



# LMBBS Spring Newsletter 2010



## Welcome to the LMBBS Spring Newsletter

We are only into May 2010, but what an exciting year it is turning out to be.

You will have all received an interim letter about our exciting new project, LMBBS Multi-Disciplinary Clinics. Tonia Hymers, Julie Sales and Chris Humphreys will take up the roles of Family Support and Family Liaison at the Clinics in Great Ormond Street, Guys, Birmingham Children's and Queen Elizabeth Hospitals and have been working closely with Professor Beales, Dr Tim Barratt and Dr Lukas Foggensteiner and teams for 'lift off' on 1<sup>st</sup> April. More information regarding this exciting venture is coming up later.

You will also find information about another exciting project, Ciliopathies Alliance UK. LMBBS is working closely with Professor Beales, Clinicians and other groups in setting up the Alliance, with an Awareness Day to be held at the Kennedy Suite, Institute of Child Health, London on the 29<sup>th</sup> November 2010.

Chris represented LMBBS as a speaker at GIG (Wales) Rare Disease Day, held at the Welsh Assembly in February, speaking about Families coping with a syndrome and Partnerships with Clinicians. LMBBS was further represented by Allan Clark, Pauline and Peter Taylor and their son Keiron at the Scottish Rare Disease Day, and Jonny Fegan at the Ireland event, so well done all. If anyone would like to represent their country next year, please contact Chris.

We are delighted to announce that we have been granted a Radio 4 Appeal, due to take place on 27<sup>th</sup> June. You will find more information about this and our new Summer fundraising initiative inside.

For more exciting events and news updates, enjoy your newsletter.

Thank you to all those who continue to support the society in so many different ways.

Phil Humphreys  
Chairman

## Announcements



### Wedding Bells in the Parker Family

Our congratulations go to Gareth, eldest son of Michael and Drina Parker, joint founders of LMBBS, and Kate, who married on 31<sup>st</sup> October 2009. Gareth's brothers, Richard and Daniel, were joint best man. Gareth and his new bride, Kate, honeymooned in New York. We wish them every happiness for the future.



### Congratulations!

Our congratulations also to Vicky and Wayne, who celebrated their engagement recently. They met through mutual friends and quickly became good friends themselves. They remained just friends for a year before Vicky finally agreed to go out on a date with Wayne. A few months later, Wayne proposed to Vicky on bended knee; we are delighted for them both.



## LMBBS Multi Disciplinary Clinics

Over the past year, the LMBB Society has been supporting Professor Beales in his bid to set up LMBBS Multi Disciplinary (MD) Clinics, with the aim of providing a 'one-stop' annual visit for those with LMBB Syndrome. We know from talking to those with the syndrome, and parents of children with the syndrome, how difficult it is to find doctors with knowledge of LMBBS, the many different aspects of it, and the impact that this has on the patient's health care. Professor Beales has gathered together experts from across the fields of Ophthalmology, Nephrology, Endocrinology and Genetics amongst others, which should bring about a major change in how LMBBS is managed, with a focus on early intervention and good health management.

We are therefore delighted to report that the first clinic was held on Friday, 16<sup>th</sup> April, 2010 at Great Ormond Street Hospital, London, the first of many to be held across four hospitals in the UK. Depending on where patients live, they will be offered an appointment in Birmingham or London; University Hospital, Birmingham (UHB) and Guys Hospital, London will hold clinics for adults, and Birmingham Children's Hospital (BCH) and Great Ormond Street Hospital, London (GOSH) will hold clinics for children.

### Which Clinicians will be available?

On the day patients may be seen by an:

- Ophthalmologist
- Endocrinologist

- Nephrologist
- Geneticist
- Clinical Psychologist
- Dietician

### **What happens at an appointment?**

Each hospital may vary in the structure of the day, however the plan is for the adult clinics to be split into morning and afternoon sessions. The morning clinic will run from 8.45am to 1.30pm (lunch included, some clinics will feature a dietician's demonstration). The afternoon clinic runs from 12.30 to 5.00pm (lunch included with attendance at the dietician's demonstration, if applicable). Each patient will be allocated a clinic room, which will be theirs for the duration. Consultations with each clinician will last approximately 25 minutes. If any further tests are required, each clinician will advise as to what action needs to be taken. The clinics at Guys will be held on Wednesdays and at UHB on Fridays.

At GOSH, the clinic is spread over a day, with the morning spent in the Ophthalmology Outpatients Department, which will allow the time and facilities for optimum assessment and management. The appointments are staggered across 9am and 10am starts, and will finish in time for lunch. Following lunch, rooms will be allocated and again, each consultation will last approximately 25 minutes. The aim is to minimize waiting and provide a more relaxed environment. There will be a play worker on hand who will ensure that appropriate toys/activities are available. LMBBS Clinics at GOSH are on Fridays.

The BCH clinics will be held on Monday afternoons, with the first one to be held on 28<sup>th</sup> June 2010. This clinic is still in the planning stages, and full details will be sent out with the appointment letters.

### **What tests may be done at my appointment?**

A blood sample may be required and patients may also be asked for a urine sample. If any other tests are required, patients will be informed about these on the day.

### **Finally**

Following the appointment, information and test results will be sent to the local health care team to ensure the patient receives optimum care locally. The patient will also receive a copy of this letter.

### **Extra Support**

An important aspect of this service is provided by the LMBB Society. The Society was successful in its bid for funding to support the clinics, and advertised three support worker positions on the LMBBS web page in December/January. Chris Humphreys, Julie Sales and Tonia Hymers have been appointed to these roles and will be available to support the patients and/or their families before, during and after the appointment to help with any concerns about attending the clinics. We understand that attending an appointment in London or Birmingham is not without difficulties for those who live outside the cities, which is why the support workers will be on hand to assist with travel and accommodation arrangements. Although the Society is unable to provide transport funding, we can assist with finding it elsewhere. If required, good quality hotel accommodation will be arranged on a bed and breakfast basis at no cost to the patient or their family. In addition, benefits information can be provided.

### **Meet the team**

Chris, Julie and Tonia, all members of the LMBBS Management Committee, have worked tirelessly over the past year to support Professor Beales in his bid for funding for the BBS MD Clinics and are very excited about the benefits the clinics will bring to the patients and their families.

### **Chris Humphreys**



Chris will co-ordinate and facilitate the clinics held at University Hospital, Birmingham and Birmingham Children's Hospital. Most of you will already know Chris well in her role of LMBBS National Co-ordinator and Conference Co-ordinator. Chris began her working life as a civilian clerk in the South Wales Police Force. Along with her husband Phil (LMBBS Chairman), they became Foster Carers in 1970. Chris became Residential Officer-in-Charge of a Local Authority Children's Home in 1974, caring for 13 teenagers, living 'over the job.' with Phil and their children.

In 1981, Chris resigned to spend more time with her family, continuing to foster and, later, child minding. In 1982, with the arrival of James, a three week old foster child, life would never be the same again for the Humphrey's household. Chris and Phil later adopted James and two other young boys, completing their family of five children and other short term foster children who came along. During that time, Chris supported many groups, including Hyperactive and Dyspraxia Group, Cathedral Children's Club, 'Improvement in education for visually impaired', 'Visually Impaired Children Taking Action', Regional Contact a Family Groups, and gained Accreditation in Counselling. More recently, Chris has been working with GIG Wales and the Ciliopathies Alliance.

Chris and Phil's lives have always revolved around children; their own children, their six grandchildren and the two young foster children they have with them at the moment. Chris combines her role of foster carer with working as a member of Social Services Fostering Team, working with prospective new Carers on 'Skills to Foster'.

Chris has been a committee member for 14 years serving as Fund Raising Officer and Secretary before taking on her current role. She can be contacted by telephone on 01633 718415 or by email at [chris.humphreys4@ntlworld.com](mailto:chris.humphreys4@ntlworld.com)



### **Julie Sales**

Julie will co-ordinate and facilitate the clinics held at Guys Hospital, London. You may already know Julie in her role of LMBBS Secretary and organiser of the Conference Children's Outing. Julie has worked in various professions, including secretarial, catering, physiotherapy and learning support. Julie has completed a counselling course and has gained experience in the past, providing support via the LMBBS helpline.

Julie is married to Kevin (LMBBS treasurer) and together they have two daughters, both of whom have LMBBS. They became aware of the charity when the children were diagnosed in 1997 and became committee members soon after. Julie has worked in a voluntary capacity for the LMBBS for the past ten years, and is currently responsible for the Society's database, webpage and LMBBS merchandise. Julie can be contacted on 01892 685311 or at [kevin.julie1@btinternet.com](mailto:kevin.julie1@btinternet.com)



### **Tonia Hymers**

Tonia will co-ordinate and facilitate the clinics held at Great Ormond Street Hospital, London, in addition to maintaining the BBS Clinics Database. Tonia is the LMBBS Newsletter Editor and Conference Creche Co-ordinator, and was, before that, the Fundraising Co-ordinator. Tonia has been working in a voluntary capacity for the Society for ten years.

Tonia worked for one of the main High Street Banks for fifteen years, before leaving to care for her family. Married to Rob, they have two sons, Daniel (13) and Connor (10). Daniel

was diagnosed with LMBBS as a baby, and the family attended their first conference in 1997. Tonia embarked on a Social Sciences Honours Degree with the Open University in 2004 and will complete it in June. You can email tonia at [toniahymers@btinternet.com](mailto:toniahymers@btinternet.com).

Chris, Julie and Tonia are committed to working together to provide the best possible service, to support the patients and to ensure the clinics are a success. Patients and their families will be contacted by one of the team prior to their appointment; however, in the meantime, the team would be delighted to hear from you, should you have any questions.



## **Benefits and Financial support Update**

Every April, the government makes a number of changes to the rules

governing the benefits system and other forms of financial support for families. Many parents report that they find it very difficult to keep abreast of these new rules and initiatives. Given this, Contact a Family have prepared the following four short articles on forthcoming changes which they believe may be of interest to parents.

### **New benefits rules – child maintenance payments ignored from April 2010**

From 12<sup>th</sup> April, child maintenance payments will no longer be treated as Income when working out if you are entitled to means tested benefits such as income Support, income based Job Seekers Allowance or income related Employment and Support Allowance. These payments are already ignored as income for Housing and Council Tax Benefit and for Tax Credits. If you receive child maintenance payments and were told in the past that your income was too high to receive a means tested benefit, you may find that these new rules allow you to qualify for the first time.

### **Carer's Allowance earnings limit to increase from April**

Carer's Allowance is the only state benefit specifically aimed at carers. However, in order to get Carer's Allowance, one of the rules is that your earnings must be no more than £95 per week. This earnings limit has been unchanged since October 2007. However, from 6th April 2010, the government is to increase this figure to £100 per week.

### **How are my earnings calculated for Carer's Allowance?**

In working out your weekly earnings, certain deductions can be made from your gross wages. For instance, any tax and national insurance you pay is deducted, alongside half of any pension contributions you make.

### **What about if I have to pay someone to look after my children while I am at work?**

If, because of your work, you have to pay someone else to care for the person you look after, or to look after your children, you may also be able to deduct these costs from your earnings. However, the maximum amount that you can deduct for alternative care costs is 50% of what would otherwise have been your earnings. No deduction is allowed if the person you pay is a close relative. These rules may allow some carers to qualify for Carer's Allowance, even though they are earning slightly more than £100 per week.

### **What are the other Carers Allowance rules?**

You must be at least 16 to claim and you can only get Carer's Allowance if the person you look after is in receipt of the care component of Disability Living Allowance at the middle or highest rate or Attendance Allowance (a benefit for elderly people). You cannot claim if you are a student involved in 21 hours or more supervised study. If you are looking after a disabled adult, then, in certain circumstances, an award of Carer's Allowance could lead to a reduction in that disabled person's benefits.

### **Extra Child Trust Fund payments for disabled children**

From April 2010, the government will start to make extra payments into the Child Trust Fund accounts of disabled children. The extra payment will be £100 per year, or £200 per year if a child is on the care component of Disability Living Allowance at the highest rate.

### **Will all disabled children receive these extra payments?**

No. In order to qualify for an additional payment from the government, your child must have been in receipt of **Disability Living Allowance (DLA)** at some point in the previous year. In addition, only children born on or after 1<sup>st</sup> September 2002 have Child Trust Fund accounts.

### **My child gets DLA. What steps do I need to take to ensure my child receives these extra payments into their account?**

The government expects to automatically identify those children who have both DLA and a child trust fund and will then make a payment directly into each child's account. Parents will receive a letter telling them, once a payment has been made.

### **When will my child be able to get the money in their account?**

A child must normally wait until they reach 18 years of age to access the money in their account. However, if your child has a terminal illness and their death could be reasonably expected within six months, you can get early access to buy things that your child needs.

## **The Savings Gateway - government help to boost your savings**

Later this year, a new government backed savings scheme called the Savings Gateway is to be introduced. Aimed at people of working age who are on lower incomes, the government will give you 50 pence for each £1 you save into your Savings Gateway account.

### **Am I eligible for a Savings Gateway account?**

You will qualify for an account if you are getting one of the following benefits or tax credits: Income Support; Incapacity Benefit; Severe Disablement Allowance; Employment and Support Allowance; Job Seekers Allowance; Child Tax Credit (your income must be below £16,040 - this limit may increase after April) or Carers Allowance (you must actually get this, not just have an underlying entitlement).

### **How do I apply for an account?**

When the scheme launches later this year, Her Majesty's Revenue and Customs (HMRC) will write to everyone who is eligible, inviting them to apply for an account and telling them how to go about doing this. Savings Gateway accounts will be offered by a range of banks, building societies and credit unions. You can only open one Savings Gateway account during the course of your lifetime – so you need to think about when would be the best time for you to do so.

### **How much can I save into my account?**

You can save whatever you like - up to a maximum of £25 a month. At the end of the two years, the government will then add a reward of 50 pence for each £1 you've saved.

### **When will I be able to open an account?**

At the time of writing, no launch date has been announced. However, it is unlikely to be before April 2010.

For help and advice regarding any of the above articles, please telephone the Contact a Family helpline (freephone) 0808 808 3555.

## **Jonny & Sharon Fegan's Fundraising weekend**



The intrepid duo (Phil and James Humphreys) flew over to Ireland on 28<sup>th</sup> August for Jonny and Sharon's fundraising weekend. Friday evening saw Phil and James, guests of Sharon and Jonny's family and friends, at the Greyhound Racing, sitting in the glass fronted restaurant overlooking the course. An excellent time was had by one and all, although I think Phil's dog is still running!!!

Saturday morning, and after a typical Irish Breakfast, Phil and James met up with all the participants of the Irish sponsored Walk, 'Up the canal and Back'. As Jonny says, it is not 'how far you walk' but that you are 'sponsored to walk'. Morris, Peter and Alice Crum travelled up to take part as well. After the walk, it was back to the hotel to get ready for the evening entertainment. Once again, everything was donated, the Club, the buffet, the entertainment and the raffle prizes. 'Mr & Mrs', 'Deal or No Deal', an Auction and a raffle were only part of the evening entertainment.



Sunday, and before flying home, Jonny had arranged a treat for James: a ride-out on a speedboat. What Jonny didn't realise was that the owner was about to give James the thrill of his life, he let him take full control across the waters; down went the throttle and off they went with Phil and Jonny hanging on for dear life. Phil was grey getting on and white getting off, but James was on a high. With adrenalin running, it was back to the harbour and treat number two for James, a dual controlled jet-ski and yes, you have guessed, he again took full control, arriving back soaked to the skin, having loved every minute. Over the amazing weekend, Jonny and Sharon raised almost £4,000.00. Well done and thank you to you both, your family and all the people of Newry, for a wonderful weekend and also for your continued support for LMBBS. James is still talking about it. Pity the batteries went on the camera out at sea!!!

By Chris Humphreys

## Waseem Farooq – A Personal Perspective



Hi my name is Waz. I am twenty six years of age and still living at home with my parents. I am registered Blind, but I have a little sight; I have got fifteen per cent in my right eye and in my left eye I have got none.

I went to Dorton College, which is in Sevenoaks, in a small Village called Seal and was there for three years. I achieved a lot of new skills; how to live independently, I learned to do basic cooking, how to use the long cane when going out on my own and I would definitely recommend it to anyone who has a visual impairment disability. I gained lots of certificates in basic food hygiene, good budget management, business studies level 1 and administration level 3.

I first started wearing glasses at Primary school age, which was the age when I was diagnosed with Laurence Moon Bardet Biedl Syndrome. Since then, my sight has deteriorated quite rapidly; I can only read font size 28, in Arial with yellow writing and a dark blue background. At 250%, this is the right font size that I can read close up to my eyes. I also wear a baseball cap which helps me for the glare of the sun and bright lights, and, of course, I use a white cane. I have recently started learning Braille now and I am really enjoying it, it is really good.

I am currently working three days a week for a Recruitment agency called Kent Social Care Professionals. My main role is Audio typing, typing up CV's for clients, for which I use a special adapted piece of equipment which is called JAWS. It is a very impressive tool; it will read documents, internet, e-mail and Face book and also reads out loud any computer commands. I have been doing this job for a quite a while now which I really enjoy.

I keep myself fit by going to my local Gym, which I attend three times a week. I am good at Rowing. I can do two thousand metres in ten minutes. I also enjoy the cycling machine and running on the treadmill, upon which I can run almost three miles in twenty minutes. My running speed is 9.5kph. I use the weights as well to keep fit.

I enjoy socialising with my friends, playing pool and snooker. I normally play well if I can get the colour red. I have noticed that I can see the colour red better than yellow when I am playing pool; I always try to get the red when I can. I enjoy watching all sports, especially football; I am a big Liverpool supporter in the premiership. My favourite player is Fernando Torres; he is a very good striker and is a good team player. I sometimes go and watch live football with my friends; we normally go and watch Tonbridge Angels, Gravesend North Fleet and Gillingham. I enjoy it because the atmosphere is very good and all the fans are very friendly and talkative to me which I really enjoy.

## Ciliopathy Alliance

Recent months have seen the beginnings of a new organisation that will benefit people with LMBBS and related syndromes.

We know from years of scientific and clinical research that many aspects of LMBS Syndrome are caused by defects in the function of *cilia*, small tail-like structures found on many types of cells in the body. There are a number of other rare diseases that also have aspects caused by defects in cilia function (ciliopathies) and these share similar symptoms to those in LMBS. These diseases include Alstrom Syndrome, Polycystic Kidney Disease, and Primary Ciliary Dyskinesia (amongst several others).

LMBS has now initiated closer ties with the support groups for these diseases and will work alongside Professor Beales and Dr Dan Jagger in order to gain mutual benefits. The aims of this collaboration will be:

- to share expertise in fund-raising
- to share expertise in changing health and benefits policy
- to carry out joint research that will help all people with rare ciliopathies
- to hold joint meetings that will share information relevant to all patients and health professionals
- to present a strong and united front for all people with rare ciliopathies

More news about this organisation should be coming soon, watch this space!

Dr Dan Jagger  
Chair, Alstrom Syndrome UK

## Wales Rare Disease Day



Chris was invited by the Genetic Interest Group (GIG) to speak at Rare Disease Day at the Welsh Assembly in Cardiff; the theme this year was 'Patients and Researchers – Partners for Life'. It couldn't have been a more appropriate subject. Chris spoke about families coping with the day to day aspects of living with a rare syndrome and how fortunate the LMBB Society is to have such a good working partnership with many eminent national and international experts.

There was time to socialise, mingling with representatives from many of Wales' Rare Syndrome Groups, before the Reception commenced.

The evening was hosted by Helen Mary Jones, Plaid Cymru's spokesperson for Health and Social Services and was supported by many Assembly Members.

Alastair Kent, chair of Rare Disease UK, said: "There are tens of thousands of people in Wales and millions of people in the UK, living with rare conditions. For many, the information available on their conditions is scarce and scientific research is lacking. A rare disease is one which affects about one in 2,000 people. They are surprisingly common – 175,000 people in Wales (3.5 million in the UK) – live with a rare condition at some point in their lives."

Professor Julian Sampson, Wales Gene Park & Institute of Medical Genetics, Cardiff University, spoke about the importance of a partnership between rare disease groups and researchers.

It was a busy week for Chris and James with interviews for BBC Wales On line, ITV Wales On line, RNIB Insight Radio and a two page spread with photos for one of Wales' Newspapers.

Autographs are available from Chris and James.!!!!!!

## Scottish Rare Disease Day



Pauline and Peter Taylor, together with their son, Keiran, who has LMBBS, attended the Rare Disease evening in the Scottish Parliament building, in Edinburgh. Around fifty people attended, including researchers, charity representatives, those with families affected by rare disease, as well as representatives from the pharmaceutical industry and NHS Scotland. This is Pauline's report on the evening:

Christine Grahame MSP hosted and opened the evening; other key speakers included Sir Ian Wilmut (the scientist who cloned Dolly the sheep) and a Personal Perspective, which made me take stock and be grateful that Keiran is relatively healthy and has the chance of a future ahead. This was the first event that Keiran had attended as an adult with LMBSS.

We were provided with a list of attendees beforehand and knew that Allan Clark, LMBBS family and committee member, would be attending, which gave LMBBS a good presence. There were names of organizations I could not even try and pronounce, but looking down the list of individuals, a name struck a chord. It was Dr John Tomlie, from Yorkhill, who was the very man who set us on our journey with LMBBS. He was very much the same, with less hair, and was interested in how Keiran had progressed and what he was doing now. We were also approached by

individuals who had done research on the human genome and LMBSS had been one of the areas they had investigated. We were also made aware of the possibility of a genetic eye clinic opening in Inverclyde.

The event was well organized and it was interesting to meet people who knew of LMBSS, which was a huge boost. If I reflect back thirteen years, it is amazing what a difference time and awareness makes. This made this event all worthwhile. All in all, a successful night with some key names noted for the future.

Pauline Taylor



### Perkins Brailers – Repairs and Servicing

The Royal National College for the Blind offer a Perkins Braille maintenance and repair service, with all work carried out by an engineer with over 30 years experience of maintaining the College's fleet of Perkins Brailers. Turnaround is fast; they guarantee to return your Perkins Braille within two weeks, and, failing that, will supply you with a replacement to tide you over until your own fully functional model is returned. Postage and packing is free through 'Articles for the Blind'. Yearly service and maintenance contracts for individual and multiple Perkins Brailers are available to schools, colleges, Sensory Support Teams, Associations/Societies for the Blind, etc. and standard servicing costs from as little as £25.

For further information, contact the Perkins Technical Advice and Repair Help Line on 0800 027 6574 or [Perkins.repairs@rncb.ac.uk](mailto:Perkins.repairs@rncb.ac.uk) or visit [www.rncb.ac.uk/perkinsbrailer](http://www.rncb.ac.uk/perkinsbrailer)



### Richard Parker – an update



Roger Thyer-Jones, 6<sup>th</sup> Degree black belt, wrote the article, 'Difference doesn't mean deficit' for last year's newsletter, about his work with Richard Parker. He wrote then, 'In the first month I worked with Richard on his posture as clearly he stooped and this threw his whole body out of alignment making ease of movement almost impossible. We then worked on balance and movement and I taught him how to move using Tai Chi... We worked on perception and hearing so that after about the third session, he was able to identify not only where I was in the studio in terms of the clock face, but the distance I was from him and he could achieve over 90% accuracy here. My aim is to get to a point where he can work with me one to one and get the same feeling we all do from that sort of partner training which all adds to the confidence cycle.'

Since then, Richard has been training privately with Roger and, pictured, you can see Richard in the studio with his official certificate, having recently taken his first grading in karate. To complete the grading, he had to demonstrate that he had a firm foundation in using blocks, kicks and punches to defend himself, as well as having a good standard of fitness. Roger said, "Richard has made real progress in terms of fitness, balance, co-ordination, power and focus. This test was not easy and it is greatly to his credit that he passed it and passed it well."

Richard is now an advanced white belt and is studying for his blue belt, which he hopes to take this year. Our congratulations to Richard on this achievement; we look forward to following his progress in the future.

## Fundraising Round-Up



I don't need to remind any of you that 2009 was a very difficult year for everyone from a financial point of view. In spite of this, fundraising, donations and grants for the Society totalled nearly £21,000, not including the generous support of our loyal group of 'Friends'. Thank you, all of you, for helping to achieve this. You have been as hardworking and inventive as ever... having said that they would have a rest following their incredible achievements in 2008, the Fegan family held a magical fundraising weekend. You can read more about this and other fundraising stories elsewhere in the newsletter. Donations were made following a Christmas Concert, held by St. Mary's Early Education and Childcare Centre in Glasgow and following the production and sale of their "Nutty News" by the Primary 6 pupils of St Mary's Primary School, Duntocher. Jo-Anne Newson's 'Newson Academy' held

collections for LMBBS at their summer and Christmas shows, the pupils of the Ringwood Specialist Language School raised money for the Society and the Downey House School held a collection for us at their Carol Service.

Beth Glacklyn asked for donations instead of presents when she became married in the summer and Anne Mortimer, (mother of Dr Dan Osborn), held a "Girls' Night In", which sounded great fun. John O'Sullivan, (Mary's brother), ran in the London Marathon and Justine Williams ran the 10K Bristol Marathon. The National Sports Raffle raised a wonderful £3,225. Thank you, Chris, for organising that and our grateful thanks to everyone who donated the very special Sports Memorabilia, including our patron, Ryan Jones.

I'm afraid that I was less successful in my applications for grants in 2009. I made nine applications, some of them very lengthy and time-consuming, and had a positive response from only two. (Grant-making bodies are affected by the recession, as well, and have limited funds to share out.) We are enormously grateful to the **Yapp Charitable Trust** who granted us £3,000.



We are thrilled to have been offered a Radio 4 Appeal which is due to take place on 27<sup>th</sup> June, unless a slot becomes free before then. The transmission times are at 07.55 and 21.26 on the Sunday and 15.27 on the following Thursday. We are in the process of writing a script and choosing a presenter. Terry and I went to London for a briefing session and met some other groups who have Appeals soon. We were all given some very sound advice, a list and a timetable of things to do. Please keep an eye on our website for updates about the Appeal.

This year promises to be just as difficult, if not worse, than 2009 in respect of applications for grants so, once more, we really need a huge effort from all of you to enable the valuable work of the Society to continue. We need to aim for £30,000 a year.



### Summer Fundraising Event.

The National Coffee Morning was moderately successful last year, raising £1,338 for the Society. However, only six groups of people participated in this event, including Jackie Chapman and family, who raised £700; congratulations to all of you who held a coffee morning, for working so hard and raising so much money. Just think, then, how much could be raised for the Society, if we **all** tried to take part in this year's plans. We are going to hold a summer event this year..... "THE BIG "S", a Summer Sundae" of activities, e.g. a Sausage Sizzle (BBQ), a Sixty's or Seventy's disco, a Sponsored Stroll, a Sunflower-growing competition, etc. If you are unable to organise something yourself, try to persuade a friend or relative to hold one for you. We will provide a fundraising pack to help which will include more suggestions for a "BIG "S" event. Don't forget to send Tonia or me any photographs that you may have of your event and a few words about it.



**The London Marathon** We have been granted a Silver Bond, this year. Thank you, Louise, for all your efforts on our behalf.



You can find details on our webpage about the Weather Lottery or you can help the Society by buying lovely craft and educational products from Yellow Moon, ([www.yellowmoon.org.uk](http://www.yellowmoon.org.uk)). You will need to quote the Source Code SLM10001, for us to benefit. Please contact me if you would prefer to have a catalogue to browse through.

It just remains for me to say thank you to all of you who have helped the Society, each in your own way. I am sorry that I can't name every single one of you and some of the amazing results of your efforts. THANK YOU, all of you.

Anne Crotty

## Shane Ryan



In the Spring Newsletter, 2009, Shane Ryan, from Ballybricken in County Limerick, told us about his passion for rowing, for which he had just won the title of 'British Indoor Rowing Adaptive Men's LTS Open Champion'.

Shane competes in the LTS category which means he is able to use his legs, trunks and arms and he competes with the Irish Adaptive Rowing Team. Since last year's report, the team has been to Munich for the Rowing World Cup (the team of 4 won bronze) and Poland for the World Championships; (the team won the B final so they are ranked 7th in the world).

Shane competed in the British Indoor Rowing Championships (BIRC) again in 2009 and came home with a silver medal, having been beaten by the UK competitor by 1 second.

In February, Shane travelled to Boston for the World Indoor Rowing Championships, which is the first time that Adaptive Rowers have taken part in this prestigious competition. All of the competitions that Shane has competed in are regular rowing competitions with events for Adaptive Rowers. This makes the sport very inclusive and they are mixing with the top rowers in the world!

Shane's mum, Marie, says, 'It really is a great sport for anyone with a visual impairment; most rowing clubs now include adaptive teams, as it is now a sport in the Paralympics. There is lots of training and discipline involved which suits Shane down to the ground. It has also done wonders for his weight and blood pressure - his doctor is delighted with him.'

Congratulations Shane; we look forward to hearing about more of your considerable achievements.

## Helen's British Tea Party

Dr Helen May-Simera



Regular readers will remember that, in 2008, Helen, who at the time was a member of Professor Beales' team, completed the Bath to Paris Cycle Ride to raise funds for the Society. Shortly after, Helen moved to the States to work, and it wasn't long before she was fundraising for the Society again, by hosting a 'Very British Tea Party'. The event was such a success, raising just over \$200 that Helen decided to repeat the event, proving that the Society is never far from her thoughts. Again, the Tea Party was a great success and this time raised around \$300, which is fantastic. Helen wrote to us back in the Autumn with an update:

'All the people thought it was a great idea and we had such fun with the raffle as well. I made lots of typical English sandwiches and confused them all with the cheese and pickle. They were expecting their kind of pickle and couldn't quite get the taste of Branston. The scones were a great success and I even managed to find real clotted cream here. I had bought Mr Kipling cakes which went down well at the end. I was quite pleased I had a few left over, so I have been munching on them all week.

It's Thanksgiving here next week, I'll be happy to have a few days off work. I seem to find the work pace much more intense here than in the UK. I am so upset to be missing the conference again next year, but hopefully I will have lots of research to up-date you all on by 2011'.

A big thank you goes to Helen for her continued support and we really look forward to catching up with her next year.

### Power Cut – Megan Humphreys



Nine year old Megan Humphreys, like most girls of her age, loves all things electric, but decided that, for 24 hours on Sunday 15th November 2009, she would give up all electric items to raise money for the LMBB Society. Megan is the niece of James Humphreys, who, regular readers will know, is a keen daredevil who combines his love of extreme sports with fundraising for the Society.

Megan gave up her hair dryer, tongs, television, games machine and computer, as well as the use of the oven, microwave and kettle, which meant sandwiches and cold meals and drinks for the day. Megan spent the day playing board games with her Mum, catching up on school work and other boring things like tidying her bedroom and playroom. She said the worst thing was giving up her television and missing the 'X Factor'. Megan raised a brilliant £245 for the Society, which is just fantastic – well done and keep up the good work, especially the homework and bedroom tidying!

### Cheer on the Carers!



Louise Butcher, a member of the Conference Childcare Team, ran in the London Marathon for us on Sunday 25<sup>th</sup> April. Here is an extract from Louise's Just Giving Page:

'So why am I running in support of the LMBB Society? Well, Danielle (18) and Hollie (15) are my second cousins, both girls were born with LMBBS and have had to overcome many challenges as they have grown up, challenges which many adults would struggle to deal with. Having watched Hollie and Danielle grow up with LMBBS, I wanted to do more, get more involved with the charity and help out as much as I could. I try to attend the LMBBS conference every year as part of the Childcare Team, and here, I see the invaluable support that the charity offers to families living with the syndrome. This year, however, I will be

unable to attend as I will be pounding the streets of London in the hope of raising as much money as I possibly can to support them.'

This was a huge undertaking. We have wished Louise every success with her training and we thank her for her on-going support. Please, please support Louise via her Just Giving page, [www.justgiving.com/Louise.Butcher](http://www.justgiving.com/Louise.Butcher).



On Sunday 11<sup>th</sup> April, Martin Cleary and Andy Castle, who are also members of the Conference Childcare Team, both ran the Paddock Wood, Kent half marathon. They have both been supporting the Society for a number of years, and we thank them for their on-going support. If you wish to sponsor Martin and Andy, please email Julie Sales on [kevin.julie1@btinternet.com](mailto:kevin.julie1@btinternet.com).

Spencer Drake, a friend of the Sales' family, completed the Tunbridge Wells half marathon to raise funds for LMBBS and sent the following report:



'I am pleased to say that I have exceeded the sponsorship target I had set and the final figure should be close to £260. I think this must have been the toughest half marathon that Tunbridge Wells has seen for a long time. Numerous parts of the course were flooded and the driving rain made running difficult. However, there is always light at the end of the tunnel... there was plenty of welcome support around the entire course and the marshals did an excellent job with handing out drinks en route. The hardest part of the course was the dreaded Fordcombe Hill, although there was a good pace setter ahead of us and we all managed to keep up the rhythm. The final run in from Langton

Green seemed to last forever but the finish line eventually appeared in the distance with the crowds cheering us in! I finished in a time of 1 hour and 48 minutes, a personal best (until next year). To sum it up, a great event, with a superb atmosphere. Great fun.



**Bardet-Biedl Australia**  
You're not alone

My name is Kathryn Murphy and some of you may remember me from the LMBBS Family Conference in 2006, where I gave a Personal Perspective of living with LMBBS. It is amazing to realise that it is nearly four years since then. I continue to baffle my doctors with strange symptoms and problems for which they continue to have no answers. I said back in 2006 that my BBS had been a roller coaster ride; well, I don't think that is a fair description anymore. On a roller coaster ride, you can get on and off. These days I don't, it's just constant, but maybe, if someone can work out what is wrong, then things will improve again. Once again, I keep trying to help all the doctors by researching as much as possible, and I would put a bet on the problem being my kidneys. Unfortunately, the medical world works in terms of data and, without proof in tests, symptoms and a strong family history, you do not get very far. The problem is getting the medical world to understand I don't do normal, I have never done normal. Anyway, what is "Normal" with BBS?

Unfortunately, I can no longer boast of having being diagnosed with RP, with no further deterioration to my sight. My sight remains good; however in August of 2008, I was advised of the first progression of RP. Fortunately, it appears to have settled again. On a day to day basis, I actually experience more troubles from my Macula. I have had to make some adjustments for this such as increasing the zoom on my computer and decreasing the brightness of the screen. Also, my favourite hobby of Scrapbooking is becoming a little difficult and, after half an hour, I start to get headaches and have trouble focusing, so am changing over to digital, using my computer, where there are more options on screen sizes and so on. Considering everything though, I am extremely fortunate. Amazingly, I am still allowed to drive at night, although I have put in my own restrictions, in that I probably don't drive more than 5 kms, and only in areas I know well and when it is dry.

For years now, I have felt that there has been a greater purpose to the reason I have BBS. Having completed a nursing degree and worked as a nurse, this has enabled me to develop a solid medical knowledge base. My greatest hope is that somehow I can use this medical knowledge, together with the first hand personal experience of living with BBS, to help other people trying to cope with the syndrome. In Australia, although more Doctors are hearing of BBS, we are still very much isolated and there is no support. In the last four years, I have been trying to change this by endeavouring to start some sort of support group. Starting up a contact service for BBS in Australia has been a huge learning experience. Finally, with the generous help of a web design company, I am delighted to be able to announce that there is finally a contact service for people with BBS in Australia, in the form of a website.

**Bardet-Biedl Australia** is our official name and can be found at: [www.bardetbiedlaustralia.org](http://www.bardetbiedlaustralia.org)

### **And finally....A Winning Combination!**

Our patron, Ryan Jones, gave an interview to Betfair and, in return, they gave him £50 to donate to a chosen charity, to place a bet on any horse racing event in the country. Ryan nominated the LMBBS and our Chairman, Phil Humphreys, was delegated to place the bet. Phil did the Society proud. He 'studied form' and placed the bet, which resulted in £220.00 for the Society; so thank you Ryan, thank you BetFair and thank you Phil.

**The views and opinions expressed in this newsletter are those of the authors of the articles. They do not necessarily express the views and policy of LMBBS. Whilst every effort is made to check the accuracy of information reproduced, readers are advised to check with the original source before acting on it.**

## **Contact Details**

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For newsletters, leaflets, tape or disk requests please contact Tonia Hymers by e-mail at [toniahymers@btinternet.com](mailto:toniahymers@btinternet.com). Alternatively telephone your request to Chris Humphries and she will pass the details on.

For fundraising information or to join the Friends of the LMBBS please contact Anne Crotty on (01255) 507977 or by e-mail at [anne\\_and\\_terry@yahoo.co.uk](mailto:anne_and_terry@yahoo.co.uk)

The LMBBS web address is [www.lmbbs.org.uk](http://www.lmbbs.org.uk). All of the above contact details are posted on our web site.

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