



LMBBS Summer Newsletter 2013

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Contact Details

National and Conference Co-ordinator, Temporary Fundraising and LMBBS Clinic Support Worker: Chris Humphreys: (01633) 718415 or by e-mail at chris.humphreys4@ntlworld.com

Newsletter Editor and LMBBS Clinic Support Worker
Tonia Hymers: 07805 685342 or by e-mail at toniahymers@btinternet.com.

Secretary and LMBBS Clinic Support Worker
Julie Sales: (01892) 685311 or by email at Kevin.julie1@btinternet.com

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

Notices

Fiaz Bokhari

We were saddened to hear of the tragic death of Fiaz Bokhari on the 28th January 2013. Our heartfelt condolences are sent to Mr & Mrs Bokhari and their family for the sad loss of their son, who passed away on 28th January 2013. Many of you will know the family, who have been members of the Society since its inception and have become close to us all.

Terry Stone

We send sincere condolences to Jean Stone and her sons Jason and Mark on the sad and sudden loss of husband and father Terry, who sadly passed away on 5th June 2013. Many of you will know the family from attending our conferences and condolences have been sent on behalf of the Society.

Foreword



Welcome to our bumper Summer Newsletter

With the cold, wet and windy days behind us, the sun shining, holidays looming and barbecues burning, it is time to relax in your garden and enjoy your newsletter.

Our successful family conference is over and once again, a wonderful weekend was had by all. Your Conference Report should be with you late summer and will bring those of you who were unable to attend, up to date with some of the latest information and research and refresh the memories of those who were there.

We recently attended the equally successful first Ciliopathy Family Conference, which was also held at the Hilton Hotel, Northampton at the end of May. This event allowed us to meet other Rare Groups and their families and provided an opportunity for everyone to come together and discuss the future they would like for children and young people with a ciliopathy. More information will be available on our web site during the summer.

Inside, you will hear about some of the fundraising events organised by our members and I cannot stress enough how grateful we are for your ongoing support. The Teague family, newly diagnosed last year, amazed and full of gratitude at the level of care and support received from the Society and Specialist Clinics, decided, along with their extended family and friends, to plan nine months of fundraising. Hear about the journey of this inspirational family and their achievements inside.

You will also find details inside of our new fundraising initiative for 2013, an opportunity to involve family, friends and colleagues in a UK Blindfold Bowling Challenge during the week of 23rd-27th September. This is a fantastic opportunity for the whole of the UK and Southern Ireland to unite, have fun and raise funds for your Society at the same time.

I look forward to hearing from you and to working with you to make this our most successful fundraising year. I know you can do it!

Regards

Chris Humphreys



In response to popular demand, LMBBS now has two Facebook pages, 'Laurence-Moon-Bardet-Biedl Society', our 'international' page, and 'LMBBS UK' for UK based members only. This is proving to be an excellent way for the Society to keep in touch with its members; more importantly though, it is there for YOU to keep in touch with each other, so, if you haven't already done so, become a 'LMBBS FaceBook Friend' and keep up to date with the Society and its supporters.

My Journey



Hello all, my name is Claire, I am 29 years old and I have two beautiful boys, Joshua who is eight years old and Ryan who is five. As every mother dreams of, you draw up a picture in your mind of a perfect family; I would like to share with you, my journey.

I was 21 years old when my first child, Joshua, was born. He was perfect and gorgeous, although very sleepy and showed signs of development delay; he was late sitting up, crawling, walking and his speech was very slow too. After so many check-ups with doctors they just said he had a learning disability, so we accepted that.

One night, in the winter months at around six, we went to go out. It was very dark, (Joshua was two years old at this point) I opened the front door and Joshua wouldn't walk, he was putting his hands in front of him and feeling the floor with his feet. We didn't know what was wrong. Joshua was also having a lot of accidents around the house, like bumping into door frames and tables, which all seemed to be when the lights were dim.

We had a check up at hospital and Joshua was referred to an Ophthalmologist. Joshua was checked three times in a twelve month period and they said his vision was fine. Joshua's dad, Lee, and I said there must be something wrong. Joshua continued to see doctors as he developed other health problems with his kidneys, bladder, hearing, sleep apnoea and he was also putting on so much weight.

Joshua was then referred to see a Geneticist; by this time our other child Ryan was born, perfect and gorgeous and looked just like Joshua. As Ryan was growing up he showed all the same signs as Joshua, so Geneticists put them through test after test, but still everything came back negative.

As the months went by, Joshua's vision just seemed to be getting much worse; in the daylight he seemed to be bumping in to things and we were now guiding Joshua around our own home. I did research on the internet, reading up on different syndromes, but found nothing that explained what Joshua and Ryan were experiencing.

Joshua was only six years old, was struggling with his weight and had already had so many ear, nose, throat and bladder operations. Most importantly, he was still struggling with his vision. Ryan was three years old and had developed all the same problems; he too has had so many operations, developed epilepsy, struggled with his weight and again, like Joshua, was unable to see in the dark or in dim light. Finally, we had an appointment for both of our children to see a Paediatrician, who suggested we have the children tested for Laurence-Moon-Bardet-Biedl Syndrome. A letter was sent to our Geneticist.

The Geneticist referred us to the Birmingham Children's Hospital specialist clinic for Laurence Moon-Bardet-Biedl-Syndrome. Joshua and Ryan had blood tests and, after three months, the results were back; Lee and I opened the letter and it confirmed our children had Laurence-Moon-Bardet-Biedl-Syndrome, a very rare disorder affecting approximately 400 people in the UK.

This explained all the health problems and confirmed their eye condition, Rod-Cone Dystrophy. I had a load of booklets explaining about the syndrome and in one way I was kind of relieved, but in another way, I felt like my whole world had come crashing down. I read all the booklets about the medical and learning side of things, but when I read that blindness happens around the age of fifteen years old, written in black and white, this was, and still is the most difficult aspect to cope with as a family. My children mean the world to me, and although I face a more challenging job of being a parent, I will continue to love and support them and all other visual impaired people. There isn't a day or even an hour goes by that I don't think about Laurence-Moon-Bardet-Biedl Syndrome, Who would have thought that just five words could change our lives so much. We have learned how all our other senses are so very important; Joshua is learning Braille and he is hearing more and more sounds, he feels my face with his cute little hands.

Joshua is now eight years old and has started cane training and is doing extremely well at his fantastic school. Ryan is now five years old and has started a new school for children with visual difficulties. The boys are amazing and never moan about anything life throws at them. We will never be able to change things, so we live life to the full with them; I am so proud to be their mother and my partner Lee is such a great support and fantastic Dad to the boys. It has been a tough ride so far. I never thought it would be this tough being a parent, but what I do know, is that I do have the perfect family, who I love very dearly.

Thank you for listening to My Journey

It's Good to Talk!



New Families Contact

Claire Anstee is our New Families Contact. Claire is married to Jason and they have three children. Claire's daughter has LMBBS and so she knows too well how difficult the early days of diagnosis can be. If you feel you would benefit from talking to another parent about the syndrome and diagnosis, Claire is more than happy to listen and help in any way she can. She can be contacted by telephone after 4.30pm, Monday to Friday, on 01604 880386 or via email at claire@anstee2.orangehome.co.uk



Adults Contact

Steve Burge is our contact for affected adults, their families or carers. Steve was diagnosed with LMBBS at eleven years old, lost his vision at the age of 21 and has been involved with the Society since 1997. Music has

always played a big part in Steve's life and he credits his love of music, especially his drumming, with helping him cope with having the syndrome. Steve can be contacted on 07833 228463, at stevebudge@live.co.uk, via Skype at budge-2005 or by post at 38 Pocklington Court, 74 Alton Road, Roehampton, London, SW15 4NN



SEN Contact

Dianne Hand is a Special Educational Needs Co-ordinator and has a son with BBS, so has a great deal of knowledge about the education system with regards to Special Educational Needs. If you have any questions about getting support for your child at school, or would just like to chat to someone with knowledge of how the system works, Dianne is more than happy to help. She can be contacted via email at diannehand@live.co.uk or by telephone on 0161 442 0989.

Don't forget you can also contact us via the LMBBS helpline: 01633 718415, further contact details are on the back page of the newsletter.



Introduction

From 8 April 2013, the Government is replacing Disability Living Allowance (DLA) for people aged between 16 and 64 with a new benefit called Personal Independence Payment (PIP).

In this factsheet we tell you how the introduction of PIP is likely to affect you if you are blind or partially sighted. This factsheet gives you general information rather than advice. If you think you need advice on a specific issue, please contact RNIB on 0303 123 9999.

Please note that:

- there are no plans to replace DLA for either children under 16 or people aged 65 and over who are already receiving DLA
- there is no change for you if you receive Attendance Allowance.

What will PIP consist of?

PIP consists of two components:

- a **daily living** component
- and a **mobility** component.

DWP may award you one or both of these components.

Both components will have a **standard rate** and an **enhanced rate**. DWP will award the:

- **standard** rate when they believe a person's ability to carry out daily living or mobility activities is **limited** as a result of their physical or mental condition
- **enhanced** rate when they believe a person's ability to carry out daily living or mobility activities is **severely limited** as a result of their physical or mental condition.

The qualifying period for PIP

When making claims for PIP you will need to meet the “**required period condition**”. This means that you must have had your disability (including sight loss) for the **three months** before you claim PIP **and** that DWP **expect your disability to last for the next nine months**.

Do I have to be assessed for PIP?

Yes. When you claim PIP, it is very likely that you will be asked to attend a face-to-face consultation which will consider your individual circumstances across **twelve activities**.

For each activity, there are different descriptors which describe a person’s ability to do something. Each of these descriptors has a score associated with it and you will score points based on the descriptors that apply to you.

For each descriptor to apply you must show that you are unable to carry out the activity “reliably, repeatedly, safely and in a timely manner”.

Of the twelve activities, ten relate to the **daily living component**:

- preparing food and drink
- taking nutrition
- managing therapy or monitoring
- bathing and grooming
- managing toilet needs or incontinence
- dressing and undressing
- communicating verbally
- reading and understanding signs, symbols and words
- engaging socially
- making financial decisions.

The other two activities relate to the **mobility component**:

- planning and following a journey
- moving around.

You can score points in more than one activity. For DWP to award you the **standard rate**, you have to score at least **8 points** in total for the relevant activities. To receive the **enhanced rate** you have to score at least **12 points** in total.

The activities include a greater acknowledgement of aids and appliances than there was for DLA, and the need for them may lead to increased points for you. Aids are devices that help you in the performance of a function, such as spectacles or something to help you carry out a task such as a liquid level indicator or braille on shower controls. We will not know exactly how DWP views using these aids until the PIP assessment process starts.

The claiming process

DWP will split the process of claiming PIP into two parts:

- the first part takes place over the phone and this is where DWP will take initial details of your claim
- in the second part, DWP will automatically generate your claim form and send it to you. Your claim form will include a barcode (to uniquely identify your form) and some parts of it will already be completed for you (your name, address and date of birth, for example).

I already receive DLA – will I automatically receive PIP?

No, there is no automatic transfer from DLA to PIP and you will have to make a claim for it. When this happens varies.

From October 2013 until October 2015

Between these two dates, DWP will invite you to make a claim for PIP:

- if your DLA is due for a renewal
- **or** if you report a change in circumstances
- **or** if you wish to have your claim for DLA re-assessed. In this case, DWP will deem you to be a “self-selector”.

If you are considering whether to report a change of circumstance or be deemed a “self-selector” please call our Helpline for further advice.

From October 2015

If none of the above applies to you, DWP will write to you from October 2015 to invite you to make a claim for PIP. DWP will then assess you against the new entitlement criteria.

Passporting to other benefits

Being able to receive one type of benefit often means that it will be easier for you to receive other benefits or concessions. This is called “passporting”. For PIP, the following will apply.

- If you receive either of the rates for the daily living component, this will allow for carer’s allowance to be claimed.
- If you receive the enhanced rate for the mobility component, this will act as a route to the Motability scheme.
- PIP will **not** give entitlement to extra support within Universal Credit.

Benefits cap

From April 2013 the Government is introducing a cap on the total amount of benefit that people of working age can receive; however, if you, your partner or your child receives either PIP or DLA, you will not be affected by this cap.

What can I do if I don’t get awarded PIP or am unhappy with the rates I am awarded?

You can ask DWP to look at their decision again if you are unhappy with it and wish to dispute it. The DWP Decision Maker will contact you to discuss the reasons for their decision and answer any questions you (or someone acting on your behalf) may have. This is called “mandatory reconsideration”.

If, after DWP have reconsidered their decision, you are still unhappy with the outcome and wish to dispute it, you can then lodge an appeal with HM Courts & Tribunal Service.

The RNIB Legal Rights Service can provide legal advice and, where possible, representation on appeals to the Tribunal.

If you have any questions about any of the information mentioned in this factsheet or want further advice, please call the **RNIB Helpline on 0303 123 9999** or go to **rnib.org.uk**.

You See it, We Dot it!



Jane Sellers set up a Braille Transcription service in October 2012, with a small quantity of work from Surrey Coalition of Disabled People. She said, "I have always wanted to do transcribing but didn't have the idea or funds until I came across EmployAbility, Surrey". EmployAbility is a service which aims to support disabled people to enable them to have the same opportunities for rewarding, meaningful employment as everyone else. Their scheme enabled Jane to get started and last year she went to Barclays and opened up a business account with them. Jane worked for Barclays for many years, and the staff were delighted that Jane went back to them when she started her business. Jenny, Jane's PA, arranged for them both to go on a business course, where they learnt about marketing. They successfully obtained funding, with help from their Disability Employment Officer, which enabled Jane to pay for Jenny's services.

In February, Jane learned that she had won a 'Making a Difference' Award and she was to attend a ceremony on the 19th April. Jane said, "It was a magical day and the ceremony was a lovely occasion. My Mum travelled over from Spain, however it was a shame my Dad couldn't attend, so I phoned him instead. The day started with me having my hair done at the local hairdressers, so it looked nice for the photos, of which many were taken. I wore a new white blouse, black trousers and a black jacket with black velour shoes."

The award Jane received was for EmployAbility's Self Employment Works Scheme, which was introduced in September 2012. Carol Pearson, from Surrey Coalition of Disabled People, gave a speech and said that they now needed to promote Jane's business, which produces good quality Braille and is good value for money. Jane also had to write a speech, which she completed as part of her English homework for English Literacy Functional Skills, entry level 2. She wrote about her Dad, Peter, who inspired her to set up her own business.

The reception was held at Denbies English Vineyard and the evening was rounded off with a buffet and a glass of Surrey Gold, a wine produced on site. Jane is delighted with her award, which she proudly displays on her mantelpiece, safely stuck down with blu-tack, to stop her friend's guide dog swooping it off with his tail!

Jane is now flying solo, with the aid of funding from Access to Work, which pays for her new PA, John. Jane is teaching John how to use the embosser and they are in the process of having some business cards made with their slogan 'You see it, We dot it!'

Silent Heroes



There are many individuals and companies that help our Society, often quietly in the background, without expecting recognition or praise and this is our chance to say 'Thank You!'

This edition, we would like to say thank you to Rob Hymers' employer, '**Harwich International Port**'. As you will have read in the 'Behind the Scenes' article, the team at HIP Freight Services have taken care of all our franking needs for the past nine years, saving us considerable work. In addition, for the past two years, the Port has provided and fuelled a mini bus to transport the creche toys (and Hymers family) to the Annual Family

Conference. For their general understanding and support over the years, and for all of the above, THANK YOU!

Guardian Angel

At a recent clinic at Great Ormond Street Hospital, there was a young mum and her son attending, who had travelled across London during rush hour. London is really difficult during this time, however factor in a child in a wheelchair and it can be impossible. I asked Mum how her journey had been and she looked close to tears. I thought she must have had an awful time, however it was just the opposite.

At her home station, she was having difficulty purchasing her ticket, not sure which zones she needed, when a business man stepped in to assist. He bought them all tickets for Charing Cross, refusing to take any payment. He helped them on and off the train, before popping them into a taxi to the hospital, insisting she allow him to pay for the return fare.

Overwhelmed, she told the taxi driver about her generous benefactor, and was stunned when upon reaching the hospital, the taxi driver refused to accept payment. Cities can be challenging during busy periods and commuters rarely look up and register the world around them, however on this particular morning, one did and what a difference he made.

Tonia



Fundraising Round-Up

You will hear later on, about the amazing Teague family, who decided to give back to the Society by undertaking a year of fundraising, incorporating several half-marathons and a full marathon. Zoey called it the best therapy ever and says it has left her a changed person. The Teagues and their family and friends are an inspiration and we are so grateful for their support.

A friend of the Teagues, Lindsay Edwards, together with Gym Clubs throughout the country raised an incredible £5000 for the Society, via a very simple fundraising scheme. New members at participating Gym Clubs nationwide, are asked to donate £1 to that month's nominated charity and Lindsay nominated LMBBS in support of her friends. What a fantastic scheme and so simple too... maybe there is a similar scheme operating at a club near you??

Our heartfelt thanks also go to:

- Graeme and Robert Roache for their cycling efforts, raising over £4,000, with further donations from Arova Ltd (£500) and Margaret Roache (£25)
- Darron Jones and members of Usk Cricket Club who have been extremely supportive to the Society over the past few years. At their recent Annual Dinner, they raised £575 with a further £700 donated from the Ladies Night raffle. Their next fund raising event is Usk Cricket Club v Newport Dragons, to take place on 4th August 2013.
- Luis Tobenas, whose son is an employee of Darron Jones for his tandem parachute jump, raising over £800.
- Emma Tadgell and family and friends who raised £180 selling cakes, with a further £284 raised by the ladies of Chapel.

- Daniel Hymers, who held a 'Bake Sale' and raised £50 for LMBBS and a further £50 for Comic Relief. Well done Dan.
- Paul Rose, a friend of Oliver Sloane, who raised £488 with a 'Dressing Down Day'. This was matched by Lloyds TSB Bank and Paul has offered the services of Lloyds TSB Volunteer Personnel to assist with any fundraising. If you have any events in the future and would like their help, please contact Chris Humphreys who will pass on your details.

There are many more of you who have been fundraising over the past year, organising raffles, cake stalls, selling handmade cards and much more. Our Society would not survive without your efforts and those who support you and our thanks go to each and every one of you.

If you would like to organise an event, and are looking for a fun fundraising initiative, look no further than our Sponsored Blindfold Bowling Challenge, featured in this newsletter.

For further information or general fundraising support, please contact Chris Humphreys: chris.humphreys4@ntlworld.com or telephone 01633 718415. Looking forward to hearing from you.



LMBBS Blindfold Bowling Challenge - 23rd-27th September 2013

We are always trying to come up with fun ways of raising money and our Sponsored Blindfold Bowling Challenge certainly ticks the box. We would like to invite all our members regardless of age or vision to take part, during the week of 23rd – 27th September 2013. The challenge involves playing one sighted game and one blindfolded game of bowling. Involve your family, friends and colleagues and ask each to put together a team of four to six players. You can give the winning team gold medals and the event can be an annual challenge amongst your family and friends.

It really is simple to organise, just follow these steps:

- Decide on the number of teams.
- Choose a captain for each and provide them with sponsor forms.
- Each captain invites his/her team members from their circle of friends
- Contact your local Bowling centre, they usually give a discount on a quiet evening.
- Sponsor money can be collected before the event and handed in on the day.
- You can give medals to the winning team (gold, plastic children's medals will do) and LMBBS stickers to the runners up.
- Hold a small raffle if you wish.
- Involve the media and tell them it is a UK Challenge. Have an information leaflet ready.

For minimal individual effort, this event has the potential to raise a great deal of money, it works like this: each team pays a £20 entry fee (£5 per head) and collects sponsorship for the event. If each team member raises around £50 in sponsorship, each team will contribute

around £200-£250 to the pot. Four teams could raise £1,000 to £1,500 and if 10 of our members take on this challenge, that's £10,000 to £15,000 raised for the Society!

For the more outrageous among you, why not make it a Sponsored Fancy Dress Blindfold Bowling Challenge!! Above all have fun, this is something the whole family can take part in. Don't forget to take photos and we can print them in the next newsletter.

For further information or general fundraising support, please contact Chris Humphreys: chris.humphreys4@ntlworld.com or telephone 01633 718415.

A letter from Australia



Conference 2012 was absolutely amazing. After the Conference, I headed north to Scotland, where I spent three weeks seeing some of this glorious country. Then it was on to the Netherlands, before landing on the Sales' doorstep, absolutely exhausted. I had a fantastic time staying with them and cannot thank everyone enough who helped and enabled me to attend the day at the LMBBS clinic on the 23rd May.

Being Australian, I knew I couldn't be seen as a patient at the clinic, however I did beg if I could please just ask the kidney specialist two questions. For so long, I had felt that my kidneys were the source of my real problems with BBS and this was a big part of my reason for coming over to the UK, to have the opportunity to talk to other sufferers and find out if they were experiencing anything like I was.

I asked the BBS Clinics Nephrologist if it was just the change in weather that accounted for my needing less salt tablets; it had been freezing cold during my entire stay in the UK and my salt needs had dropped from 7 tablets to 2-3 tablets; I was concentrating my urine really well. The answer was 'yes'. Most of my doctors in Australia feel the same and really, I need to come back to the UK right now or else hibernate in an air conditioned room; I hate summer. For the previous two weeks, I had needed 9-10 salt tablets every day; it is exhausting having such fluctuating salt levels.

The other question was: Were doctors from the clinic recommending any treatment specific to Nephrogenic Diabetes Insipidus for BBS. The answer was 'no, just per the management of anyone who has the condition'. The doctor then wanted to know what treatment I was on and my laboratory values. I mentioned I was taking bicarbonate and I will never forget the words, 'has anyone ever mentioned Renal Tubular Acidosis to you?' (They hadn't). Although I was concerned at taking up too much time, the Doctor wanted to talk about the importance of Vitamin D, 'not that it is something that would be a problem in Australia', he said. Well it was my turn to teach him something. Australia has high levels of Vitamin D deficiency due to it being too hot to go out into the sun and because of the fear of cancers caused by the sun.

Arriving home, I had bad pain in my lower legs and bone scans revealed major hot spots in my hips, knees and shins. There was major concern about a stress fracture in my right hip, however following multiple MRIs and an X-Ray, an Orthopaedic Surgeon ruled it out. Next, I saw a Rheumatologist who raised concerns about my renal care, he told me that there are renal specialists who specialise in transplants and those who specialise in renal tubular dysfunction issues and asked which of these was looking after me. Both my Mum and I had been worrying about that for the past two years, as we felt that my specialist concentrated more on transplants. In addition, I had close to the record lowest Vitamin D level in the practice. Questions were raised about my Parathyroid not working properly. I was diagnosed with Osteopaenia, which can be a pre-cursor to Osteoporosis; I will need bone density scans

every two years. The doctor was not surprised by my pain considering how soft my bones must be.

It was about now I had time to breathe and remembered the words of the kidney doctor in the UK. What was this Renal Tubular Acidosis thing? Could I have it? Well bullseye, I have never seen my old kidney specialist again, supposedly the best in Queensland. The best in which area though? I had seen him every six months and my bicarbonate levels and urinalysis should have flagged up acidosis, yet nothing was done. For years, I kept telling him about my need and great love of drinking heaps of milk and asked whether that could have anything to do with my kidneys. He said no, I just like drinking milk. So maybe now you can appreciate how thankful I am to have had the opportunity of speaking to the doctor that day during my trip to the UK, because it is more than likely that in a year or two, the first I would have known about it would have been a broken hip. The pain did improve for a while but unfortunately it has worsened again. This is possibly because my Vitamin D dose was decreased too dramatically. At the moment, I am keeping up my milk intake, Vitamin D and am waiting to hear about Endocrine specialists with a particular specialty in bone metabolism.

And so the saga known as BBS or 'The Never Ending Story' continues...
Kathryn Murphy

BBS Clinics

Following another very busy and successful year, we find ourselves at the beginning of our fourth year working alongside the specialist NHS BBS Clinics, with two years remaining of the original five year contract. We have again seen an increase in patient numbers, however the rate of increase has slowed down. Having had around 90 new patients during years one and two, we have had just 40 new referrals over the past year, bringing the total number of affected adults and children to around 420. All four centres have booked additional clinic dates to cope with the increase in patient numbers and continue to run at around 18 monthly. We do ask that if you have any concerns, or feel that you or your child need to be seen sooner, please contact Tonia or Julie and we will do our best to book you in.

We continue to be supported by a wonderful small team of volunteer clinic support workers and our thanks go to them for their continued dedication and commitment. If you live reasonably close to London or Birmingham and feel you would enjoy this opportunity, please contact either Julie or Tonia for more information. Although this is a voluntary role, all expenses will of course be paid.

Patient feedback consistently rates the specialist service as being excellent with the majority of patients marking themselves as being 'very happy' with their experience. Much of our work is done outside of clinics: facilitating clinics, liaising with families and clinicians, producing newsletters and managing the database and website, however nothing gives greater satisfaction than a successful clinic day. It is so gratifying to know that we make a significant difference to patient experience, from supporting a patient through a blood test, making cups of tea, taking guide dogs for a walk, to ensuring patients know their benefits entitlement, our remit is broad.

We have developed a Benefit and Social Care Assessment, which enables us to identify where families or individuals need extra support. This is sent out prior to clinic and ensures we take the correct information/resources with us on the day. We have found that many are missing out on crucial benefits or lack adequate support, which highlights the importance of this aspect of the BBS specialist service.

In June 2012, all four centres came together once again to discuss best practice and results from the previous year and we presented a report based on patient experience and feedback. One of the issues we highlighted was that there seemed to be a lack of understanding of some aspects of the syndrome. We suggested that a training programme be developed for BBS Clinicians, in collaboration with adults with the Syndrome and their parents/carers covering the emotional/communication aspects of the syndrome and how this can be experienced, together with guiding skills and VI awareness. We are delighted that the team at Guys Hospital, London have successfully obtained a grant to begin this project and we will work with them throughout its development.

Our involvement continues in a research project aimed at linking genotype (genes) with phenotype (symptoms displayed). Many of you have kindly completed the questionnaire at clinic and we are starting to see promising results. The long-term aim of the study is to improve diagnosis, disease prediction and long-term management of the syndrome.

Looking ahead, the NSCT will be making way for the new NHS Commissioning Group and our funding and contract will be affected, although the service provided will remain unchanged for the time being.

We look forward to seeing you all soon.

Tonia Hymers
toniahymers@btinternet.com
01255 551886 or 07986 447429

Julie Sales
kevin.julie1@btinternet.com
01892 685311 or 07773 252585



Duxford Christmas Party

15th December 2012 was a very exciting day for five year old Savannah.

The LMBBS were invited to nominate families to attend a Christmas Party at Duxford American Airbase in Cambridge, along with many other children with disabilities and their families. The invitation was accepted on Savannah's behalf and, as her Grandmother, Marie, and Uncle Shaun will vouch, Savannah was treated like a 'little princess' from start to finish. Carers were there for every child, food was in abundance and all diets were catered for. At the end of the party, Father Christmas arrived with a named present for every child. It was a very special day and our thanks go to all those involved with the organisation of the event.



The Amazing Team Teague

As a thank you to the LMBBS, following an appointment at the specialist clinic at Great Ormond Street Hospital, Zoey and Gary Teague, along with their children Freddie and Maisie (AKA Team Teague) decided to embark on a nine month fundraising mission to raise £5000 for the

Society. They said, "We were taken back by the love and compassion shown towards us at a very difficult time by the members of Dr Beales team... ..we are on a mission to go completely and utterly bonkers to do anything we can to raise the most amount of money for LMBBS.... ..To give back and to be positive that future sciences and research could change this condition is amazing... ..Maisie is our true inspiration, a totally amazing little girl who has hopes and dreams like all of us who live life to the full, we are doing this for her.

The family began training and planned their journey for the coming year, which would include: the London Duathlon, the Henley Half-Marathon, the Wokingham Half-Marathon, and the Reading Half-Marathon, finishing with the Brighton Marathon in April 2013. Alongside this they planned further events such as a charity car boot sale, a spinathon, a race night/quiz night and possibly a charity recipe book.

Team Teague's 'marathon' year began with the London Duathlon, Zoey wrote, "I'm pleased to say that we came away with four medals. The day was a mix of emotions. Maisie and Freddie blew us away with their amazing determination, we were very proud of them. Gary was sensational and it was rather emotional watching him put himself through endurance that I've never seen before. As for myself, I got through it - just about!"

The next event for Zoey and Gary was the Henley Half-Marathon, however training was leaving them tired and sore. Maisie kept them going and ensured they persevered with their training; Zoey said, "She's one in a million, I could never get away without running on my running days....she's on my case!"



In the meantime, Team Teague's friends and family continued to support them, rising to the fundraising challenge and adding to their incredible amount raised; Leigh Morgan raised £360 competing in the Virgin London Triathlon, Lindsay Knox, and her gym group raised £5000 and Zoey's mum ran a sponsored spinathon at David Lloyd Gym, Reading. Members gave up their locker £1s and another £300 was raised for the Society. Zoey's Mum was 'chuffed to bits', so much so that she planned a sponsored rowathon for early the following year. Looking ahead, Gary's partner at work was committed to doing the London ride (100 miles) to raise more money for LMBBS.

Following the successful completion of the Henley Half-Marathon in October 2012, Zoey and Gary completed the Wokingham Half-Marathon in February 2013. This was to be followed by the Reading Half-Marathon before finishing with the 'Big One', the Brighton Marathon. Zoey in particular was finding it difficult, she said, "we have been running now for nearly seven months and can honestly say that I've never endured anything like this before; I'm tired, my body aches, I've had a few injuries along the way BUT I will flip the coin over and say that never have I done anything so rewarding in all my life. Surely if you take on a challenge in life it means just that, so I'm glad it hurts, I'm glad it's hard because at least we can say we went through hell to reach our final goal."

Alongside the training, Team Teague turned their attention to a big charity race night, to be held in March. They managed to get a free venue, their local Post Office Club, and planned entertainment, a small auction and 'the best raffle in town!' Mum of 'Team Teague' helped with the organising and Maisie planned on selling pop corn, sweets and cakes. They had an 'amazing evening' and raised a staggering £1218 taking their total to around £3600.

Back to the running, completion of the Reading Half-Marathon saw personal bests for both Gary and Zoey and their attention turned to the fast approaching Brighton Full Marathon. Zoey said, "To say I'm scared is an understatement, we will do it, through blood sweat and



tears. Maisie and Freddie will be waiting for us at the end.... they will be so proud of us, so that's what will get us through and with it being our last event...
...our kids waiting for us... priceless!

Our huge congratulations and heartfelt thanks go to Team Teague, Zoey's mum and all the friends and family who supported them over their Marathon Year. A truly inspirational group of people.



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Visually-impaired musicians' lives: Trajectories of musical practice, participation and learning

The “Visually-impaired musicians’ lives” (VIML) project is a UK national investigation into the musical practices, participation and learning experiences of blind and partially-sighted people. VIML is funded by the Arts and Humanities Research Council (AHRC) at the Institute of Education, University of London, and has two official partners, the Royal National Institute of Blind People (RNIB) and Royal Academy of Music, London.

Dr David Baker is the Project Leader and Professor Lucy Green his Co-Investigator and Mentor. Through an online questionnaire and confidential interviews, VIML will investigate the experiences of all types of visually-impaired musician, including instrumentalists, singers, composers and music teachers. We are interested in both amateurs and professionals, even adults who have just begun to learn an instrument. Participants will be invited to a conference, where findings will be shared, and exciting “music days” of practical music-making in London will be created as a collaborative undertaking with the Open Academy.

If you are visually-impaired and an instrumentalist, singer, composer, music teacher, or sighted and are working with visually-impaired musical learners, please visit the website below or make contact for further information.

Website and blog: <http://vimusicians.ioe.ac.uk>
E-mail: david.baker@ioe.ac.uk



Healthy Living: making good choices

Drinking enough of the right type of drinks is as important for our health as the food we eat. Recently lots of studies have made the news showing that drinking enough fluid can improve our concentration levels, exam success and well being for so many other reasons. When you think about it, it makes sense: our bodies are made up of about two-thirds water and so keeping this level balanced must be important.

How much should we drink a day?

A useful guide is to have at least six drinks each day. The size of these drinks increases as you grow from a small child to an adult. Six drinks means having a drink with your breakfast, mid-morning, lunch, mid-afternoon, evening meal and during the evening/before bed. In hotter weather we need more fluid than usual, use your thirst as a guide to ensure you drink enough.

Can our drink choices affect our weight?

Recent studies have shown that stopping sugary drinks and replacing them with water or sugar-free (diet) alternatives leads to weight loss and a reduction in body mass index (BMI). It is known that we don't alter the amount of food we eat if we have a higher or lower calorie drink. So if you have a sugary drink, such as a fizzy drink with your evening meal you will eat the same amount of food but also have the extra calories from the drink. This really adds up over time and your weight will steadily creep up.

So if you are keen to lose a little bit of weight or to start eating a bit more healthily in time for summer, why don't you think again about what you are drinking?

So what are the healthiest drinks?

Water is the best choice because it contains no calories at all, try experimenting:

- Try sparkling water if you like fizzy drinks.
- Add a slice of lemon or lime, it can make a huge difference to the taste. (Freeze slices of lemon and lime ready to use).
- Children may like fun ice cubes, try adding different fruit pieces to your ice cube tray to jazz up your drink, a few raspberries or grape pieces work well.
- Water is often the only drink allowed at school, if so, make water the only choice with your lunches at home too.

Milk is a good source of calcium. In our diets we need enough calcium to keep our bones healthy. Calcium rich foods include cheese, yoghurt, sardines, pilchards and calcium fortified foods including breakfast cereals and calcium enriched soya milk. Calcium is found in smaller amounts in many other foods, for example, bread, hummus and spinach. If you eat a varied diet you will not need to drink lots of milk to meet your calcium requirements. If you have concerns about whether you/your child is getting enough, chat to your clinic dietitian before changing your diet. So what sort of milk is best? If you are over 5 years of age, try to drink skimmed or 1% fat milk as these are the lowest energy (calorie) choices. If your child loves to drink milk and is under 5 years of age chat to your clinic dietitian as a lower fat milk

and a suitable vitamin supplement may be the best choice for your child to prevent too rapid a rate of weight gain.

Fruit juice and fruit smoothies are often perceived to be healthy choices but are they as good as you think? A small glass (150ml) of 100% fruit juice can count as 1 of your 5-a-day. If you drink extra this doesn't count as you are not eating the fibre found in fruit and vegetables. Smoothies sometimes count as 2 of your 5-a-day but this depends on its fruit pulp and juice content.

However these drinks contain naturally occurring fruit sugars so aren't low in calories. Did you know they have a very similar energy content to ordinary fizzy drinks? If you don't believe me, try squeezing oranges at home to see how many you need to get a 150ml glass of juice. It typically takes eight oranges but depends on the size and variety of orange you use. So it is easy to see how drinking juice means you consume far more fruit sugar than you would have eaten from the whole orange.

If you'd like to drink fruit juice or a smoothie, have it with your breakfast to help you absorb the iron from your breakfast cereal. It is worth diluting fruit juice with water to reduce its energy content and sticking to no more than 150ml of juice or smoothie a day.

Also watch out for "fruit drinks" as these can contain minimal amounts of fruit juice, lots of sugar and do not count towards your 5-a-day. Squashes too can contain large quantities of sugar, look for 'Sugar Free' or No Added Sugar'. Try to have squash very well diluted and only for occasional drinks. Another reason to avoid sugary or sweet drinks is they may make you want to eat other sweet foods and so you end up eating much more than you expected.

Fizzy drinks contain lots of sugar and usually nothing much else. Remember research has shown that people do not adjust the amount of food they eat when they have sugary drinks, so these just become extra calories and then weight gain follows. A can of fizzy drink typically contains 140 calories and 7 teaspoons of sugar. If you drink a can a day this means over a year you will have 51,100 calories from this fizzy drink. If you swap to drinking water or a diet fizzy drink you will lose weight (as long as everything else stays the same!) You should lose about a stone in weight over a year without too much hard work!

Tea and coffee are fine to include as part of a balanced diet in older children and adults. (However they should both be avoided in young children as they contain caffeine and can inhibit the absorption of iron from your food). To make these as healthy as possible use a lower fat milk, skimmed or 1% fat milk for example. It may interest you to know that a study found that most people found the taste of 1% fat milk as acceptable as semi-skimmed milk so why don't you swap today!

Watch out for high street coffees, lattes for example, as these have many more calories than you would expect due to their size and the amount of milk they contain. Always ask for a skinny coffee so skimmed milk is used. Tea can be the best option in coffee shops as you add less milk to tea so will have a cheaper and lower calorie option too! They also do lots of speciality teas that can be taken black, why don't you try something new?

What about energy and sports drink?

Energy drinks are best avoided! They often contain high levels of caffeine and also lots of sugar which means lots of calories. (They are not suitable for children.) Sports drinks are only needed if you are doing endurance training. After exercise it is best to rehydrate with water, otherwise you may find you undo all your hard work.

What about alcohol?

Obviously this section is written for adults only. Calories from alcohol can quickly add up! Did you know that an average wine drinker in England consumes 2,000 alcohol calories each month? One in three drinkers increase their energy intake by having snacks such as crisps and nuts with their drink. A heavy drinking session is also followed with a less healthy breakfast the next day by 60% of drinkers for example, a fry up to help ease their hangover. Does any of this sound familiar?

If so the following tips may help you cut back the calories you are getting from alcohol:

- ✓ Try alternating an alcoholic drink with water or a diet soft drink on a night out
- ✓ Try stretching your drinks by adding a splash of soda water to white wine to make a spritzer, or have extra diet cola or mixers with your gin or vodka
- ✓ Have a healthy dinner before you start drinking so snacks are less tempting!
- ✓ Remember the government recommendations for safer drinking: men should have a maximum of 3-4 units of alcohol and for women the maximum is 2-3 units a day. (A 175ml glass of wine typically contains 2.1 units and a pint of lager/beer/cider 2-3 units).
- ✓ Have a couple of days off alcohol each week and if you've had a heavy drinking session avoid alcohol for at least 48 hours.

If you are trying to lose weight, looking at your alcohol intake and how you eat when drinking, or the day after, really may help. If you do drink and eat more than usual at a special event like the family conference weekend, try to get back to your regular diet as soon as you are fully recovered!

Remember:

Changing any aspect of your diet should be done gradually. Make one change at a time; you could start by drinking water with your lunch. After a couple of weeks this will have become a habit and then you can think about the next change you'd like to make.

If you do have a day when you've drunk more sugary or alcoholic drinks than usual, don't give up, just restart with your healthier diet and drink choices the next day.

Sarah Flack

Principal Dietitian, Great Ormond Street Hospital, London

If you would like to chat to Sarah please call her on 020 7405 9200 extn 5941 on a Thursday or a Friday

Change of Address

If you move house, please remember to pass on your new address to everyone involved in your or your child's health care. Many people don't realise that the computer system at their GP surgery is not linked to the hospital where they receive specialist care. Likewise, if a patient notifies their hospital, this information will not necessarily be passed on to other health care bodies. So, if you move, let your GP know, let your hospital(s) know and let us at the LMBBS know.

Blind Football Manchester

LMBBS member Liam is involved with a new Blind Football Team in Manchester and they need new members:

“Currently we are a little group and only have a handful of players and would like to encourage more people to join our group and play blind football in Manchester. We have a committee and two trainers/volunteers and we train on a Thursday evening from 7pm till 8pm at North Manchester Academy Sports Hall, Queens Road, Manchester.”

Blind Football is a very exciting game and is becoming extremely popular throughout the UK.



If you live in Manchester and would like to have a go, contact Ruth Walmsley on 07780 466 410 for more information.

Go Ellie

Need some inspiration? Check out Ellie Jones, general legend and part of the Allstars Squad at Usk Tennis Club. Recently, she has reached a rally of ten after three months practice.

What made this an exceptional achievement is that Ellie doesn't watch the ball - she listens to it!

With special modified tennis balls with bells inside, partially sighted Ellie times her shots to the noise the

ball makes when it bounces.

She is currently working on her serve and has already aced coach, Rhys Jones a few times! Keep up the good work Ellie.



Well Done Katie

Awe inspiring Katie Oates, 14, took part in the British Gas half mile Great North Swim in Lake Windermere in June 2012. Katie, who has chronic renal failure, was competing as a personal challenge and her friend, India, was due to swim too as support for Katie. Devastatingly, just two weeks before the swim, India was diagnosed with Lymphoma and had to pull out. Determined to do her friend proud, Katie swam with her mum, Emma, and together with another friend, they raised £1300 in ten days for the Teenage Cancer Trust. The family were followed all weekend

by the BBC who televised Katie's story. Katie met Cassie Pattern, Olympic bronze medal long distance open water swimmer, Steve Parry, Olympic swimmer and Blue Peter's Helen Skelton. We are delighted that India will be competing in the Great North Swim this year with Katie... a remarkable feat, considering India's journey over the past year. We wish these truly inspirational young ladies all the very best for their swim and for the future.



Are you ready for some serious 'Sports without Limits'?

Do you fancy being part of a team and enjoying the buzz of match victory? Want to get out and get fit while meeting new people? Is football, cricket, goal ball, judo or tennis your thing? Then try out the Royal London Society for Blind People's (RLSB) sports programme for vision impaired young people aged 11-25.

'Sports without Limits' offer lots of opportunities for young people who go to mainstream schools to get into sports, with sessions taking place in London, Kent and Sussex. Their coaches use adapted equipment and specialist VI training methods to teach cricket, football, goalball, judo, athletics and tennis from beginners level, right up to national level. 'Sports without Limits' is free; they feel that every child, regardless of their ability should be able to experience the thrill and excitement that taking up a sport can bring.

At a Sports without Limits cricket session, you will be taught how to bowl a V.I. cricket ball, how to catch and throw, and of course, how to bat. The communication and team work skills involved in blind cricket are vital, and play a key part in their sessions. RLSB also run free social groups in London. Sailing, Rock climbing and Tandem Biking are just a few of the activities they offer.

In their goalball sessions, you will learn how to defend your own goal, as well as how to effectively shoot at the opposing team's goal. Your team work and communication skills will also improve, as these skills are very important in goalball, as they are in all blind or visually impaired team sports. Judo and martial arts in general, can be inaccessible to people with a visual impairment, however RLSB's experienced coaches run safe and enjoyable sessions where you will learn how to perform a number of judo throws safely, as well as how to land safely yourself!

The V.I. football sessions will teach you how to control and pass the ball, how to tackle safely and effectively, and how to dribble and shoot! Their sessions always include a match, so you will also get the chance to develop your positioning and a better understanding of how the game works.

The young people attending sports sessions help to decide which sports they offer, so expect to see swimming, archery, shooting and tennis on the programme soon.

For more information about RLSB and their sports programme, please go to the RLSB website: www.rlsb.org.uk

We hope you have enjoyed this newsletter, don't forget, all of the contact details can be found at the beginning.

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