Craig and I had a little girl called Chloe; she was bubbly, fun and full of energy. We visited Thorpe Park. The bus journey was long but everyone was full of spirit. We took Chloe on the boat across the water. She stood up and screamed in absolute excitement that she was going to the farm.

I cried. The feeling for me was so amazing, this little girl touched my heart – she was living with this disability and for one moment she forgot
and the excitement took over. I was lost for words all weekend. I met blind people who had done amazing things and to me they were inspirational.

We returned home, I sat and cried not tears of sadness, but admiration for the families I had met.

These families live day to day lives, just like us, and as they had said to me "you just get on with it". These to me are special people. Just like Florence is to us, after this weekend I felt I had added to my family, extended it. I wanted to stand on the tallest building and tell the world how special they all are.

Every day I wake, I get a huge feeling of happiness knowing that my husband and I can contribute to helping these families in any way we can. It's so easy being on the outside looking in, but for Craig and me we no longer stand on the outside. We feel that these families belong in our lives. I just hope that we can express and care enough for them to know that.

The conference has changed me as an individual. I am more patient with my children and more understanding of their needs. Julie, Kevin and all the families we have met have taught me that.

This is our fourth conference and our feelings haven't changed Thank you everyone for allowing us into your lives.