



# LMBBS Winter Newsletter 2010



Welcome to the LMBBS Winter Newsletter



## Announcements

Congratulations to Helen (May-Simera) and Chris who eloped in October. Helen says they 'married under a tree in gorgeous sunshine in beautiful Vermont, then stayed on for a few days, in a hotel with amazing views, did lots of hiking and ate fabulous food'. We are thrilled for them both and wish them every happiness in the future.

## The Saga of the BBC Radio 4 Appeal – Anne Crotty

**BBC  
RADIO**



92–95 FM

In September 2009, we received a letter informing us that we had been granted a BBC Radio 4 Appeal. I could hardly believe it and was smiling for the rest of the day. This is the story of how it all happened.

The whole process took over a year and was beset by minor hiccups from the very beginning. I wasn't very hopeful of success because we rarely meet the criteria required by big organizations. We asked to be considered for a BBC 1 Lifeline and Radio 4 Appeal and, if possible, for it to coincide with our "Big S" 'Summer Sundae' fundraising initiative.

On 3<sup>rd</sup> July, I received an e-mail asking for the final page of the application to be re-submitted as the signature was dated 2007! The request by Christina Roski was really friendly and understanding and this set the tone of all my dealings with the BBC staff. Following the fantastic news in September that our application had been successful, in December, we heard that we had been allocated an appeal slot on 27<sup>th</sup> June, 2010 and we were invited to attend an appeal briefing in January 2010, at Broadcasting House.

In early January, the venue for the briefing changed to the BBC Television Centre and Terry and I duly travelled to White City, where we met several other people like us from small groups. We met Sally Flatman, the producer, and

other members of the team and received comprehensive information about the whole process, including a 3-month count-down.

We knew that, for a Radio 4 appeal, we would have to write our own script and find a presenter to read it. One of the first items on our count-down list was to consider who to approach regarding presenting our script and, after consulting with the committee, I sent a list of eight names to Sally Flatman.

I did not receive a response from Sally but assumed that this was because our appeal was not due to go out for some time; however it turned out that the email address was incorrect. Fortunately, in the meantime, we had started work on our script. Tonia, Allan and I each wrote a script and then I amalgamated them into one and sent it to Sally. We had now lost a lot of time and began a period of almost daily e-mails and telephone calls, discussing suitable presenters and tweaking the script, bearing in mind that we were appealing to Radio 4 listeners, a very special breed! I was immensely grateful to Sally and team who helped me find contact numbers for agents of suitable presenters, none of whom, however, were able to help.

In the end, we were less than four weeks away from the date of the Appeal and needed to book a studio. I was beginning to get a bit panicky about it all and it was in my thoughts by day and night! After some discussion with Sally and an OK from the committee, at the beginning of June, it was decided that I would be the presenter and we adjusted the script accordingly. I began to sleep a bit better but developed a sinking feeling in my stomach that stayed there until it was all over. In the middle of all this, I was trying to think about publicity and, once the presenter had been decided, liaised with Karen Scott of Capita, who organized the telephone and online donations, some letters and postal donations on our behalf. I was also trying to obtain suitable pictures for the Radio 4 website and was still constantly adjusting the script. I was delighted to receive an e-mail from Sally on 9<sup>th</sup> June, marked FINAL script; the Studio was booked for the 15<sup>th</sup> June.

The dreaded day dawned; Tonia travelled to London from Essex and Terry and I travelled up from Devon, where we were on holiday. We set off for Tiverton at 10am, allowing plenty of time to get to London and spend a little time with Tonia before the recording. We had a 35 minute drive to the station and when we got there, the car park was full! We had to retrace our 'steps' to find the overflow car park, where the ticket machines were out of order! We walked to the station and enquired about paying for the car park, only to be given a telephone number. I then had to talk to a machine and pay for a virtual ticket by card, something I have never done before. Fortunately, it worked; we bought our rail tickets and had a little time to recover from the first trauma of the day before the train arrived.

Once in the train, we sat back to enjoy the journey, got to Taunton and stopped there for an hour! Another train had jumped a red light which called a halt to all train movements from that station. An announcement came over the tannoy, 'Is there a doctor or a nurse on the train?' Fortunately, a nurse came forward. I had been retired for far too long to feel that I could help. The train manager and staff were fantastic, keeping us regularly informed and giving the passengers free tea and coffee. We explained that we needed to be at the BBC by 3pm because of studio time and they found out where we had to go to get a taxi and roughly how long it would take. I kept in touch with Tonia by text and spoke to the Studio on the phone. It looked as if we should be OK. When we arrived at Paddington, however, there was an enormous queue for taxis. Our stress levels were building! We arrived at Broadcasting House with minutes to spare and were greeted by Tonia. Panic over! We waited in the lobby to be escorted to the studio.

Sally and Ruth had managed to extend our studio time so we were able to go ahead without feeling rushed. It was a most interesting experience. The tiny studio was divided into two by a large glass window. I had to sit by myself with the microphone on one side of the glass, while everyone else squeezed in with Steve, the sound engineer, on the other side. The script was tweaked several times more and we did several 'takes'. After that, Tonia went to the microphone and was interviewed by Sally for Facebook. She did really well as this was unplanned and she didn't know what Sally was going to ask her. We were encouraged to take photographs and Sally and Ruth behaved as if they had all the time in the world. As we left Broadcasting house, Tonia saw Gary Kemp, she is sure he winked at her!



Once outside, we said our goodbyes and Terry and I headed for Paddington. Our journey back to Tiverton was uneventful and our walk back to the car park took us past a lonely fisherman in the dark. Very peaceful! We drove out of the car park in the direction that we had come, only to find that the road was closed and we were diverted into the wilds of the Devon countryside; the day had ended as it had begun! I have been very humbled by the generosity of those who donated to the Appeal. Many did so anonymously, many wanted no acknowledgement, some sent moving stories of their own and some sent humorous messages. Many said how moved they were by the appeal; I have been moved by their response. Apart from the letters of thanks, I have claimed the Gift Aid and

kept records required by the BBC for their Monitoring and Feedback form, which has duly been completed and returned.

I cannot speak highly enough of all the staff at the BBC. During the most frenetic period of the process, Sally and team were dealing with many charities as an appeal is broadcast every week; however, we were always made to feel as if ours was the only one that mattered. They were consistently understanding, good-humoured and helpful, gently guiding me in the right direction. The 'final product' was excellent and to hear Tonia on Audiobook, on Facebook, was the icing on the cake.

## LMBBS Radio 4 Script



For those of you who missed the Radio 4 Appeal, here is the final script.

'It was dawn on a North Essex beach. The sea was calm and rabbits were hopping about among the colourful beach huts. In a nearby cottage hospital, a baby with red hair, called Joe, was born. His excited parents examined their beautiful little boy from head to toe. They were concerned to discover an extra digit on his left hand but the midwives reassured them that this could easily be removed at a later date.

Little did everyone know that this tiny little finger was the outward sign of a rare condition called the Laurence-Moon-Bardet-Biedl Syndrome, or LMBBS.

Joe's mother discovered the full facts of this syndrome while in the library, with Joe watching innocently from his pushchair... and sobbed. Tests had already indicated kidney disease, hypertension, obesity and delayed development. Diagnosis was finally confirmed when Joe was 10 months old and, soon after, the family were told the devastating news that he had a progressive eye condition that would lead to blindness.

Joe is my grandchild.

Luckily, Joe's parents soon discovered the Laurence-Moon-Bardet-Biedl Society, a support group for those with the syndrome, their families and carers.

The family were invited to the LMBBS Family Weekend and Conference. That was 12 years ago - they haven't missed one since.

The Family Weekend provides a stimulating break for parents and a fun-filled time for the children, who, for once in their lives, don't feel 'different'. Joe's Mum says, 'it feels like going home.....for just one weekend a year, we find true acceptance and peace.'

I now volunteer as the charity's fundraising co-ordinator and raising money for this conference is a big challenge. But, if a thousand people were to donate £20, I can tell you - next year's weekend would be safe. So please help us to continue providing these very special weekends for Joe and all the families who need them... () ... thank you.'



Building a brighter future for young people with complex needs

RNIB Rushton School and Children's Home, based in Coventry, is being transformed to provide state-of-the-art purpose-built facilities for children with complex needs and sight loss from all over the UK.

RNIB Rushton offers specialist education, year-round residential care, tailored therapies and one-to-one health support for young people aged up to 19 years old who are blind or partially sighted with multiple, complex needs. The new facilities, due for completion next year, will double the current number of places available and will include new school amenities with specialist sensory environments, music rooms, bungalows with outdoor play areas and a water therapy suite.

Sue Card's son Tom attends Rushton. Sue said, "The expertise and care Tom has received at the school and home is exemplary. The support he has received has helped him to develop at so many levels - in ways I would never have thought possible."

Mark Sanderson, Director of RNIB Rushton School and Children's Home, said: "We are at an incredibly exciting stage of the development. The new facility will place RNIB at the forefront of care and education providers for young people with complex needs and sight loss, and will set the standard for other facilities around the world.

Our exciting redevelopment builds on the firm foundations of our strong heritage and will enable more young people to reach their full potential for learning, independence and fulfilment in everyday life."

To request an information pack or arrange to visit, contact Liz Gutteridge on 024 7636 9531, email [liz.gutteridge@rnib.org.uk](mailto:liz.gutteridge@rnib.org.uk), or, for more information, visit [www.rnib.co.uk](http://www.rnib.co.uk).

## Taxicard & Capital Call

If you live in London, are mobility impaired and have trouble using public transport, you may be able to use subsidised taxi and minicab travel in London. The two schemes are called Taxicard and Capital Call. You can use Taxicard wherever you live in London and can join Capital Call as well if you live in Bexley, Ealing, Enfield, Haringey, Hillingdon, Hounslow, Lambeth, Lewisham, Merton and Southwark.

Who is eligible?

You may be eligible for both Taxicard and Capital Call if you:

- Receive the higher rate mobility component of the Disability Living Allowance or the higher rate Attendance Allowance
- Are registered blind
- Receive the War Pension Mobility Component

If none of these apply to you, you may still be eligible if:

- Your GP endorses your application. You may have to have a mobility assessment



## Taxicard

You generally pay a flat fare of £1.50 per trip, plus anything above the subsidised amount.

In most boroughs, Taxicard trips are subsidised up to:

- £10.30 for trips during the day
- £11.30 for trips at weekends
- £12.80 for trips at night

## How to apply

Contact 020 7934 9791 or visit the website [www.taxicard.org.uk](http://www.taxicard.org.uk).

## Capital Call

Telephone Capital Call and the operator will book a minicab for you. Not all licensed minicabs work with Capital Call, so, to receive the subsidised fares, you must book through Capital Call.

You will be given an annual travel budget of £200, to spend how you wish on Capital Call trips, booked through the call centre. You must pay the first £1.50 of any journey, and then your contribution increases as the value of the journey goes up. Once your budget is used up, you will have to wait until it is topped up again the following April before making any more trips. Your Capital Call operator will be able to tell you how much you have left to spend at any time.

For further information, telephone Capital Call on 020 7275 2446

Email: [capitalcall@hackneyct.org](mailto:capitalcall@hackneyct.org)

You need to be a member of Taxicard to join Capital Call.



LMBBS Clinics

## Healthy Eating

Weight management provides quite a challenge to those with LMBBS and is an aspect of the Syndrome that is addressed at the new BBS Clinics. Sarah Flack is the dietician at Great Ormond Street Hospital for Children, one of the venues where the clinics are being held. Sarah has kindly agreed to become a regular contributor to the newsletter, with articles promoting good diet management.

## Introduction

My name is Sarah Flack and those of you who have been to the new LMBBS clinics at Great Ormond Street Hospital will already have met me as your dietitian. For those who will come to the GOSH clinics in the future I thought it might be useful to know a little about me. I've worked at GOSH for 14 years and have covered a variety of clinical areas. Up until two years ago I specialised in looking after children with kidney problems and in this role saw children with LMBBS. I have two children, aged two and six, and so now work part time. I am really enjoying being involved with the new multi-disciplinary BBS clinics.

## The Dietitian's Role in the one-stop BBS Clinics

Each of the 4 annual clinics offers an appointment with a dietitian. At GOSH you will be asked to complete a food diary before/after clinic. This is used to give a greater insight into what is usually eaten at home. This allows recommendations to be made to ensure your child receives the optimum diet that is well balanced and nutritionally adequate, providing the right energy intake for growth whilst controlling their weight gain.

There is a lunchtime session which is a chance to sample a healthy buffet lunch and chat with other families, BBS support workers and me to share ideas or ask questions.

## Healthy Lunch Boxes

It is only a month into the new school term and already making my daughter's packed lunch has become a bit of a job. If I and many of the parents at school already see it as a chore, does this make it hard to provide an interesting, well balanced and varied lunch? My daughter likes to talk to me about her lunch every day and also all the foods that her friends have in their lunch box. Interestingly she never mentions fruits or vegetables - just the chocolate biscuits, crisps, savoury snacks and cakes! So why am I worried about what is in our children's lunch boxes? Contrary to what you may think, studies have shown that packed lunches aren't necessarily a healthier alternative to school dinners.

You may have read the newspaper stories earlier this year following the publication of one of these studies – the headlines were scary: '99% junk in kids' packed lunches' and '99 out of 100 packed lunches eaten at school are unhealthy'.



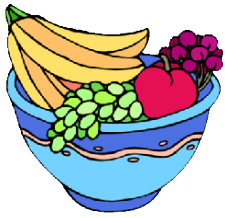
It has been shown that children eating packed lunches eat approximately double the amount of sugar and 50% more saturated fat in their lunch than in the school meal. Foods contributing to this higher fat and sugar intake included crisps and savoury snacks (found in more than half of all the surveyed packed lunches), chocolate confectionary, full coated chocolate biscuits, cakes and biscuits. On a positive note more than half of children eating a packed lunch ate fruit. However only a very small number of children had a vegetable included in their lunch. If you are reading this thinking that you already include fruit and vegetables daily and limit 'treat' foods - congratulations! If you are keen for ideas on how to fill up your child – the best foods to include more of are those with the lowest energy density, for example, low fat choices and foods with a high water content. If you think your lunch boxes could be improved read on for some ideas.

All meals need to be based around starchy carbohydrate foods which slowly release energy for the body to use. If you can, choose high fibre varieties (for example wholemeal bread or wholemeal chapattis) - the fibre bulks up the food but cannot be fully digested and helps you feel full for longer. Using a different type of bread each day can make sandwiches more interesting. Why not try multigrain and seed rolls, bagels, baguettes, pitta breads, chapattis, wraps...the list is endless!



Think about what you put in the sandwich. Do you need to use margarine/butter or a low fat spread? The answer is no if the filling is moist e.g. cottage cheese, egg mayonnaise (low fat mayo!) Try to think of lower fat alternatives to your current sandwich fillings e.g.

Current sandwich	Healthier options
Sliced Cheddar cheese with butter thickly spread on the bread	Grate the cheese – this will reduce the amount used Try a reduced or half fat cheese – the difference may not even be noticed Add a vegetable to help bulk out the sandwich so you can use less cheese e.g. tomato or cucumber Use a scrape of reduced fat spread
Ham - 2 thick slices with the fat left on made with full fat margarine spread on the bread	Choose 1 slice lean ham and remove any visible fat Use a scrape of reduced fat spread Add salad or cucumber
Tuna in oil mixed with mayonnaise and margarine spread on the bread	Choose tuna in water or brine Mix with reduced fat mayonnaise Add sweetcorn or cucumber No spread required on the bread as this is a moist filling



We all know that we should be eating five portions of fruit and vegetables each day but what does this actually mean? I am often asked what size a child's portion should be – a simple rule is as much as they can hold in their hand. Remember it is best if five different types of fruits or vegetables are eaten each day. To ensure your child has 'five-a-day' at least two portions need to be included in the packed lunch. Try including one vegetable e.g. raw carrot, cucumber or celery sticks. A low fat dip could be included too - salsa is very low in fat and a great way of boosting vegetable intake. For the fruit, try including at least one portion. If your child isn't that keen on fruit, try making a fresh fruit kebab or fruit bags – it looks colourful and enticing to eat. Use ready peeled orange pieces, grapes, blueberries, strawberries etc.

Water or sugar free squash are the best choices for drinks. It is best to avoid all sugary fizzy drinks. Fruit juice is also best avoided as this too will increase your child's energy intake. Your child will get adequate vitamin C by eating five different fruits and vegetables each day.



The snack food market is huge and very profitable for the manufacturers. It can be very difficult as a parent to decide which foods are the healthiest choices, due to clever marketing, and packaging often attracts children's attention. Not all low fat or low sugar snack foods are a good choice. Often the energy content of these products is very similar to the original food as they have extra sugar added if the fat is removed or vice versa. Some healthy food choices become less so when they are coated in yoghurt or chocolate for example. It may surprise you but there can be more fat in yoghurt coated dried fruit snacks than in the same snack covered in chocolate.

Savoury snacks are a minefield too and should be kept for treats. Low fat crisps can be just 15 kcals less than a packet of standard crisps, which is a very minimal saving. Look instead for baked potato style snacks or corn based snacks e.g. Quavers or Skips to make a greater energy (calorie) saving. Buying 'diet' type brands needs to be an individual family's decision - the argument against using these products is that your child develops a taste for these unhealthier foods, possibly choosing these over healthier options. My personal preference is to use the regular 'treat food' as a treat. I have found it useful to discuss when treat foods will be allowed in my daughter's lunchbox. We both now know that crisps will be allowed as a treat on a school trip day only. There may be a treat in her lunchbox on days when she is particularly active. On a day when she does a PE lesson and after school sports, she may find a small square of flapjack added to her lunch. Alternative snacks could be a currant bun, plain biscuit, chewy cereal bar or a fun-sized bar of chocolate.

For other ideas or extra information please contact your clinic dietitian or look at the following websites:

Food Standards Agency: [www.eatwell.gov.uk/healthydiet](http://www.eatwell.gov.uk/healthydiet)

British Dietetic Association: [www.bda.uk.com/foodfacts](http://www.bda.uk.com/foodfacts)

Department of Health: [www.nhs.uk/Change4Life](http://www.nhs.uk/Change4Life) and [www.nhs.uk/livewell/5aday](http://www.nhs.uk/livewell/5aday)

Sarah Flack  
Principal Dietitian  
Great Ormond Street Hospital  
Contact details: 0207 405 9200 extn 5941 (Thursday & Friday)  
October, 2010

### BBS Clinical Nurse Specialist

Hello, my name is Kath Sparks and I have been appointed as Clinical Nurse Specialist (CNS) in Bardet-Biedl syndrome for Great Ormond Street Hospital. I also help run the adult clinics in Guys Hospital. So far I have been involved with six clinics, three in each trust, and it has been lovely to meet some of the families affected by Bardet-Biedl syndrome.

I trained as a children's nurse at Great Ormond Street Hospital over ten years ago and have had experience in various fields during that time. I began looking into ways in which bloods were taken from children a few years ago and, with my previous colleagues, endeavoured to change the way blood tests were taken. Our aim was to minimize any trauma the child felt and to alleviate pain during the procedure so that the next time blood tests were needed, the child didn't feel scared or as scared. I have used local anaesthetic and distraction techniques on the children I

have seen in the clinics and offered local anaesthetic to some of the clients in the adult clinics. I feel it is very important to give the children a positive experience so they don't become phobic of blood tests, as this is a fundamental way of monitoring their condition. I try and let patients have some control over where and when their tests are performed as I believe this helps them feel less stressed during their clinic appointments. I will always have time prior to taking the tests to talk to the patients/parents regarding the need for the tests and the ability to obtain them.

My role is both exciting and challenging as it involves providing a service for people and families, where I am a key member of a team looking after their needs. This role involves providing support for patients and families and liaising with other multi-disciplinary teams involved with the care of those affected, both within GOSH and in the community. This also includes working closely with support groups and ensuring families are well informed of all services available to them. Their support and welcome for me has been overwhelming.

Kath Sparks  
CNS Bardet-Biedl  
[sparkk@gosh.nhs.co.uk](mailto:sparkk@gosh.nhs.co.uk)

If you or your child are concerned about having blood taken and you feel it would be beneficial to discuss this, or any other aspect of clinic attendance, prior to attending an appointment, please don't hesitate to contact Kath or one of the BBS Support Workers; we are happy to help.

## Fundraising Roundup

Once more, you have been generously giving your time, energy and money to help your Society.

The 'Friends of LMBBS' are donating regularly and generously and, with Gift Aid, their annual giving is bringing in over £3,500. If anyone else would like to become a 'Friend', please contact me. Many of you are saving coins and filling collection boxes. We have received very welcome donations; the donors include Waitrose, the Basingstoke Townswomen's Guild, the Students of Tranquil Space Bethesda Yoga class in America, William Grant and Sons (thanks to a nomination by Andrew Ramsey) and some donations have been made in memory of loved ones. I wish I could name every one of our faithful supporters – members, family and friends – but space won't allow it. You know who you are and rest assured that every penny you have contributed is very much appreciated and vital to the continuance of the work of the Society. Thank you, all of you.

Some of you have participated in the 'BIG S', the 'Summer Sundae' of fundraising events. The amount raised, so far, is around £7,000. Haven't you done well! Chris Begley did a sponsored Skydive, Emmy Anstee had a Sponsored Walk with family and friends, Pauline Taylor held a Race Night (see separate article), Neil Roache ran 10K in the Great Scottish Run, Danielle Sales had a Sponsored Swim, Beth Hoskins and Miriam Schmidts walked the Wirral Coastal Walk and Lindsay Mapley ran the Henley Half Marathon. Two of our Conference Carers, Amanda and Jason Wilbraham, held a 60's Night and raised £550. Lloyds TSB added £400, bringing the total to a very impressive £950.

Amanda is pictured below with her friend, ready to party in their retro gear. Rob Hymers, also pictured, took part in the Three Peaks Challenge, with the aim of climbing Ben Nevis in Scotland, Scarfell Pike in the Lake District and Snowdon in Wales within 24 hours. He completed the challenge in 27 hours, raising £550 in the process, a fantastic achievement.



More of you have promised to participate, including James Humphreys, who is going to go Paragliding over the Black Mountains in Wales (he has a sponsor page on [justgiving.com](http://justgiving.com)), and Helen May-Simera is planning another 'Very British Tea Party' in America. Allan Clark is going to walk the West Highland Way next year and he has already received a great deal of support from his friends who have donated the proceeds of their fundraising ventures to his cause; thanks go to Stuart Alison and friends (sponsored walk), Robert Watson and friends and Paul Griffin and David Banks (Great Scottish Run). Allan also has a page on [justgiving.com](http://justgiving.com).





Even though the summer is over, it is still not too late to participate, although you may prefer to organize an Autumn Adventure or Winter Warmer event, instead! I still have some fundraising packs available.

I have the Christmas catalogues for Yellow Moon, perfect for Christmas Gifts, Stocking Fillers and Craft items. LMBBS now receives 20% of every product you buy in this catalogue. You can contact me for a catalogue or go online to [www.yellowmoon.co.uk](http://www.yellowmoon.co.uk). Please note that, if you shop online, only items marked with a green F in a green square will bring 20% cashback for the Society. In both cases, you must quote the Source Code SLM10001 for LMBBS to benefit.

Finally, last but definitely not least, I have fantastic news about our BBC Radio 4 Appeal. We have been overwhelmed by the generosity of the Radio 4 listeners and others, very many of whom donated anonymously, who have donated over £10,000 (including Gift Aid). We received our latest cheque over three months after the broadcast! These donations were made specifically for our Annual Residential Weekend and Conference next year. However, we still need more funds to ensure that we can enjoy the excellent standard that we have become used to. We desperately need a sponsor for this event, so if anyone has any suggestions to make in this respect, they would be very gratefully received.

Anne Crotty

## Holidays for the visually impaired



My name is Adele Fricker and I am 37 years old and registered blind.

I go on holidays with travel companies who provide holidays for visually impaired people.

Jubilee Sailing Trust has a fleet of two tall masted sailing ships, which cater for disabled people to go sailing out to sea. The tall ships are working ships that go around the British waters for either a week or five days. My holiday included food and drinks and I had the opportunity to go ashore at two places. You sleep in bunks and they have showers on the ship. There are generally forty people on the ship with a qualified crew and you work a shift pattern of four watches. For each disabled

person, there is a volunteer who will assist you during your time on board. The voyage I had went from England to Le Havre in France. Further information can be obtained from [www.jst.org.uk](http://www.jst.org.uk) or by telephone on 02308 449108.

There are two other companies that I have travelled with who specialise in holidays for the visually impaired and offer exciting holidays. Both companies provide a sighted volunteer on a one-to-one basis. The holidays enable you to meet a wide variety of people who come from all parts of the United Kingdom.

'Traveleyes' is a company run by a visually impaired person and is based in Leeds. The normal group on each holiday is around 16 people with 8 visually impaired and 8 volunteers. They operate throughout the whole of the year with various holidays varying from being very active or just relaxing, the choice is yours. Contact details are [www.traveleyes-international.com](http://www.traveleyes-international.com) or tel. 08448 040221.

'Vitalise' is an established company and also has holidays in the UK and like Traveleyes they also provide sighted volunteer travellers. Contact details are [www.vitalise.org.uk](http://www.vitalise.org.uk) or tel. 0845 3300149.

I have been on holiday to the following countries with both Traveleyes & Vitalise

USA: Grand Canyon and San Francisco

Spain: Andalucia including Seville and Ronda  
Italy: Rome and Florence and Sorrento  
Ireland: Dublin and the Ring of Kerry  
Croatia: Dubrovnik and Split  
Cuba: Havana and the island towns  
Malta: Valletta and the island towns  
Thailand and Cambodia  
Sailing in a yacht around the British Virgin Islands.

I am hoping to go in November on a tour covering the North and South islands of Japan.  
I would be pleased to give you further details of my holiday experiences and you can contact me at my e-mail address: frickeradele@aol.com.

## Exhibition News

### Sight Village, Birmingham

Our presence at Sight Village, Birmingham, was co-ordinated by Chris Humphreys, who attended the event, together with her son, Ross, and LMBBS member, Craig Barrass. The venue this year was New Bingley Hall, which had the advantage of allowing all exhibitors space on the same floor, with plenty of room between the aisles. LMBBS were well placed in the hall and met old and new friends, members and professionals.

Chris reports, 'Sight Village always provides an opportunity to network; new links with other organisations are forged and it gives us the chance to seek out people to speak at our conference in the future or to attend as a delegate. Sight Village is a humbling experience; so many blind and partially sighted people make the effort to travel to Birmingham and you always know when a new coach has arrived by the sudden deluge of visitors and, of course, their guide dogs, who patiently sit and wait for their owners to move around. At one point, we had four guide dogs sitting patiently at one time, whilst their owners sat at our table, some making new friendships, others just grateful to meet someone else with the syndrome.'

The highlight of the event was the now famous 'Karaoke Night' at the Quality Hotel, which provided another chance to network.

The dates for next year's Sight Village, Birmingham are 12<sup>th</sup>/13<sup>th</sup>/14<sup>th</sup> July 2011; we look forward to seeing you there.

### Sight Village, London

Our presence at Sight Village, London, was co-ordinated by Tonia Hymers, who attended the event with Committee members, Richard Zimble and Julie Sales. It was held in Kensington Town Hall, which is a lovely modern building with great facilities and plenty of space. Getting to the venue on day two proved a real problem for many this year, due to the tube strike; however, many did make the effort and, although attendance suffered, especially on day two, it was not as badly affected as it could have been.

Tonia reports, 'Our stand was visited by the usual mix of specialist teachers, social workers and other professionals. It was reassuring to meet so many dedicated people who go the extra mile to help those in their care, which is especially heartening, considering the number of stories we heard of families and individuals struggling to come to terms with the diagnosis and with the lack of support or information available. In addition to the leaflets we have about the syndrome and the support we provide, we were able to pass on information about the new LMBBS clinics, which should make a big difference to those who attend.'

Richard and I took advantage of a lull on day two and visited the other exhibitors. Richard was a great help in bringing me up to speed on all the different technology and services available for the visually impaired and we chatted to 'Look' and 'Guide Dogs for the Blind' among others. We were excited to hear that Guide Dogs are now being offered to visually impaired teenagers, from the age of 13 onwards, with really good results.

A highlight of the exhibition was when a gentleman recognised us from the Radio 4 appeal and came over to say hello. He was so impressed with the script and especially liked the fact that we asked for a specific amount, £20, which he duly popped in the post for us. It was nice to be able to thank him personally and let him know what a success the appeal had been.'

#### Manchester Disability Event

The Manchester Disability Event was held on the 18<sup>th</sup> and 19<sup>th</sup> November at the Novotel Hotel in Manchester. Our presence there was co-ordinated by Chris, who attended with her son, Ross. We will find out how they got on in the next newsletter. There will also be a report on the Ciliopathy Alliance Launch, which was held at the Institute of Child Health, London on the 29<sup>th</sup> November. We will bring further news and dates of exhibitions to you as we get them.

#### Race Night

Pauline Taylor

At the beginning of the year, I decided to hold a local charity night on behalf of the LMBB Society, as it had been two years since my last event. As a family, the knowledge and information we have gained from the society has benefitted us greatly, throughout Keiran's early years and into adulthood. Without this knowledge, we would still be hitting brick walls and trying to educate those in the medical, education and social work professions.

Shortly after I decided to have a charity night, we got the appeal for THE BIG "S" event, which gave me that extra 'get up and GO' and which is why we decided on the month of "S"eptember, hoping for a late Indian Summer. For our last fundraiser, in 2008, we set a target of £1000 and raised £1700, so this year we set a target of £1500.

I cannot say how quickly I realised what a task our fundraising co-ordinator, Anne Crotty, has in front of her, year on year. The letters of rejections, no replies and then there are the companies who only support their chosen charity. The perseverance paid off, however, and I was very grateful that there were still companies and persons out there who were willing to help out with race sponsorship, donations and prizes. Our top prizes were a VIP ticket for Motherwell Football Club, a bicycle, a hotel stay for two and a printer, as well as many more prizes, drinks and vouchers.

The word went out; we sold 110 tickets. The 17th September came round and we had a Race night, disco and food.. the pies and mushy peas went down a treat. We received a surprise £500 donation through a family member who donated a prize he had won at work, after receiving my email about the Race night, and we smashed our target with a total of £2500.

Last week, I received the LMBBS Conference report; we were not able to attend the conference this year but have been lucky to go the last 3 years. Those who have been to the event can only imagine the amount of work that goes into the organisation, never mind the cost, which is why I did what I could to raise some much needed funds. We need to remember the Society relies on donations for all running costs, as well as the conference, every year.

I hope the Big "S" event has been successful. I know it's not easy to raise money but, if we all do our bit, every penny can count.

## A Life-Changing Diagnosis



Alison with her beloved dogs

I've been considering writing something for the newsletter for a while and was finally persuaded by Tonia after she saw my daughter Alison's website. It was shortly after Emma Turnbull's article had appeared in the last news letter. I was fascinated by many things in that article not least that babies are now being diagnosed during pregnancy. It's all so different from our experiences of life without a diagnosis until age 21 and yet so familiar in some ways, not least the months of sickness and weight loss during pregnancy although I must hold the record as it started within 48 hours of conception and continued until an hour after Alison's birth. If I were asked to sum things up in one sentence I would say that much has changed but the situation's still the same. I wholeheartedly agree with Emma that you just have to get on with it because Alison proves that if you don't, you'll be the ones who suffer and miss out.

I truly believe that the new multi-disciplinary clinics are a huge step forwards. Alison and I returned from the clinic at Guys Hospital with a whole new perspective on her future. At 62 and having spent more than five years battling cancer and the side effects of drugs, my life had become one of constant worry about Alison's future and how she'd cope without me. Suddenly all that has disappeared and we can begin to look forward again.

Alison has been told that the disease in her eyes has run its course; there's virtually no more sight loss possible from rod cone dystrophy, just a minor amount over the next ten years. That has to be the best news that every parent can look forward to. Rod cone dystrophy does not mean a future world of total darkness as we all imagined. When Mr Mohammed told her, she instantly realised that it means she'll be able to carry on her life in much the same way as she does now. Her comment? 'I feel as if I've been given a million pounds.'

Just knowing that her problems can be managed is a massive relief after years of believing that there was nothing that could be done. She has an amazing gift, the ability to accept what life has dealt her and to carry on as normally as possible. She never complains about her lack of sight; she has no measurable visual field and the macular has also gone, but she makes the most of the miniscule bit that's left. Now there is the hope of future stem cell therapy or an implant, her cataracts will be removed, her kidneys scanned and monitored and a desperately needed appointment at the BBS sleep clinic to check her lung function.

It's all a far cry from the treatment meted out during her childhood. Having a diagnosis in itself had changed our lives overnight. After 21 years her problems were no longer due to a neurotic mother; we finally had a named condition and to go with it a totally changed attitude from the medical profession in general. Alison was born in 1972 after a difficult pregnancy and delivery. She had night blindness from the start and by two weeks of age, a massive squint. At ten days she was constipated and the suggestion by the health visitor that I put a small amount of sugar in her bottle resulted in projectile vomiting and diarrhoea, but she still gained weight at a rate of knots. Our elderly GP was convinced she was diabetic but hospital doctors were a different matter. As far as they were concerned I was transmitting my anxieties to her.

After several visits to the University Hospital of Wales eye clinic I was told she didn't have a squint, it was the bridge of her nose that made her look like a 'little mongol'. Needless to say after an insistence on seeing someone else, the squint was so severe it was off the measuring scale. Now they were demanding to know why I had left it so long before doing something about it. By 20 months she was wearing glasses and a patch. The prescription for her lenses was arrived at by wrapping her, screaming and writhing, in a blanket, several nurses holding her down and a consultant putting drops in her eyes and trying to refract. The verdict – she was long sighted just like most children with squints. By the time she was 3 there was no improvement in her vision and I was accused of not making her wear the patches. Not bad for a mother who used to walk the streets for hours with a toddler in a pushchair with a bandage wrapped round her head, a hat on top and fingerless mittens to stop her pulling the patches off. More dissatisfaction and another change of consultant. This time she sat in a chair for refraction and lo and behold she was short sighted. She'd had the wrong prescription for around eighteen months but there was never an apology. After



seven months of the correct lenses her eyesight had improved enough for cosmetic surgery to straighten the eye.

In the meantime she had been in hospital for two weeks for tests to solve the sugar problems. Being on solid food had made the sickness and diarrhoea problems worse and she couldn't tolerate milk sugar (lactose) or fruit (fructose) in any form. After three hour glucose tolerance tests her blood sugars rose as high as a diabetic but because they eventually came down naturally, it was determined to have been a mistake in the path labs. In those days it was considered impossible for a child to be born with Type 2 Diabetes.

Over the years I have been told that there was something the matter with me, that I wanted them to find something wrong with my child when she was perfectly fine. A leading paediatrician said she was just a slow but perfectly normal child; sick children were always underweight, Alison was overweight and a leading ophthalmologist told me to go away and have more children and forget about her, she'd never amount to anything. It all served to make me more and more determined to prove them wrong and for many years I battled on without support. Luckily as a qualified teacher I was able to educate her at home so that she didn't remain too far behind her classmates at school. The failure to keep up was a gradual process, she started school ahead of most children, already able to read like an eight year old and then over the next few years the other children made faster progress than her.

Then started the fight to have her removed from mainstream education, resulting in a placement at a school for the physically disabled, where she was referred to by the headteacher as 'Much ado about nothing.' They placed her on a diet table and because she didn't lose weight, of course, it was my fault, I must have been over feeding her at home. At 12 years of age it was decided, against our wishes, that she be placed in a large comprehensive school. What an object of ridicule she was! She was a in a class of 11 year olds, so she was a whole year older, head and shoulders taller, overweight and wearing glasses and surgical boots. The favourite occupation of the other children was to run down the corridors to the stairs and switch off the lights leaving her to struggle to find her way to the next class and end up in hot water for always being late.

It was sheer luck that I had been at college with a member of the staff and she persuaded another teacher to use Alison as her child study for her degree course. She was able to carry out all sorts of developmental tests which highlighted massive, but unacknowledged, problems. We were able to use the test results in another battle to have her transferred back to special education. The placement was in a school for children with moderate to severe learning disabilities, 'just to keep me quiet'. We sacrificed the quality of education for her happiness and I continued to teach her at home.



Eventually it was Alison's love of horses and her impressive riding skills which were to lead us to a diagnosis. In 1991 she was a member of the Great Britain team at the world championships for disabled riders in Denmark. In order to ensure fair competition riders are graded according to the degree of disability and their classification is checked at international competition. Her classifier was a Dutchman, an ophthalmologist, who instantly recognised her condition and was able to give us guidance. It still took another year and a half of battles before we eventually had an appointment at Bristol Royal Infirmary for electro diagnostic testing which proved beyond doubt that I had been right. Two months later, after an insistence from the consultant at University Hospital of Wales that the condition was non progressive, we finally arrived at the clinic of Professor Alan Bird at Moorfields. He was able to give us the diagnosis that changed our lives.

As Professor Beales said, I was totally vindicated. People suddenly changed from antagonistic to sympathetic and curiously interested. Less stress in our lives allowed Alison to blossom into the bright, bubbly, capable and confident person she now is. She is our beautiful loving daughter who is finally achieving her full potential and is very happy. Her days are filled with riding and caring for her two horses and her guide dog and retired dog. She is as independent as is possible and totally computer literate. Competitions still play a huge part in her life and she has been very successful against able bodied as well as disabled people.

Now, thanks to the BBS multi disciplinary clinics, Alison can look forward to a bright future instead of believing that she needs to make the most of what she has now because it could end at any time. My prayer is that no other family ever has to fight as we have. Earlier diagnosis can only mean earlier management and the proper education for our children.

Alison will feature in the December edition of the Guide Dogs magazine 'Forward'. You can find out more about her on her website: [www.diydressage.co.uk](http://www.diydressage.co.uk)

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