

# LMBBS Spring Newsletter 2011

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## Contact Details

Julie Sales, LMBBS Secretary and Clinic Support Worker, can be contacted on 01892 685311 or via email at [Kevin.julie1@btinternet.com](mailto:Kevin.julie1@btinternet.com).

Tonia Hymers, LMBBS Newsletter Editor and Clinic Support Worker can be contacted on 01255 551886 or by email at [toniahymers@btinternet.com](mailto:toniahymers@btinternet.com).

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

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

## Get Well Soon Chris

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As some of you know, our National Co-ordinator and Conference Organiser, Christine Humphreys, has been extremely unwell since December last year. Chris has spent the past few months in hospital, managing to get home for just a short period, due to problems with her throat and breathing. She is proving to be a medical enigma and, at the time of going to press, the doctors are still puzzled as to the cause of Chris' health difficulties. We are keeping up to date with Chris' progress via regular calls to Chris' husband, Phil. In addition, whenever Chris has had access to her laptop, she has been straight back online to keep up to date with all things LMBBS. In her last email, she sounded amazingly upbeat despite facing considerable day to day challenges and thanked everyone for their love and support. On behalf of everyone in the Society, we send lots of love and good wishes for Chris' continued recovery.



## Rare Disease Day 2011

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Rare Disease Day

Tonia Hymers and Anne Crotty were invited to attend a reception at Westminster to mark Rare Disease Day 2011, together with other members of the Ciliopathy Alliance; it also provided an ideal opportunity to raise awareness of LMBBS.



The host of the Event was Mr Julian Huppert MP, who was passionate about the need to recognise Rare Diseases. Speakers were Liz Kendall, Shadow Health Minister, Professor Sir John Burn, Professor of Clinical Genetics at the Institute of Human Genetics, Mr Alastair Kent OBE, Chair of Rare Disease UK (RDUK) and Director of Genetic Alliance UK, Earl Howe, Parliamentary Under-Secretary of State for Quality at the Department of Health and Ms Jayne Hughes.

Jayne Hughes is a parent of Amy, who has Cockayne syndrome, and has founded the support group, 'Amy and Friends'. Cockayne Syndrome is a premature ageing disorder that leads to a shortened life expectancy. Ms Hughes experienced the same difficulties as many of our families, with delayed diagnosis and difficulty in finding appropriate help and support for her daughter. She did, eventually, gain the correct diagnosis and appropriate support for her daughter and some interesting research was taking place which could lead to a new treatment for those with Cockayne Syndrome. Unfortunately, this recently came to an abrupt end when the company involved with producing the drug folded, leaving the future development of this treatment uncertain. Ms Hughes, a remarkable speaker, finished her talk on a positive note, highlighting all the help and support that is now available for her daughter.

Professor Burn spoke about the importance of meeting families with rare syndromes and how research into rare conditions can have remarkable results. He described his involvement, many years ago, with a group of people with Turban Syndrome and subsequent research which highlighted a cause of cancer, something which, he said, was only possible because of the lack of 'red tape' in those days. In present times, such developments would never happen because of the difficulties of initiating research, due to the stranglehold of 'red tape'. Professor Burn asked those with influence, who were present, to cut it by half.



All the speakers supported the premise that the NHS should treat all people equally and everyone should receive the same standard of care, regardless of their situation or condition. Coming to the end of our first year since the start of BBS Multi-Disciplinary Clinics, we felt extremely fortunate to be one of very few rare disease groups to be getting such specialist care.

The House of Commons and Westminster were beautiful and we got a real sense of the history of our seat of government. All the staff were friendly and helpful, the refreshments that were provided

were delicious and Big Ben looked absolutely stunning in the dark! We will keep you informed of the date for next year's Rare Disease Day events, which are held in Wales, Northern Ireland, Scotland and England and, hopefully, we can ensure that LMBBS is well represented right across the UK.



For parents of a disabled child, many issues start and end with money; money for education, money for support at home and in school, money for special equipment, money for travel to school, hospital appointments and so on. For some, it can be impossible to work without adequate child care support. If you know your child would do better with support, or in a specific school, getting a statement of educational needs might become a major battle. If you rely on local support groups, you're at the mercy of council and charity budgets; if they get cut, so might the support group. If your child has complex needs, this becomes yet another challenge, unless you have a dedicated social worker or health visitor to help you track down what you're entitled to.

Given these challenges, perhaps it is hardly surprising that, according to national research, some 55% of families with disabled children, or children with additional needs, live on or near the poverty line (source: [http://www.thesafezone.co.uk/parents/special\\_needs.php](http://www.thesafezone.co.uk/parents/special_needs.php)). Apart from family finances, disability charities point to inadequate public service provision as another possible source of stress. Social services might have little awareness of a child's complex needs. Poor communication might exist between care professionals and the non verbal children they serve. Add to this, increased levels of social exclusion arising from bullying and harassment, the levels of stress in families with disabled children must be alarmingly high.

Small wonder then, that parents of children with disabilities have been identified as more at risk of depression and distress than other parents. Available support for families with a disabled child is generally felt to be most beneficial when it involves the whole family, rather than the child on their own. Key issues, such as the adequacy of social support, child behaviour or how well the family is functioning, are felt to be better supported when parent-led rather than professionally prescribed. In fact, family-centred care will, ideally, always ensure that ultimate control over decision making lies with the parents.

Family support workers provide a lifeline for many families with disabled children, yet many dedicated parents, committed to making the best life for their disabled child, end up ignoring their own needs and sadly, splitting up. Can more timely support help here? Marriage Care hope so. As part of a new initiative, they have recently secured funding to support 3,000 hours of counselling for parents of disabled children in the UK.

Marriage Care has been providing relationship support to couples facing challenge and change since 1946. The Big Society is not a new idea: all their trained counsellors are locally active volunteers. Over time, they have developed considerable insight into what builds and maintains adult relationships. People often feel overwhelmed by what they have to face, particularly where the outlook, as with many diagnoses of disability, is unlikely to improve much. They struggle to cope, especially if they feel under added pressure to soldier on regardless. Others feel isolated and simply

lack a voice. Counselling can't always breathe new life into a relationship but people generally leave, feeling more in control of their lives and happier to make the decisions that will ultimately help them and their families reach contentment and "move on".

For more information, go to [www.marriagecare.org.uk](http://www.marriagecare.org.uk).

Terry Prendergast, Chief Executive

## New Registry for Rare Diabetes

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EURO-WABB is a new project aiming to establish a registry for Alström, Wolfram and Bardet Biedl Syndromes and improve diagnostics, care, knowledge and treatment.

Within the EU Health Programme 2008-2013 and its call for promoting health through the creation of new registers for rare diseases, EURO-WABB is supported by the EU Directorate General for Health and Consumers (DG Sanco). The EURO-WABB project began on 1 January 2011 and will run for 3 years.



"The general objective of EURO-Rare Diabetes is to support efficient diagnosis, treatment and research for the rare genetic diseases Wolfram, Alström and Bardet Biedl (WABB) syndromes in Europe," says Professor Timothy Barrett (Birmingham University, UK) leader of the project. Currently, there are 13 participating members from 10 countries.

The idea for the EURO-Rare Diabetes project came from Nolwenn Jaffre, President of the French Wolfram Association, who gathered Wolfram syndrome researchers from around the world in Paris in October 2009. The final application included researchers in Alström syndrome, Bardet Biedl syndrome and Alström Syndrome-UK as partners. Alström Syndrome-UK is a charity dedicated to Alström patients and those caring for them.

EURO-Rare Diabetes is divided into six components called Work Packages (Coordination, Dissemination, Evaluation, Core Datasets and Pathways, Genetics, Virtual Registry and Information Environment) and Kay Parkinson of Alström Syndrome-UK is one of the partners in charge of Registry and Information Environment. "This is the largest European project in which our charity has ever been involved. Most of the time we are invisible and being involved in EURO-Rare Diabetes raises awareness of the charity and the syndrome. We are seeing benefits already, and, in 2011, the charity's role is to provide stakeholder analysis, and I would be pleased to hear from anyone with an interest. We will develop a dissemination plan and a report on the learning needs of health professionals," declares Kay Parkinson.

Rare Diabetes Syndromes are a group of rare, inherited disorders linked by intolerance of the body to glucose, and, though the register is mainly directed towards the three WABB syndromes, its scope includes some other rare diabetes syndromes. Long-term studies on these syndromes are desperately needed to understand their natural history, relate genetic diagnosis to predicting outcomes, establish a basis for evidence-based management, and to develop new treatments. "There are to date no orphan drug treatments available, nor access to well characterised cohorts of patients. The lack of specific health policies for these diseases and the scarcity of expertise, translate into delayed diagnosis and difficult access to care. There are almost no multidisciplinary teams of experts in these diseases. Genetic testing centres are concentrated in a few member

states and there is unequal patient access to testing across the EU, America and Japan,” states Professor Timothy Barrett.

The Registry will tackle all these problems by establishing the natural history of the three diseases; assessing clinical effectiveness of management and quality of care; providing a database of anonymous patient data for recruitment into treatment studies; establishing genotype-phenotype correlations. Professor Barrett and the other participants are keen to hear from health professionals, inside and outside of the EU, caring for patients with one of these syndromes, as well as affected patients and family members. “In practice this means we will use questionnaires and focus groups of health professionals, to find out what support they need to take part; then decide on the core dataset for the web based registry, to make it easy for health professionals to use. We will create a rare disease gene chip to identify all the known mutations quickly and make it available across Europe. We will also do questionnaires and focus groups for patients and health professionals to find out their learning and information needs; write education material and patient information on Alström and the other diseases; and finally use the registry to support ‘meet the expert’ platforms, and advice for families,” declares Professor Barrett.

Increasing knowledge of Alström, Bardet Biedl and Wolfram syndromes, supporting new research by allowing controlled access to investigators for epidemiological, clinical, genetic and interventional studies, advocating on behalf of families for improved quality of services both at national and European levels: the objectives of EURO-Rare Diabetes are ambitious and eagerly awaited by rare diabetes patients.

This article was first published in the March 2011 issue of the EURORDIS newsletter

Author: Nathacha Appanah

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## Fundraising Round-up

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Following a successful second half of 2010, this year has started exceedingly well, with grants of £1,000 being received from VICTA and £2,000 being received from the Hedley Foundation. We have also been promised £5,000 from the Foyle Foundation. These are all towards our Annual Weekend in April and we thank those concerned for ensuring that we will be able to have a great time this year.

This special weekend needs considerable funding, year on year, and we can't be certain of receiving the generous help that we have received over the last nine months. Your response to the 'Big "S" Summer Sundae' initiative was amazing. The total came to over £10,000 and you will read about two of the events, Emmy's Walk and Chris's Skydive, elsewhere in the newsletter. On top of this, there were all the other donations and fundraising efforts of our amazing band of supporters, including Gemma Allen who ran the Bupa Great South Run and raised around £300. Thank you, all of you.

So far, this year, Pauline Taylor's colleagues at HP held a Charity Raffle to mark Rare Disease Week in aid of LMBBS and have raised £700. The Kirby Group has donated £500 and the members of Christchurch have donated £200, following a Beatles night. We have received nearly £500 in memory of Mrs Joan Vagg who passed away in January. Our thoughts are with her family at this time.

This year's fundraising initiative will be based on 'Save Our Conference (SOC)', as the Annual Weekend is the biggest expenditure of the year and we can't always be as fortunate in our fundraising for it, as we have been this year. More details will be available soon.

If you are unable to organize your own fundraising event, perhaps you would be able to sponsor someone who is undertaking a challenge on behalf of LMBBS. Allan Clark is going to walk the West Highland Way from 7<sup>th</sup> to 11<sup>th</sup> of April, Helen May-Smera is training hard for the gruelling 'Iron Man' Triathlon on 29<sup>th</sup> October in America and Genny Martin is running the Sheffield Half-Marathon on 8<sup>th</sup> May. They all have a page on the Justgiving website ([www.justgiving.co.uk](http://www.justgiving.co.uk)), as has James Humphreys, who hopes to go paragliding over the Black Mountains in Wales, when family circumstances allow. Darron Jones and fellow MITIE employees are running the Cardiff Half-Marathon on 16<sup>th</sup> October.



Yellow Moon will be producing a Christmas Catalogue in the Autumn and we will get 20% Cashback on all items purchased in that catalogue.

If you wish to take part in the Weather Lottery, please contact me.

I am sorry that I can't name every single one of you for all that you have done and are doing for the Society. Every penny that we receive is greatly appreciated for, as you know, LMBBS relies almost entirely on your support. Thank you, all of you.

Anne Crotty



### **Disability Living Allowance - New rules for April 2011**

From 11<sup>th</sup> April, the law is changing to allow severely Visually Impaired people to apply for the higher rate mobility component of Disability Living Allowance (DLA). To qualify, you must be registered Severely Visually Impaired by a Consultant Ophthalmologist and have

- a best corrected visual acuity of less than 3/60, or
- a best corrected visual acuity of between 3/60 and 6/60, a complete loss of peripheral visual field and a central visual field of no more than 10 degrees in total.

### **How to claim Disability Living Allowance:**

If you think you may qualify, contact the Department for Work and Pensions (DWP) as soon as possible, as the date the application form is requested will be treated as the date from which Disability Living Allowance can be paid, as long as the form is returned within six weeks of that date. If the claim is delayed, benefit may be lost.

If you have an eye test coming up, make sure you receive your results in a letter, so this can be used as evidence for the claim. To find out more, see contact details below:

Benefit Enquiry Line: 0800 88 22 00

Web Page: [www.directgov.co.uk](http://www.directgov.co.uk)

If you experience difficulties claiming DLA, disagree with a decision or want further information, you can contact the RNIB Helpline for advice. Call 0303 123 9999 or email [helpline@rnib.org.uk](mailto:helpline@rnib.org.uk).

## **Direct payments**

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Direct payments are local council payments for people who have been assessed as needing help from social services and who would like to arrange and pay for their own care and support services, instead of receiving them directly from the local council. A person must be able to give their consent to receiving direct payments and be able to manage them, even if they need help to do this on a day-to-day basis.

### **If you already receive social services**

Your local council must offer you the option of direct payments in place of the services you currently receive. There are some limited circumstances where you are not given this choice. Your council will be able to tell you about these.

### **If you are not receiving social services**

To get direct payments, you need to contact your local council to ask them to assess your needs. Social services - and therefore direct payments - are normally available if you are:

- disabled and aged 16 or over
- a carer aged 16 or over, including people with parental responsibility for a disabled child
- an older person

### **If you have been refused social services**

If your local council has decided that you do not need social care services, it will not offer you direct payments. If you think your needs or circumstances have now changed, ask your council for a new assessment.

### **How much you can get**

The amount you receive will depend on the assessment your council makes of your needs.

### **How it is paid**

Direct payments are made directly into your bank, building society, Post Office or National Savings account. If you need someone who cares for you to collect your money, or you are registered blind, payment can be made by sending a cheque which can be cashed at the Post Office.

### **How to apply for direct payments**

If you already get services, ask your local council about direct payments.

If you are applying for services for the first time, your social worker should discuss the direct payments option with you when they assess your care needs.

### **What you can use direct payments for**

The money is for you to use to pay for the services and equipment which will meet the needs the local council has assessed you as having. As a general principle, councils should let you choose how best to meet your assessed needs, as long as they are satisfied that agreed support arrangements are being met.

### **What you cannot use direct payments for**

You cannot use direct payments to pay for permanent residential accommodation - but you may be able to use direct payments to pay for occasional short periods in residential accommodation, if your council agrees that is what you need. Unless your council decides that exceptional circumstances make it necessary, you cannot use direct payments to pay for a service from:

- your spouse (husband or wife)
- your civil partner
- a partner with whom you live as a couple
- a close relative with whom you live, or the spouse or partner of that close relative

### **Direct payments for disabled children**

Direct payments can be made to parents and carers aged 16 or over. This includes people with parental responsibility for a disabled child.

#### **If you have parental responsibility for a disabled child**

Following an assessment, direct payments can be used for a variety of services which offer your child stimulation, new experiences and independence. These include:

- short breaks
- nursery placement providing specialist support for your child
- assistance to attend an activity, for example a youth club
- personal care

#### **Employing somebody to provide support for your child**

Where agreed following an assessment, you can use direct payments to employ a person to provide your child with support. Local councils can provide guidance about safe practice in employing carer-workers to work with your child. You can ask your council to help carry out a number of checks, including a police check, to ensure the safety of your child. As the employer, the responsibility remains with you.

#### **Child care services and holiday time**

As a parent, or someone with parental responsibility, you should be supported to stay in work, or to return to work, if this is what you want to do. If you have been assessed as needing help from social

services, direct payments may help you make sure you have the right type of after-school or holiday provision to meet the particular needs of your child.

### **Specific queries**

If you have a specific enquiry about your own situation, your **local council** or **Social Worker** is the best place to start. Each council implements direct payments in its own way. They will also be able to put you in touch with local support services.

***We would very much like to hear from adults and families who are successfully accessing Direct Payments, how they are used and their experience of the process to help those who are thinking about accessing this system for the first time. Please contact Tonia if you feel you can help via [toniahymers@btinternet.com](mailto:toniahymers@btinternet.com). Confidentiality is assured where requested.***



Households where a member (adult or child) is blind or severely sight impaired can qualify for a 50% concession on their TV Licence. To claim this concession, you need a photocopy of one of the following:

- A complete certificate which has been signed by an ophthalmologist (a CVI or BD8 1990): or
- If you live in Great Britain, a certificate or any other document which has been issued by or on behalf of a local authority, showing that the applicant is registered as severely sight impaired or blind.
- If you live on the Isle of Man, a certificate or any other document which has been issued by or on behalf of the DHSS which shows that the applicant is registered as blind.
- If you live in Northern Ireland, a certificate or any other document that has been issued by a Health and Social Services Trust.

Just send your document, along with a letter, which includes your name, address and contact phone number, to the following address:

TV LICENSING  
BRISTOL  
BS98 1TL

**Please do not send original documents.**

Tel: 0300 790 6087

Fax: 0300 790 6012

Email: [enquiries@TVLicensing.co.uk](mailto:enquiries@TVLicensing.co.uk)

Web : <http://www.tvlicensing.co.uk>

All of the above information is for your *guidance* only. Full and up to date information on the above and other benefits is available on the government website,

[www.directgov.co.uk](http://www.directgov.co.uk)

Many thanks to Louise Martin for contribution to this article.

## Recycle 4 Charity

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You can raise money for Laurence-Moon-Bardet-Biedl Society by recycling your empty inkjet cartridges, mobile phones and laser or toner cartridges.

This service is 100% free and provides donations of up to £1 for all listed empty original inkjets, up to £30+ for mobile phones and up to £5 per laser/toner cartridge received. Please let friends, family and local businesses know about this recycling service. The more people we can get using this service, the more funds we will create for LMBBS. Check if your cartridges are compatible on [www.recycle4charity.co.uk](http://www.recycle4charity.co.uk). All freepost boxes and bags contain our charity's logo which will help promote our recycling efforts and can be obtained free of charge from [www.recycle4charity.co.uk](http://www.recycle4charity.co.uk). You should have received a collection bag with this newsletter; however, if you would like further information, please contact Anne Crotty on 01255 507977 or email [anne\\_and\\_terry@yahoo.co.uk](mailto:anne_and_terry@yahoo.co.uk).

## Skydive over Swansea

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As mentioned in the Autumn Newsletter, Chris Begley very bravely completed a sponsored sky dive to raise funds for the Society; he writes:

'Doing the skydive was an amazing experience, one of which I loved the thought of being able to do, so to actually get the chance to do it, while raising a very good sum of money for LMBBS, was more than a pleasure. My family were amazing in helping raise over £1,300 for LMBBS and were there on the day to witness the amazing event. It is an experience never to forget and one I shall hopefully do again! To be 13,500ft up, seeing the views of Swansea, it was breathtaking and a beautiful setting, all be it falling from the sky at 170mph!'

Well done Chris and huge thanks!



What an amazing first year we have had, supporting the new multi-disciplinary BBS clinics, and how fast it has flown by! So far, we have seen 49 children and 116 adults and are around two thirds of the way through our patient database. We estimate it will probably take around 15 months to get through the list initially, so those who haven't yet been given an appointment, should hear from us over the next few months. If you feel you need to be

seen urgently, please contact one of the LMBBS Support Workers and we will book you in as soon as is possible.

We have really enjoyed meeting so many new people, those who are living with the syndrome, their families and carers, and have seen first-hand what an impact the clinics are having. Parents in particular have been overwhelmed at the support and knowledge available to them, often following years of struggling on alone. For older children, the transition from paediatric to adult clinical services at BBS Clinics is a positive one, with continued high standards of care and understanding amongst all the medical and support team. For adults with the syndrome, it is a relief to be amongst healthcare professionals who already know about LMBBS and to be able to just talk about how it is affecting them personally. Lunchtimes have been a very enjoyable experience, giving everyone the chance to chat informally, either with each other or, in some clinics, with the clinicians as well.

We distribute questionnaires to everyone who attends a clinic and to date have received extremely positive feedback. We make a point of asking for good, honest, constructive criticism, to ensure the clinics remain patient focused, and any less than positive comments are acted on where possible. Of course, not everyone returns the forms as, once home, life moves on and these things tend to be forgotten. If you have a form at home, please, please take a few minutes to fill it in and return it to us – we don't need your name, just where the clinic was and the month it was in. From the following comments, you can see what a positive experience it has been for those attending:

“All the doctors, nurses and consultants were very patient and understanding and took lots of time answering our questions. We really feel that we could not have received better care.”

“The clinic provides reassurance that there is a support body ready to answer questions and more importantly to liaise with the local medical practice.”

“This is a wonderful service provided by the hospital and we are grateful for this clinic. A lot of hard work has obviously been done by the support workers to make all this possible and we thank you for this and their dedication.”

The role of the Support Workers has been diverse. Pre-clinic, we help with travel arrangements, organise hotel accommodation and liaise with the hospital on the patient's behalf. For patients staying at a hotel the night before the clinic, we meet them at the hotel, or station if possible and assist with checking in and dinner arrangements. On clinic days, we also assist patients to and from

the station where possible and arrange travel refunds where appropriate. We also make lots of tea and coffee and provide general support and information. On occasion, further appointments are generated, where the clinicians have identified issues that need investigating further and we are happy to help with arrangements for attending these also.

Our aim is to have two LMBB Society representatives at each clinic, to ensure adequate support is available; however, this is not always possible. We would, therefore, like to build up a 'bank' of willing volunteers, to assist the Support Workers, so, if you live reasonably close to London or Birmingham and feel you would enjoy this opportunity, please contact either Julie Sales or Tonia Hymers for more information. Although this is a voluntary role, all expenses will of course be paid.

We look forward to seeing you all soon.

Tonia Hymers email: [toniahymers@btinternet.com](mailto:toniahymers@btinternet.com) Tel: 01255 551886 or 07986 447429

Julie Sales email: [kevin.julie1@btinternet.com](mailto:kevin.julie1@btinternet.com) Tel: 01892 685311 or 07773 252585

## **Healthy Eating and Dietary Issues**

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### **Is a diet with a high dairy content the answer to weight loss?**

Calcium and milk intake have recently been in the press, reporting positive benefits to health. If you didn't read any of these articles, here is a quick summary. Some research has shown that people consuming the recommended servings of dairy foods were likely to be slimmer than those who did not. Another study showed that consumption of milk and dairy foods as part of a calorie controlled diet is associated with greater weight loss. Is this the magic answer to weight control for all of us?

Unfortunately, when you look at these studies in greater detail, there are concerns with how these conclusions have been reached. Sometimes studies have not been designed to look at this outcome. Also, there have been concerns that reported food intake may have been under or over reported, thus negating any effect calcium intake may have had. Other factors known to be associated with successful weight loss have also not been assessed or reported e.g. exercise and total energy intake.

Most of these studies recommend further work, exploring this possible relationship between dairy/calcium intake and successful weight loss/control. So where does this leave us now? Even if dairy intake does not prevent weight concerns, it is important to ensure we / our children are eating adequate amounts. Dairy foods are good sources of calcium and other micronutrients important for optimum bone health. I think it is also important to ensure sufficient dairy foods are included if you / your child are trying a weight loss plan - all too often milk and milk based foods are reduced on these types of plan.

## What different types of milk are available?

Most supermarkets stock at least 3 different types of milk which all have a differing fat and energy content. Whole milk (full-fat milk) is usually sold by supermarkets in bottles with blue labels; it contains 68 kcals per 100ml and 3.5% fat. Semi skimmed milk, which is the most popular type of milk used in the UK (usually labelled green), has less fat (1.7%) and so is less energy dense (50kcals per 100ml). Skimmed milk (usually with red labels) has had even more of the fat removed, to leave only 0.1-0.3% fat and so it has the lowest energy content, just 35 kcals per 100ml.

Have you noticed the new 1% fat milk in your supermarket? It is available in most supermarkets but labelled with a variety of coloured labels - I have spotted purple and orange so far. Have you thought about changing to this new milk? The following may help you decide to give it a go. The Food Standards Agency published research showing that people who regularly used semi-skimmed milk would be happy to switch to drinking 1% fat milk. Three-quarters of consumers involved in this trial liked the taste just as much as semi-skimmed milk. Interestingly, 85% of those who gave it to their children said they did not notice the switch when given the lower-fat milk. If you and all of your family are older than 5, why not try this simple change to 1% fat milk now!

## Are there any different recommendations for children?

The UK guidelines for children between 1 and 2 years of age are to drink full-fat milk as it contains more calories to help them grow. Between 2 and 5 years, children can have semi-skimmed milk if they're eating a varied, well-balanced diet and are growing well. The lower fat milks (1% milk and skimmed milk) are not recommended for children under 5 as they don't contain sufficient vitamin A.

However, in BBS clinic at Great Ormond Street Hospital, if children are eating a varied, well balanced diet, I would recommend using the lowest possible fat milk together with an appropriate vitamin supplement. Each child requires an individual assessment to ensure they receive the best option for them and their family. If you have a child under 5 and you would like to try using a lower fat milk, contact your clinic dietitian. They can help you make this switch by recommending the correct type of vitamin supplement and discussing the best milk option for your family.

## How much calcium do we require?

Dietary requirements are called reference nutrient intakes (RNIs). The amount shown below will provide sufficient calcium for 97% of children/adults in a population:

Age	RNI for Calcium	Example of how to meet
1 to 3 years	350mg	300ml skimmed or 1% milk on cereal and as drinks (approx. 375mg calcium) or 200ml skimmed milk and 80g yoghurt (approx. 360mg calcium)

Age	RNI for Calcium	Example of how to meet
4 to 6 years	450mg	200ml skimmed or 1% milk on cereal & in hot drinks, 80g pot of low fat yogurt & 1 small triangle of low fat cheese spread (approx.450mg calcium)
7 to 10 years	550mg	200ml skimmed or 1% milk on cereal & in hot drinks, 125g pot of low fat yogurt & 20g of low fat cheese (approx. 600 mg calcium)
11 to 18 years, male	1000mg	300ml skimmed or 1% milk on cereal & in hot drinks, 150g pot of low fat yogurt, 30g of low fat cheese & 2 slices brown bread (approx. 970mg calcium)
11 to 18 years, female	800mg	200ml skimmed or 1% milk on cereal & in hot drinks, 150g pot of low fat yogurt, 30g of low fat cheese & 2 slices brown bread (approx. 840mg calcium)
19 years and over	700mg	200ml skimmed or 1% milk on cereal & in hot drinks, 150g pot of low fat yogurt, 2 tablespoons cottage cheese & 2 slices brown bread (approx. 690mg calcium)

If you avoid cows' milk in your / your child's diet, it is best to always use a calcium enriched milk substitute. The following table gives details of the calcium content of commonly eaten foods.

Food	Calcium (mg)
<b>Milk &amp; Dairy Foods</b>	
Glass of skimmed milk (200ml)	250
Milk per cup of tea or coffee (35ml)	44
Milk on cereal (100ml)	125
Portion reduced fat Cheddar cheese (30g)	252
Pot of low fat fruit yogurt (150g)	210

<b>Food</b>	<b>Calcium (mg)</b>
1 tablespoon cottage cheese (40g)	51
Low fat custard (120g)	168
<b>Cereals</b>	
2 large slices of white or brown bread (72g)	130
2 large slices of wholemeal bread (72g)	76
<b>Fish</b>	
Sardines in tomato sauce (100g)	430
Salmon canned with bones (100g)	91
<b>Nuts &amp; Seeds</b>	
1 tablespoon sesame seeds (12g)	80
Plain peanuts (50g)	30
6 whole almonds (13g)	31
<b>Soya</b>	
Calcium enriched soya milk (200ml)	178
Tofu (60g)	306
<b>Vegetables &amp; Fruit</b>	
1 tablespoon cooked spinach (40g)	64
1 cooked broccoli spear (45g)	18
Small can of baked beans (150g)	80
1 medium orange (160mg)	75
1 tablespoon of canned red kidney beans (35g)	25

Source: Food Standards Agency (2002) McCance and Widdowson's The Composition of Food, Sixth summary edition. Cambridge: Royal Society of Chemistry.

If you have any concerns about your / your child's calcium intake, please get in touch with your clinic dietitian.

Sarah Flack, Principal Dietitian, 30th March 2011

Contact number: 0207 405 9200 extn 5941 (Thursday & Friday)

## Genny Runs More Than a Bath!

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Genny Martin, sister of Alex Jones (who has LMBBS), has decided that she should get fit for Spring. She will be taking part in the Sheffield Half Marathon on Sunday, May 8<sup>th</sup> in order to get fit, and also to raise money for LMBBS. Genny is a final year student at Nottingham University and hopes to gain an Honours Degree in Psychology. In her 'spare' time, she volunteers for a student charity and for St John ambulance. She also has a part time job, working in a school for the deaf. Genny has not done any serious running since school, but she has been practising, and has high hopes of completing the half marathon in a reasonable time. If you would like to encourage her, she would be over the moon to receive your sponsorship. You can sponsor her via the JustGiving website: [www.justgiving.com/genny-martin/](http://www.justgiving.com/genny-martin/).

Louise Martin



*Working together to fight diseases caused by cilium dysfunction*

## Successful Launch of the Ciliopathy Alliance

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As reported in the Spring Newsletter last year, LMBBS is one of four founder members of the newly formed Ciliopathy Alliance. Research has shown that Laurence-Moon-Bardet-Biedl Syndrome is a ciliopathy and, by uniting with other groups, the aim is to increase awareness and benefit from future research.

The Awareness Day and Launch of the Ciliopathy Alliance took place on 29<sup>th</sup> November, last year. A short presentation was given by each of the four founding support groups, who are Laurence-Moon-Bardet-Biedl Society, Alstrom Syndrome UK, Primary Ciliary Dyskinesia and Polycystic Kidney Disease, highlighting the devastating effects of these conditions. There were also talks from scientists and clinicians involved with the Alliance, NHS representatives (National Commissioning Group), and policy-making groups (Genetic Alliance UK). The proceedings were supported by grants through ASUK, Deafness Research UK and the Beacons for Public Engagement.

Feedback from the day was very good and a lot of interest was generated among scientists, medical professionals and those affected by ciliopathies. Meetings are now taking place to formalize the Alliance and begin scientific discussions and fundraising. A Charitable Company is being set up and

all four founding support groups will have a member on the board of Trustees/Directors. Drina and Michael Parker have kindly agreed to take on this role for LMBBS this year.

The aims of the Alliance are to:

- Share knowledge and understanding.
- Promote awareness of ciliopathies and the respective patient organisations.
- Encourage collaborative research and facilitate where possible.
- Apply for research funding from government, trusts and foundations.

For more details of the Alliance go to [www.ciliopathyalliance.org.uk](http://www.ciliopathyalliance.org.uk)

## Walk for Emmy

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I don't think I can really express how much going to the LMBBS Family Conference benefited us. We attended our first conference before we had even got our firm diagnosis of LMBBS for Emmy, who was then only 6 years old. Just on one Friday evening, we learnt so much and met such amazing people, and most important of all left that evening having that feeling of being understood.

After attending last year's conference, my husband and I were in the car driving home and both thinking we have to do something to help with the fundraising for the charity; we cannot attend next year without giving something back. We thought of a couple of ideas but decided it had to be simple, easy to organise and something the whole family could take part in. We live near our local reservoir which is a 7 ½ mile walk, so the decision was made on a sponsored walk - our 'LMBBS walk for Emmy'. I started contacting our family and friends to see who would consider being part of this fundraiser. The response was overwhelming, don't get me wrong we always knew we had fantastic family and friends but they really did show their support.

We made certificates to award all the walkers, we borrowed t-shirts from the Society, set up a 'Just Giving' page and I even went on the local radio! I cannot begin to tell you how nervous I was and anyone who knows me will know that this is really not my sort of thing. I did stumble over my

words at the beginning of the interview, but I did it! I got through it and hopefully raised some awareness of BBS. One of our walkers, when going round her workplace to get sponsorship, was told by a colleague that she didn't normally give much to this sort of thing but she'd heard me on the radio and was very touched by what she heard.

There were about 30 of us, adults and children, walking on the day and we were blessed with lovely weather (thankfully) and had a good day just walking together for a great cause. We set an initial target on our JustGiving page of £1,000 and our family, friends and work colleagues all really touched us with their generosity. On the day of the walk we soon began to realise that not only had we reached our target, but that with all the sponsorship on the forms, we had probably doubled it!

We invited everyone back to our house for a BBQ after the walk and to celebrate our fundraiser. We sold BBS wristbands to anyone who wanted them and had a 'Name the Teddy' competition too. I went round everyone and got an approximate figure from them on how much they had hoped to raise and we did an announcement, which we got Emmy to say to everyone, that all their efforts meant that we were going to raise approx £3,500 for the charity. We were all quite emotional at this point and it just goes to show how something as simple as a walk can raise a lot of money.

I hope this will encourage people to give it a go and who knows what can be achieved for the charity.

Claire Anstee

## What A Star!

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Our very lovely Helen May-Simera is still working extremely hard in the lab over in the States, in her words, 'converting everyone to study LMBBS'. Helen holds the Society and its members very close to her heart, since working as part of Professor Beales' team at the Institute of Child Health. Following her move to the States a couple of years ago, she has remained very close to the Society, continuing to raise funds and awareness whenever she can. Helen has decided to take on an incredible challenge to raise even more funds for the LMBBS, for which we are enormously grateful. Please, please show your appreciation and support for this amazing young woman by visiting her JustGiving page: [www.justgiving.co.uk/Helen May-Simera](http://www.justgiving.co.uk/Helen_May-Simera). Read on to hear about Helen's next big challenge:

*Dear LMBBS Gang*

As some of you might already know, I have decided to attempt an Iron Man to raise money for the Laurence-Moon-Bardet-Biedl Society. I wanted to do something different, something big and something crazy, so that lots of people will sponsor me and I can raise lots of cash for the society.

I did my first ever triathlon about five years ago and was instantly addicted to them. However, an Iron man is no ordinary triathlon. It starts with a 2.4 mile swim, followed by a 112 mile bike ride and finishes with a full 26 mile marathon. Whilst I have done a couple of half Iron men before (where everything is half the distance), this is going to be a real challenge for me; I love riding my bike and I am happy running long distances, however, the swim is my biggest challenge along with the fact that I need to get around the whole course in under 17 hours.

I have decided to do the Iron Man in Willmington, North Carolina, here in the US. It's at the end of the summer on October 29<sup>th</sup>, so I will have lots of time to train for it. I have a few races over the summer lined up to keep me motivated and on track with my training. The swimming is going to be the hardest as I HATE swimming, so any advice from any swimmers out there will be greatly appreciated; however, apparently, the swim part on this particular race is with the tide so it's somewhat easier.

I am enjoying keeping up with everyone on Facebook and can't wait to see you in April. I'll keep you all posted along the way with how my training is going.

With much love to you all.

Helen  
The Official LMBBS Triathlete!



In response to popular demand, LMBBS now has its own FaceBook page, which is proving to be an excellent way for the Society to keep in touch with its members, as well as proving to be a useful fundraising tool. 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 'LMBBSFaceBook Friend' and keep up to date with the Society and its supporters. For those of you not on FaceBook, we also have an online Forum on the LMBBS Website, so, come on guys, STAY IN TOUCH.

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**We hope you have enjoyed this newsletter, don't forget, all of the contact details can be found at the beginning of the CD. The views and opinions expressed in this newsletter are those of the authors of the articles. They do not necessarily express the views and policy of LMBBS. Whilst every effort is made to check the accuracy of information reproduced, readers are advised to check with the original source before acting on it.**