



# LMBBS Winter Newsletter 2013-2014

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## Foreword

First of all, I want to thank everyone for voting for me at this year's AGM and enabling me to become Vice-Chairman. It is a role that I hope to make my own and I look forward to supporting the rest of the committee in the vital work that they do all year round.

Well I am sure that those of you who know me well, know that I am not one of those people who stays still for very long, and this year has been no exception.

The summer saw me travelling solo once more to the USA, to San Francisco, to see my Father and meet with my younger brother, Timmy, who lives in Sydney.

The holiday was amazing and saw me taking in the music scene of down town San Francisco and also the dizzy heights of the Sentinel Dome locked within the centre of Yosemite National park.

At the moment, I am busy preparing for the launch of the DAB radio station which I run in Surrey called Redstone FM. We are launching our DAB service in December and I am very excited about this.

The year has not been without its BBS challenges, but I never let this stop me from living life to the full.

The LMBBS Committee and I are now in the process of planning the Family Conference for 2014 and hope to see all of you next year. Whether you are a newly diagnosed family or old timer like myself, the conference is a magical weekend of love and support and is a chance to make new friends, who before long you will know as your LMBBS family. Finally, stay warm this winter and I look forward to seeing you all at Conference.

Steve Burge  
Vice Chairman.

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## Congratulations!



Our congratulations go to Bob and Jill Staples who celebrated two special occasions this year; Bob turned 80 in June and together they celebrated their 30<sup>th</sup> wedding anniversary in July.

Jill writes, "On July 6<sup>th</sup>, a party celebrating both occasions took place for 196 family and friends. Several friends from LMBBS were there including Marie, Shaun and Suvannah Owen who had travelled from Wolverhampton and Ian McDonald and Pauline Reynolds and family. Also there of course was my nephew, Stephen with his carer, Kelly. A good time was had by all with plenty of food and an excellent disco. I gave Bob a few surprises by tracing many of his old Boys Brigade lads and some of his old officers who turned up to celebrate with us. In total, £500 was raised in donations instead of presents

and we would like to thank everyone for coming and giving generously



Family Fund is a registered UK charity which has provided grants for low-income families, raising children aged 17 and under, for over 40 years. It can help with essential items such as washing machines, fridges and clothing but can also consider grants for sensory toys, computers and much needed family breaks together.

The Family Fund website ([www.familyfund.org.uk](http://www.familyfund.org.uk)) is very informative and apart from telling you where you might be able to make savings you hadn't thought of before, it also makes you aware of other agencies that could help your family.

They may not be able to help you with everything you ask for, but they are certainly worth a look.

If any of you have benefitted from the Family Fund and would like to share your experience, or if you know of other groups that can help, especially ones that support all age groups, we would love to hear from you.



### My Journey by Ollie Sloane

I became interested in fundraising after going to the LMBBS Conference two years ago. I've always struggled with being visually impaired, however with the conference and clinics in London, I find some hope and inspiration. Following a parent's suggestion at the Conference, I now do archery. I played VI football for five or six years, but the blindfolds kept giving me eye infections, so I've had a break from it this past year. After listening to Hannah Young, who spoke at last year's Conference, I've also considered cricket, although I haven't been along yet.

I also enjoy the LMBBS Facebook page and want to say 'Hi' to Nicola, who I started chatting to after we found out we were both doing 'The Great North Run'.

I have already done a couple of quizzes and card sales which my mum helps with (thanks also to Julie and Hollie for helping to make some cards).

In September, with the help of my brother, Josh, I completed the Great North Run. My family and aunt and uncle all went up for the weekend and we made a great weekend of it. We met up with Dan from Birmingham at the starting line; I wonder how he did, our time was 2 hours 51 minutes.



It was absolutely amazing. For those who watched it, we were on TV waiting to start behind David Weir and Tirunesh Dibaba. We had orange baseball caps on and lots of our friends said they saw us. Mo Farrah started his race half an hour after us and it was fantastic to have him race past us after only three miles. Not many people can say they've had him sprint past them. We were interviewed by the Newcastle Chronicle - did any of you see it!

We raised £600, so a big thank you to the people who gave me money at the Conference and also to those who donated online.

I am planning on doing a quiz night in February, we've done these for the past few years and it is a great evening with lovely raffle prizes. If anybody lives near, or in Kent and wishes to come, my email address is: [oliejhon@msn.com](mailto:oliejhon@msn.com).

Looking forward to seeing everybody next year.

Ollie

## **Organise! Request! Use! Go!**

### Top Tips for Parents or Carers of Children with Special Needs

As a parent or carer of a child with special needs, you will no doubt find yourself attending many meetings relating to medical and educational issues. Before these appointments, many parents feel unprepared or feel that they have a lack of control about what is going on, which can lead to anxiety and frustration. Feeling like this can minimise the benefit that these important meetings have and so the following suggestions are designed to boost confidence and lead to a more productive encounter.

### **Organise**

It is essential that you keep all documents for future reference.

- ✓ Buy a ring binder in which you can keep any reports you receive. Sharing knowledge of your child's health may be important so you need to have the information to hand.
- ✓ Insert a calendar at the front of the folder for marking things like medical appointments, medication and periods of illness.

- ✓ Have a second binder for educational issues such as Individual Education Programmes (IEPs), home visit reports and reports from agencies such as Occupational Health, Physiotherapy and the Sensory Support Service.
- ✓ If possible, keep an on-going diary that is an overview of all issues.
- ✓ Keep these files in an accessible place so they can be updated easily.
- ✓ Include a 'Questions' page at the front of the file so you can jot down any questions as they arise. This will serve as a reminder of what you want to ask when you next go to an appointment/meeting.

### **Request**

- ✓ Always request that you receive copies of documents or reports from doctors and therapy providers. It is not uncommon to have to chase these up, so be persistent because it is essential that you have a record of what is going on with your child.
- ✓ Make sure you receive updated IEPs and school reports so that you can monitor your child's progress.
- ✓ Ask school when you can expect things like IEPs, reports and parents meetings so that you know when these things are due.
- ✓ When necessary, request a meeting with your child's teacher or SENCo so that you can share relevant information that schools will need in order to update or modify any programmes of work or support plans.

### **Use**

- ✓ Look into what is offered from support groups in your area. They may have ideas that will help parents and carers as well as your child.
- ✓ Find out about any grants that may be available.

### **Go**

- ✓ Take your Medical and/or Education file with you when you go to a meeting so you always have the information you may need to refer to.
- ✓ If you have been compiling a list of questions, remember to take it with you to any meeting.
- ✓ Take a notepad and pen with you to any meeting so you can jot down any notes and write reminders of things that you will need to follow up.
- ✓ If possible take someone with you to meetings. It's a good idea to have someone there for support and to talk things through afterwards. They may also remember things that were said that you missed.

Medical and educational meetings can be very overwhelming, but they are also essential. Through good organisation and thorough preparation, we can maximise their usefulness to the full. Finally, remember to keep notes, because no doubt you will need to refer back to what happened at these meetings at some stage in the future; it is so easy to forget details.

Hopefully these suggestions will help you, the parents and carers, to feel more in control and support you in your role as advocate for your children. If any of you have any helpful tips you would like to pass on to other parents, we would love to hear from you.

Dianne Hand  
LMBBS Mum and SENCo

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## 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 a big 'thank you' to Claire Gillman, Karen Masters and their employer, Barclays Bank. Claire and Karen, both members of the Conference Childcare Team have raised a considerable amount of money between them over the years, holding parties and raffles, which has all been matched by Barclays, £1 for £1. In fact, Barclays' involvement goes back around fifteen years, when they donated £500 to the LMBBS to pay for the Conference Creche toys. We remember having a great time choosing everything from the Early Learning catalogue and the excitement on delivery day! It was money well spent as the toys are loved and played with at every conference before being carefully cleaned and packed away ready for another year. Thank you Barclays and thank you Claire and Karen.



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## Volunteers Please!

Have you a spare day or even a few hours once a year and would like to do something to help the LMBBS? Then Sight Village may be for you. Sight Village is a two-day exhibition, in Birmingham and London, offering the latest in everyday living aids, technology, specialist holidays, general advice and much more.

Sight Village is held in Birmingham every July and in London every November and we try to have an LMBBS Stand at one or both events every year, to raise awareness of the syndrome; it is a great event and one we recommend both to visit and to attend as an LMBBS representative.

Exhibitors, including large and small Charities as well as the major exhibitors in the field of vision, travel from all parts of the UK. The atmosphere is friendly and 6<sup>th</sup> Form Volunteer Guides are provided by the local school. There is always time to look around the exhibition and find out about the latest technology and it can be very fulfilling, chatting to the visitors to the LMBBS stand, who are often teachers or social workers, supporting people with the syndrome wanting to extend their knowledge.

To all our volunteers who made it possible for the LMBBS to attend Birmingham and London Sight Village in 2013, a big thank you. If any of you are interested in helping out in 2014,

please contact Chris Humphreys at [chris.humphreys4@ntlworld.com](mailto:chris.humphreys4@ntlworld.com) or by telephone on 01633 718415 and she will be only to be pleased to provide further information.

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## Sports Day Success



My name is Jules Thomas and I am the Chairman of Dis-Sport Beavers, which is a local disabled sports club in Basingstoke, which meets weekly and caters for all types of disabled people. In association with another charity called the DD Hampshire Games Association, I have been running our local sports day for the past 14 years. It has actually been going for 38 years and is held at a local comprehensive school. Unfortunately, the previous committee were getting too old and wanted to stop running the Games, so as Chairman of Dis-Sport Beavers, I offered to take on the running of it.

The games were started in 1975 by a local business man called Douglas Rowsam and this year was our 38th year. We had about 86 competitors who came from all over the South of England including Gosport, Southampton and Farnborough and we had nine different teams and quite a few individuals from Basingstoke as well. Competitors took part in 17 different events ranging from field events such as Shot, Discus, Club, Precision Javelin, Petanque, Skittles, Table Games and so on. The Mayor and the President of the Rotary presented the medals and trophies to all the competitors.

We couldn't put on this event without the volunteer helpers from Rotary, Basingstoke Lions, Loddon Valley Lions and Unicorn, as well as all the individual helpers. At the moment I am busy trying to fund raise, because it costs us about £1400 to run it all, as we have to pay for medals, venue hire, paramedic in case of any accidents, equipment and so on.

Everyone that came this year really enjoyed themselves and seemed to have a really great time. The weather kept fine for us, the Rotary did a BBQ and everyone is looking forward to coming back again next year.

Jules Thomas

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## The Great North Run



I actually did it! I ran the Great North Run!

For anyone who knows me, I am most definitely not a runner, BUT I loved every minute of it and raised over £500 for LMBBS and over £500 for RNIB. My children came along and cheered me on at the 8 mile mark, which kept me going and reminded me why I was doing it when it was getting tough.

For anyone thinking about doing the Great North Run, or any other marathon, you should give it a go, the

atmosphere is amazing! I felt proud to be part of an amazing event and so pleased to have raised over £1000 for two great charities.

Nicola Jordan

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## Health and Wellbeing: Nutrition Labelling

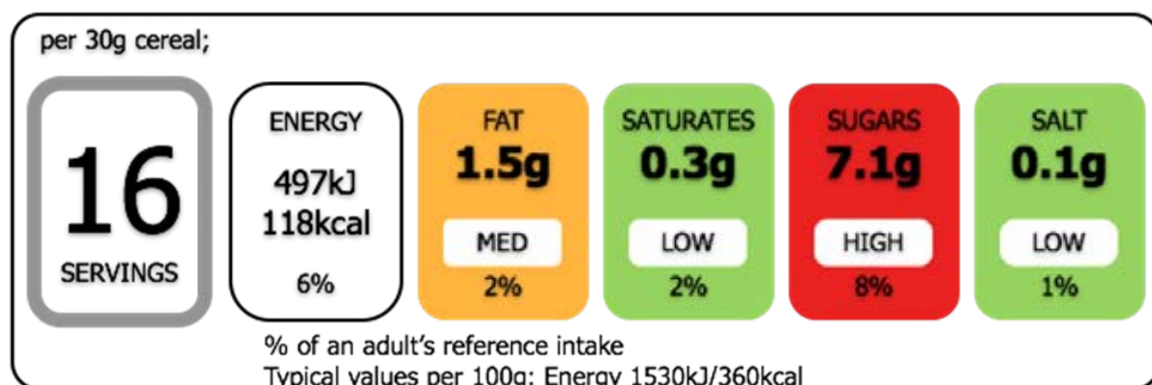
I have been asked several times recently, how to use the nutritional data on food packs, so thought it would make a good newsletter topic. However I do realize, for those of you with vision issues this may be difficult, but hopefully the end section will have some useful reminders for us all.

How do we tell which “ready meal” is the better choice? Currently the nutritional data on the front of a food label is presented in a variety of ways. Some manufacturers comment on total sugars in a product, others only about the added sugar; which makes it very hard to compare products. The colours used to indicate whether a food contains a small or large amount of fat or sugar are not consistent. Unsurprisingly research has shown that we can misunderstand this nutritional information. Some shoppers don’t look at the nutritional information as it takes too long to compare products and after looking it can be too hard to decide which is the healthiest option anyway!

For people wanting clearer nutritional information about the food they buy, there is now some good news. All major UK supermarkets and many food manufacturers have agreed to introduce a new consistent front-of-pack nutrition label. Current estimates are that over 60% of food producers will be on board with this campaign and the government hopes that others will join the scheme in the future. Two major companies not taking part at this time are Coca Cola and Cadburys.

You may have already noticed products with these new labels. Manufacturers have agreed to introduce these front-of pack nutrition labels by December 2014.

### So what information will be on the new food label?



### Portion or serving size

Clear information will be provided for the portion size, for example, a quarter of a pie or one burger. Some foods like bread have variable portion sizes. In this case the details will usually be given per slice.

Use this information to check whether you were planning to eat more than the manufacturers' recommended portion size. Remember these portion sizes are for adults so for children portion sizes will need to be adjusted to ensure they are suitable for their age.

### **Energy**

The energy content of the food will be given for both the serving size and per 100g. It is important to notice that the first figure will be given in kilojoules (the metric measurement for energy). Most of us are more familiar with calories (short for kilocalories) - so look for the bottom or smaller number on the label. It is worth noting that 1 calorie is the same as 4.2 kilojoules to avoid any unnecessary surprises!

### **Fat and Saturated Fat**

Information will be given for both total fat and saturated fat. Too high an intake of saturated fat has been linked with high cholesterol levels and heart disease. As a reminder, saturated fats include harder fats like lard, butter and fat on meat. These are found in fatty meat products, pastries, cakes and biscuits.

### **Sugars**

The added sugar in a product will be listed on the front of the pack - this will make it possible to compare products.

### **Salt**

Information will be recorded for salt - much more useful than sodium which is currently on many food labels.

On the new labeling system, the fat, saturates, sugars and salt is colour coded either green, amber or red. So just like traffic lights, green means "go", so green choices can be eaten more freely but if a food has many red labels stop to think about your portion size first!

A chocolate flavoured breakfast cereal has a red labelled sugar content warning that this is a high sugar food. It has an amber labelled fat content, showing it has a moderate fat content. If this was your or your child's favourite cereal, you could look for a breakfast cereal with more green labels to eat more regularly. A couple of ways to reduce intake of this cereal would be:

Try eating an unsweetened cereal like porridge, wholegrain wheat biscuit style cereal, bran flakes, cornflakes or rice pops on week days or school days. Weekend and holiday breakfasts could be this chocolate flavoured breakfast cereal.

Or you could try mixing a variety of cereals together to reduce the amount of chocolate flavoured cereal eaten.

### **So what key things should I look out for on the front label on food packaging?**

First check the colours - all green, put it in your shopping basket! Some red labels - have a look at how much of your reference intake the product provides. (The reference intake is the recommended daily maximum for an adult). For example, honey, jams and marmalades would all have a red label for sugar content. If you are only going to have it on a piece of toast at breakfast - this is fine to eat as part of a balanced diet.

If the food has lots of amber labels - this is fine to eat most of the time. If there are lots of red labels, think about the food and your portion size! Sometimes it would be worth looking to see if there is a similar but healthier choice available. Other times it is important to enjoy the food, as it may contain lots of other important nutrients which are needed in our diets - but just watch your portion size!



Eating a healthy, well balanced diet is never all about the colours of the labels on our food. It is still important to look at your diet as a whole and consider the following.

Eat a range of foods from the different food groups to ensure you have a balanced diet:

- 1 Have at least five portions of different fruit and vegetables each day. Try to include different coloured fruits and vegetables – this will ensure a good mix of vitamins and minerals in your diet. Remember to try to cover half of your plate with these foods at lunch and in the evening.
- 2 Have a quarter plate of meat, fish, eggs or pulses (beans and lentils) at lunch and your evening meal. If you eat fish - try to have two servings per week. Choose an oily fish (mackerel, trout, sardines, salmon or fresh tuna) as one serving per week.
- 3 Try to include adequate amounts of dairy foods - milk, yoghurt and cheese. (If you can't tolerate milk ensure you choose a calcium enriched milk replacement). For those over five years of age try to choose lower fat options, for example, 1% fat or skimmed milk, low fat yoghurt, cottage cheese and reduced fat cheese or cheese spread. If you have a younger child chat to your dietitian, as a vitamin supplement may be needed with these lower fat options.
- 4 Aim to include one portion of a starchy food like bread, chapattis, rice, noodles, potatoes, breakfast cereals and pasta at each main meal. One portion should cover no more than a quarter of your plate for lunch and evening meals.

Remember to enjoy eating but try to keep portions moderate. Only eat if you feel hungry and stop if you feel full! If you think you are hungry but haven't had much to drink - try a drink first! Try to have at least six to eight drinks every day.

Exercise and activity is important too - try to do something active that you enjoy every day.

Sarah Flack

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## News from Scotland

Committee member, Alan Clark, attended the first meeting of the Cross Party Group on Rare Diseases during the summer. Five Members of Scottish Parliament (MSP) attended, including Alan's local MSP, Jackie Baillie.

The proposed purpose of the group is:

- To act as a channel of communications between the Scottish Parliament and families affected by rare diseases in Scotland.



- To act as a channel of communications between the Scottish Parliament and people and organisations working in rare disease research, treatment, care and prevention.
- To monitor the development and implementation of a Strategy for Rare Diseases in Scotland.
- To identify areas where inequalities exist in the provision of care and campaign for their improvement.
- To examine areas of policy or service provision relating to rare diseases in Scotland.

Alan is representing LMBBS within this working group and is very much looking forward to getting involved and also to raising the profile of the syndrome. More information can be found at [www.scottish.parliament.uk/msps/67466.aspx](http://www.scottish.parliament.uk/msps/67466.aspx).



### Fundraising Round Up

First of all, a huge 'Thank You' to everyone who has raised funds, donated and helped in any way over the past year – what a generous bunch you are.

Thank you to the walkers, runners, swimmers, cyclists and party animals, you have been amazing. Ollie Sloane and Nicola Jordan took part in the Great North Run – you can read about their experience elsewhere in the newsletter. On what was one of the worst downpours in August 2012, the Usk Cricket Team had to call off their charity match, however the baking of Ellie Jones and her band of helpers saved the day and the sale of their cakes raised a whopping £125 for the Society, while the forlorn members and supporters were left commiserating in the Club House. Well done to all concerned and we hope the sun shines for you next year.

We are very grateful to have received donations from the Bor Charitable family Trust and The United Church, Kingswood as well as from many generous individuals. Tom Oates got a nice surprise at his Year 11 Prom this year. The students of the Prom Committee decided that LMBBS would be their chosen charity and Tom was presented with a cheque for £250. What a lovely group of young people- many thanks to you all.



In the last newsletter, Chris Humphreys challenged all our members to a blindfold bowling event in September, however it was not the success we had hoped for, with only one or two families taking part. Chris, together with her family and friends held their own blindfold bowling fundraiser on 24<sup>th</sup> September 2013 and had a great time. Chris said, "there was lots of laughter, especially in the blindfolded game, when the bowls jumped from lane to lane... ..we raised the fantastic sum of £1005, which included a donation of £110 from Bowlplex. We aim to make the evening an annual event." Imagine if just nine other families followed in their footsteps... together they would make over £10,000 for the Society.

Elsewhere, Edward Evans, brother of Joseph, undertook the 27 mile Jurassic Coast Trek to raise funds for LMBBS. His employer, Bibby Finance, matched the amount raised, bringing the total to £664 for LMBBS. Our grateful thanks go to Edward and Bibby Finance. The staff and students of Newson Academy of Performing Arts collected funds for the Society during one of their fabulous productions in the summer – thanks guys, keep up the good work.

Beth Hoskins completed her first marathon in the States for the Society, cheered on by Helen May-Simera. Beth works in Professor Beales' laboratory testing DNA for BBS gene mutations, so not only is she working hard for LMBBS in the lab, but she is fundraising for us too – thanks Beth, you are a star! Beth has written about her marathon experience in this newsletter.

Our loyal band of 'Friends' continue to give valuable support to the Society, and we thank them for their commitment. There are different ways of becoming a 'Friend', some set up a standing order from as little as £2 per month, some pledge to fundraise in some way and others have a collection box and save their pennies. Our membership is growing, but very slowly and we desperately need more Friends to ensure a secure future for the Society. The Friends scheme is of vital importance as the standing orders and fundraising provide a reliable income, which is used in many different ways. If you or someone you know would like to become a Friend of the LMBBS, please complete the form which can be found within this newsletter.



We were delighted to receive a grant of £3,500 from Children In Need in April towards the cost of the children's activities at our conference weekend; the children and young adults had a great time and we are extremely grateful to Children In Need for their contribution. We are also excited to be shortlisted for a grant from

Genetic Disorders UK (Jeans 4 Genes) for £16,000 which would enable us to employ a Children's Development Officer. We would really like to bring someone on board to support our families with accessing local services, to ensure they are well supported in the home and at school. Tonia Hymers and Julie Sales presented our application to a panel of around ten individuals from across many sectors and we will know the outcome in January. If we are unsuccessful, we will try again next year. In the meantime, we are always on the lookout for more grants to apply for, so if you hear of any, which you think could suit us, please let us know.



Looking ahead to the New Year, for those of you who would like to do something to raise funds but are unsure of where to start, how about holding an Afternoon Tea for LMBBS? February always needs cheering up and what could be more cheery than tea and cake with friends? If you are associated with a school, church or group such as Brownies or Cubs, perhaps they would be happy to join in. All you need is facilities for having a cup of tea or coffee and a chat. We can supply leaflets, balloons and stickers as well as small collection boxes, so go for it, get fundraising and have a great time in the process!

We are delighted to introduce our new Fundraising Co-ordinator, Emma Oates, who will take over this page from our next newsletter. Emma has two children affected by LMBBS, Tom and Katie and has been a member of our committee for 18 months. We are sure Emma will bring a fresh perspective to the role and look forward to supporting her over the coming year. Emma's contact details are on the back of the newsletter.

Finally, we wish that we could name all of you who have done so much to help the Society, without every single one of you, the Society could not continue the valuable work that it does. Keep up the good work!

## Your Society Needs You



Over the past three years, the LMBB Society membership has grown hugely, largely due to the success of the specialist clinics. With a growing and diverse membership spread across all of England, Scotland, Wales and Ireland, not to mention Europe and the rest of the world, it is hard for one committee to look after the needs of everyone as well as to keep driving the charity forward to bigger and better things.

If you feel you have skills that could benefit the Society, please do not hesitate in coming forward, it is far better to have many people contributing a small amount than a few people trying to do too much. For more information or to express an interest, please email: [toniahymers@btinternet.com](mailto:toniahymers@btinternet.com).

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## Our Very Positive Perspective



We didn't know anything was "wrong" with Evie until a visit to her eye consultant last year. She had been wearing glasses for short sightedness for over six months and despite being told that it was very rare for a child of this age to be short sighted, we just thought that was it. Her eye consultant had remembered a syndrome called Bardet-Biedl syndrome or BBS and thought Evie had a few markers of this. He knew that she had an extra toe on her left foot, complete with bone and nail, which was removed when she was 15 months old. Other indications were her vision issues, her being behind on her milestones and her being a big girl for her age. After the appointment he said that he would refer us to a geneticist.

When we got home we got straight onto Google (like you do, but know you shouldn't!) We scared ourselves stupid! Blindness? Renal problems? Renal failure? Heart problems? Fertility issues? Diabetes? Speech development issues (well, that one made sense) I cried.....a lot. How had this happened? After wanting a baby for so long, now there was a chance that she could have some serious problems.

I was first told that the geneticist appointment would be the following Spring, ten months away. I quickly got that sorted by being a very overbearing Mummy! November - that would be when we went. Meanwhile I phoned my local GP and told him that I was worried that if there were any issues with her kidneys, then it was surely best to find out now. He agreed and sorted out a blood test for her, which came back clear, so at least that was a relief.

Talking to people at the time was incredibly difficult, I tried to stay a little optimistic, but my husband just knew that she had it. Friends and family didn't think anything was wrong and tried to reassure us that she was just a bit slower than other children (I hate that phrase!)

November eventually came around. The visit itself started off on the wrong foot, as I could not park anywhere. I had to drop John and Evie off, park in a street ten minutes away and run back! We saw an ophthalmologist first and Evie couldn't have cared less, she doesn't like eye appointments one bit. When we were then told that she was going to have drops put

in, I nearly lost it! Why are we messing around having her eyes examined, when this was supposed to be about getting some answers?

They didn't put drops in, I think I scared them, whoops! I just didn't see the point in getting my baby all upset and worked up when she didn't need something doing.

We got called in to see the geneticist and he took some history from both of us whilst Evie played happily on the floor with a student doctor. After going through everything he basically said that yes, it was very probable that she had it. I looked over at John and broke down. Tissues were passed round and Evie, oblivious to it all, came over and gave me a pair of glasses to try on! Eventually, as the tears dried a little I asked if there was a blood test that could be done (I already knew there was from asking other BBS parents on a Facebook page), but he said no, there was no blood test. We were told about a clinic at Great Ormond Street, which again I knew of as I had already started speaking to lots of families and had been put in touch with the LMBBS charity. So we left, our hearts heavy.

That night, I couldn't sleep. How could I, with all the worries still fresh in my head? Turns out that Little Miss was wide awake too, so I went into her room. She stood up at the side of her cot and I held her and sobbed. I apologised to her over and over again. She just enjoyed the cuddles in the middle of the night!

Over the next few weeks, things got a little easier, the tears dried up more and I put away my self-pity and sorrow, after all, where was that going to get me? We still had our beautiful, funny, caring, cheeky little girl and nothing was going to change that.

I got in touch with two wonderful people from the LMBBS charity. They were a huge source of support to me and put me in contact with even more families. Sometimes I think the hardest thing for people is thinking that you are alone and that is where the charity comes into its own. I started organising a trip to GOSH (that sounded very scary at first!)

We went to London in March this year. The BBS clinic is held over two days, the first is for all eye related exams and blood tests and the second is about seeing a whole host of professionals. We got the train and I have to say that Evie was an absolute star for the entire journey, she loved watching the world go by. She was less happy about being bundled into her pushchair when we got to King's Cross Station! We walked to the patient accommodation, dropped off our case and went over the road and into Great Ormond Street. Wow, what a hospital! Within a few minutes of getting there we were called into the first appointment. The professionals kind of ignore you and concentrate solely on your child, which I think is fantastic. Evie was very shy at first. They were showing her a few DVDs and asking if she liked them, she kept saying "no" to everything, until a Peppa Pig one was placed before her. She grabbed it and said "wow!" Good old Peppa!

This exam was for an ERG, I didn't think we had a chance at completing this, but she sat quite happily on my knee whilst the DVD was put on. Electrodes were then placed on her head and they started the test. She did so well, we eventually had to open a packet of magic chocolate buttons to keep her still, but she really did a thousand times better than we ever could have imagined. We had several appointments, one was for eye drops, oh no! Again, she was a star. I don't know if it was because she was more relaxed or because the staff there are just better at dealing with kids, they really are brilliant. The result of the ERG was discussed with the consultant and it confirmed that she had Rod Cone Dystrophy, just another stepping stone to a clinical diagnosis.

The blood test didn't quite go smoothly, but they did manage to get some and then showered her in stickers (which are one of her very favourite things) and gave her a choice of toy out of a box. Again, it's the little things like that that make GOSH so special. We had a lovely tea at Pizza Express and a bit of a walk afterwards. She LOVED London, the noises, the lights, the buses!

The next morning we went across the road again, this time for a day of meetings with various professionals. A nephrologist, dietician, speech and language therapist, clinical psychologist, endocrinologist and the best of the best, a geneticist. Now this is where I have to say that Professor Beales, the leading geneticist in BBS, is amazing and he is doing some fantastic work. It is down to him that these wonderful clinics in London and Birmingham exist and after seeing Evie, he was happy to give a clinical diagnosis of BBS. It was a very long, tiring and emotional couple of days, but at the same time, wonderful. All these people that knew what BBS was!

We were stopping another night in London as we felt it would be too much for her to get the train back, so off we went to Covent Garden and spoilt her rotten! We 'Built a Bear', spent a fortune in the Disney Store, had ice cream, met with a Facebook friend and took her to M&M world.

Since Evie's clinical diagnosis, we have had so much support. She has her regular eye appointments, she has had a hearing check, the speech and language therapists visit her regularly at preschool, she has had a kidney scan (which showed her kidneys are fine at the moment, phew!) We have a paediatrician on board now who is great for coordinating everyone and we see an early years specialist for children with visual impairment. In fact, next week, both the early years specialist and speech and language therapist are doing an observation at preschool, talk about being the centre of attention!

In April this year we went to the annual BBS Conference in Northampton where I was finally able to meet many of the people I had been talking to online, what a miserable bunch they are!!! Of course I am kidding. They are all so lovely and it sounds a huge cliché, but it was like we were all part of a great big family. No small talk was needed about what BBS was, everyone just knew and knew more than we did. I was actually really sad to leave them on the Sunday, I enjoyed being in our little BBS bubble together!

I will wrap this up for now, man I can waffle! I still have sad days, things can catch me unaware. An episode of Bones that I've ended up watching twice very close together is about a baby that has a chance of being born blind, recessive genes, 1 in however many chance etc and it makes me cry my heart out. But most of the time, I just need to look at my beautiful, carefree, brave, courageous girl and think, yep, we will fight this syndrome together!

Lindsay Swales

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## **Friends of LMBBS**

Regular readers and those involved in the charitable sector will know that raising funds and applying for grants can be very unpredictable and a charity's successful steady growth can falter in leaner times.

At the LMBB Society, which relies solely on fundraising, donations and grants, income determines whether there will be a conference in a particular year or how many newsletters will be produced. Producing or re-writing leaflets can be particularly costly and again relies on the charity being in a strong financial position.

The financial stability of our small charity relies on *regular* donations and we are often asked why we don't charge for membership to the Society. This would indeed provide that much

needed regular income, however it has always been our policy that membership is entirely free.

Several years ago, we set up 'Friends' of the LMBBS, a fundraising initiative, giving those members who wished to regularly support the society, the means to do so and we have slowly built up a strong, loyal band of 'Friends'.

#### **You can become a 'Friend' of the LMBBS in the following ways:**

- You can take a collection box and collect for the Society, or organise a fundraising event
- Complete a regular payment form and make monthly donations to the Society from as little as £1 per month
- Or you can combine both of the above.

If you feel you would like to become a 'Friend', or you would like more information, please contact Kevin Sales, who will be pleased to assist you.

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#### **Beth Hoskins: Marathon Runner**



After years of volunteering and cheer-leading for Helen (May-Simera) at various events, I thought it was about time that I challenged myself to do something that I never thought I could do, run the US Air Force marathon. I had only ever run a couple of 5K races before, so I didn't have much experience of running or training for anything. However I found a 24 week training plan for beginners on-line, and an app for my iPhone which told me how far and where I had to go, and that helped a lot.

The training started off quite well. I particularly enjoyed the first day where all I had to do was rest, however running five days a week was a bit of a shock to the system and the last couple of long runs were really tough to get through. One unexpected bonus of all the training runs was getting to explore new bits of London that I'd never seen before; doing the National Lottery Anniversary Run around the Olympic park, which finished on the track in the Olympic Stadium was a particular highlight.

With lots of friends and family in the US, I had a very busy trip with stops in three different states before I even got to Ohio for the marathon. It was nice to see so many supportive faces on my travels though and it helped to take my mind off the main reason for being there. It wasn't until a couple of days before the race that I started to get really nervous. It was great to have Helen there to keep me calm and she was full of helpful tips like putting plastic plate covers on my shoes to stop them from getting wet from the damp grass. I was less convinced by her decision to wear a bin-bag to stay warm and dry before the start as my waterproof jacket seemed a much better choice!

The race itself was much more enjoyable than I expected; there were drink stations complete with cheering volunteers almost every mile, so the time, and miles, passed quite

quickly. Helen came to find me at mile 25 and ran the last bit with me; after a good few hours on my own, hearing her supportive screams really helped.

It still seems weird to think of myself as a marathon runner. One of the reasons for choosing this marathon was because the medal is so big and heavy - maybe I should start carrying it around with me more often to help it sink in!

Thanks to my friends and family who have supported me throughout my training and been very generous with their sponsorship, especially my co-workers in the lab. But most of all, thanks to Helen who, after many, many years of trying, has finally turned me into a runner.



In response to popular demand, LMBBS 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 done so already, become a 'LMBBS FaceBook Friend' and keep up to date with the Society and its supporters.

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## **This is Me**

Hi there, my name is Aneeba Ahmed, I am 22 years old.



Let me tell you about my story.

I was diagnosed at birth with a rare genetic condition which is called Laurence-Moon-Bardet-Biedl Syndrome; I was born with six fingers and toes, the extra ones have been removed on each hand and foot, and I have to wear special footwear.

I am visually impaired which means that I can't see well in my left eye, so you have to sit on my right and also walk on my right. I can't even see in the dark or see colours.

I use a white stick (which is called a cane). This helps me get around places, but I also need guiding, if I don't know a place very well. I also need a lot of light in a classroom and I wear special sunglasses because when I come back into a dark room from a light place, my eyes go white and also I go blurry.

I have always worn glasses since I was eight months old and I also have wobbly eyes, which means that I can drop things or break things by accident and also I can accidentally bump into you by mistake. I used to have childhood asthma, but I don't have it anymore, but I still do have eczema; I cannot wear silk clothing, I can only wear cotton.

I use a computer with a black on white keyboard with which I use special software like:



Zoomtext, Jaws, Narrorts and Lightning. I use a CCTV because I can't see small print and if I haven't got a CCTV, I can see large print; I use 36 Arial black bold. I use thick black lined paper and I use a pen with black ink because I can't use blue ink or normal line paper. But I can't write for very long because I get tired easily.

I can't hear very well in my left ear, which means you sometimes have to speak a bit louder if I can't hear you. I used to have grommets when I was 2 and 12 and I have got a t-tube (an invisible hearing aid) in my left ear and I sometimes have earaches.

I have got high blood pressure, which I am on medication for and I used to have a weight problem, so I can't have anything that can affect my blood pressure or my LMBBS and I do get very emotional very easily. I have speech problems, which means sometimes, it can be difficult for you to understand me and if I get upset or tense, I use my phone to communicate with you.

Finally, I have got Spondylosis which means that I can't straighten my back or my legs, if I am standing up or if I am lying down, also when I walk far distances, my legs get tired easily.

I am now studying at Wilson Stuart School where I am doing a 19 to 25 LIVE programme four days a week, three days on the site and one day on a work placement. I am studying Maths, English, Personal Social Development, ICT and Customer Services, plus I have Tutorial. I also have different options on a Friday which are Independent Living Skills, Gardening and Business Enterprise. I get Thursdays off and on Tuesdays I do a work placement. This is either at Priestley Smith School, where I do some Braille and help some students with their Art and Music, or I go to One Stop Shopping Centre where I am helping them with a Braille guidebook.

In my spare-time, when I am not at Wilson Stuart School, I like singing, listening to music, socialising with family and friends and I like to talk to other people on Facebook about LMBBS. I also like to do my Braille as well.

So you can see that LMBBS has not stopped me from achieving my goals in life and I would like to say:

'Don't let LMBBS stop *you* from achieving *your* goals and don't let it get you down'

Aneeba Ahmed



### **Brave, Crazy or Mad!**

Ten years ago, when I was last in Turkey on holiday with my family, my Grandfather wanted to have a go at parasailing, but chickened out at the last moment and I had been thinking about it ever since. I decided this time around that I would have a go.

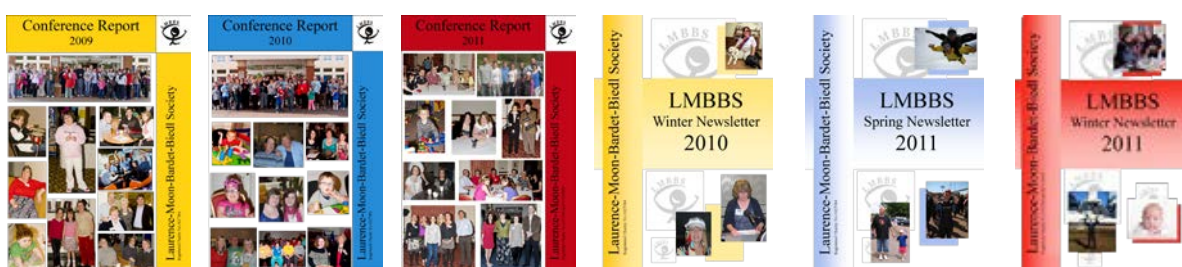
I was up there some time and reached the giddy height of 1,200 feet!! When I mentioned it to other people they said I must be Brave, Crazy, or Mad! Mind you I did wonder myself, when I felt like I was slipping. I looked down, saw all the blue sea and closed my eyes again.

I'm really pleased I did it though  
Tom Rawlings

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## LMBBS Leaflets, Newsletters and Merchandise

If you would like to order leaflets or newsletters, free of charge, please email: [toniahymers@btinternet.com](mailto:toniahymers@btinternet.com) or telephone Tonia on 07805 685342.



We also have a variety of merchandise available, a sample of which is shown below; email [kevin.julie1@btinternet.com](mailto:kevin.julie1@btinternet.com) or telephone Julie on 01892 685311 for availability and prices.



**LMBBS Clinics Admin Support**

Chris Humphreys  
01633 718415  
chris.humphreys4@ntlworld.com

**Newsletter Editor****LMBBS Clinic Support Worker**

Tonia Hymers  
07805 685342  
toniahymers@btinternet.com

**Secretary****LMBBS Clinic Support Worker**

Julie Sales  
01892 685311  
kevin.julie1@btinternet.com

**New Families Contact**

Claire Anstee  
After 4pm, Monday – Friday  
01604 880386  
claire@anstee2.orangehome.co.uk

**Adults/Young Adults Contact**

Steve Burge  
07833 228463  
steveburge@live.co.uk

**Special Educational Needs Contact**

Dianne Hand  
0161 442 0989.  
diannehand@live.co.uk

**Fundraising Co-ordinator**

Emma Oates  
01765 676578  
07861 770164

**Friends of the LMBBS**

Kevin Sales  
01892 685311  
kevin.julie1@btinternet.com

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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