MPS

Winter 2012

Society for Mucopolysaccharide Diseases

Please **donate**
online www.mpssociety.co.uk,
phone 0845 389 9901,
text MPSS01 £2/£5/£10 to 70070
or post your donation
to our office, MPS House.

The MPS Society

Founded in 1982, the Society for Mucopolysaccharide Diseases (the MPS Society) is the only national charity specialising in MPS and Related Diseases in the UK, representing and supporting affected children and adults, their families, carers and professionals. The MPS Society:

Acts as a **support network** for those affected by MPS and Related Diseases

Promotes and supports **research** into MPS and Related Diseases

Brings about more **public awareness** of MPS and Related Diseases

MPS & Related Diseases

One baby born every eight days in the UK is diagnosed with an MPS or Related Disease.

These multi-organ storage diseases cause progressive physical disability and in many cases, severe degenerative mental deterioration resulting in death in childhood.

At present there is no cure for these devastating diseases, only treatment for the symptoms as they arise.

Where does your money go?

A donation of ***£2 per month*** could help us to offer so much more support in so many ways:

Access to clinical management and palliative care

MPS Regional Specialist clinics

Support with disability benefits

Paving a child's way in accessing education

Upholding rights in employment

Advising on home adaptations

Bereavement support

Front cover photo:
Debbie and Mark Burniston at the Sherwood Pines 10k for MPS

Society for Mucopolysaccharide Diseases

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Registered as a Company limited by guarantee
in England & Wales No. 7726882
Registered Charity No. 1143472
Charity registered in Scotland SCO41012

Magazine Deadlines

Spring	1 Mar 2013	Summer	1 Jun 2013
Autumn	1 Sep 2013	Winter	1 Dec 2013

To submit content email magazine@mpssociety.co.uk

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Welcome to the Winter 2012 edition of the MPS Magazine. Thank you to everyone who took part in our online MPS Magazine survey with Survey Monkey. Many of you had some very positive comments about the magazine. We have taken on board your feedback and have included many more of your stories. We know you like reading them so please do send them to us!

If you have any material to submit, please email magazine@mpssociety.co.uk or phone 0845 389 9901. We would love to hear from you! In the meantime, enjoy the magazine!

Once you have read this MPS Magazine, please pass it on to your family, friends and colleagues. Help us spread the word about MPS and related diseases and the work we do. www.mpssociety.co.uk

Chairman's New Year Message

In our 30th year, I have found the efforts of the MPS Society members totally inspiring. I can't thank enough the hundreds of you who wrote this autumn to the Prime Minister, Deputy Prime Minister and your MPs' supporting the MPS staff team in their tireless efforts to protect the highly Specialised Service for Lysosomal Storage Diseases. You need to know your letters made a real difference and have left those at the heart of Government in no doubt that we are not going to accept a dumbed down, remote LSD service that is disengaged from patient stakeholders without a fight.

I am also totally overwhelmed by the efforts of the families of those with a child or children with Sanfilippo who have decided to turn their poignant family stories into money to fund the Genistein Clinical Trial. We can't thank Justine Angelli and Tessa Pettman enough for offering the MPS III families a page on "Share a Gift" and give friends, family and the public a unique opportunity to raise 'rare money' for 'rare disease research'. Of course, people have to be signposted to the pages on "Share a Gift" in order to generate money. Additionally, the press activity by the families these past few months has also been inspirational. It is not easy to tell your story to the press but the results of the "Share a Gift" campaign speak for themselves. So many thanks to those families. Needless to say, the hoaxer who carried out a protracted dialogue with Bobby Gill's Mum and the MPS Society promising £500,000 for the Genistein Clinical Trial has not thrown the families and the MPS Society off course in raising the £600,000 required to enable the team at Manchester to start recruiting.

Leaving aside the Genistein fundraising, the economic climate continues to present the MPS Society with significant challenges and this coming year is likely to be no exception. I need to share with you that the MPS Society are genuinely concerned that we will struggle to raise the funds to maintain our core activities, especially the individual advocacy service.
I therefore use this message to invite you all to tell your friends, family, workplace and children's schools about Wicked Genes, now registered as the working fundraising name of the MPS Society and help us to beat the trend and raise millions of pounds for the advocacy service, support activities and research that translates into therapies.

Finally, as we go into a new year, the MPS Board of Trustees and Staff send our good wishes to each and every member and their families.

Sue Peach
Chair of Trustees

MPS New Year Appeal

Over the past months our Advocacy team has been out and about meeting our families and members at their homes and at clinic. Everyone has been so warm and friendly, telling us how important the vital support services that the MPS Society provides are.

It is nearly two years since the MPS Society's relationship with Jeans for Genes came to an end and we have had to find new ways of continuing to fund Support, Research and Awareness as we no longer benefit from the Jeans for Genes partnership and we receive no government funding.

Unfortunately, as we all know, times are hard and like many other charities we have found it increasingly difficult to secure income from trusts and through corporate donations. As you know we have launched Wicked Genes as one way to try and plug the gap in our income to try and engage with a wider audience beyond our supporter base and make the charity more accessible to continue to raise vital funds for MPS support, research and awareness.

We are working hard to secure that elusive trust income and to try and engage with corporate support, but here are 3 really simple things you can do to help continue the vital work of the MPS Society for the next 30 years

- Set up a **direct debit** by either filling out the magazine slip or going online
- **Donate by text** - text WCKD44 £2/£5 or £10 to 70070 to make a one off donation
- Make a one off **donation by card** either online at www.mpssociety.co.uk, over the phone on 0845 389 9901 or using the donation form in the covering letter which accompanied this magazine.

Any and all support is very gratefully received.

Message from the Chief Executive

Dear Reader

For 30 years the MPS Society's advocacy team has been dedicated to supporting individuals and families rights and ensuring fair access to support and services across the UK.

They may have stood up for your child's rights in relation to education ensuring that they were able to reach their full educational potential. They may have sourced grant funding to help provide essential equipment and they may have supported you in applying for essential benefits, even going to appeal or tribunal where needed.

For many, an advocacy officer may have been your first contact, when you received the devastating news that you or your child has an MPS or related disease. The advocacy team is here to support you practically and emotionally, today and tomorrow but we need your help.

This team consists of only five advocacy officers. These five people are on the end of the phone for every one of our 1,200 members across the UK whenever needed. They are there at clinics, they are at their desks, and they are travelling to see you at home from the top to the bottom of the UK, in fact somehow that small team of five are everywhere, available 365 days a year. They are utterly unique and wickedly wonderful.

The MPS Society receives no government funding and relies totally on donations. These donations come in the form of grants and trusts but most importantly its supporter fundraising. Anyone can be a supporter of the Society and are invaluable to help raise much needed funds. Supporter fundraising comes in many different forms, from a cake sale to a masquerade ball, from a pub quiz to a trek across Vietnam. Some will fundraise through their place of employment, applying for charity of the year, or organising a dress down day.

We are a small charity supporting children and adults with very rare genetic diseases. We have to be able to continue to fund support, research and awareness and we desperately need you, your families, your colleagues and friends help!

At present there are 160,000 charities in the UK, it is a pretty crowded place to be and at the moment we are struggling to get ourselves heard.

Every little bit helps and although we appreciate that times are hard for everyone we desperately need more supporters to help raise valuable income for the Society. So how could you do this?

- Tell as many people about the Society, what we do and how we help
- Set up a regular donation. You can do so through your salary by asking your employer about PAYE (Pay as you earn) a much more tax effective way of donating to your charity.
- Can you, family, friends, your place of work organise a fundraising event?
- Can you get your school involved?
- Have you ever thought of jumping out of a plane, running a marathon or climbing Kilimanjaro?

If you think that you or someone you know can support the Society however small, please get in touch with our Wicked Fundraising team by email at wickedgenes@mpssociety.co.uk or by phone on 0845 389 9901.

Thank you

Christine Lavery
Chief Executive

Wicked Genes

www.wickedgenes.co.uk

News from the **Management Committee**

The Society's Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Trustee Board Meeting held in September 2012.

Treasurer's Report

The Treasurer presented the income and expenditure report for the period 1 January – 31 August 2012. It was noted that the MPS Society was encountering a reduced level of fundraising and to manage risk the Trustees created a Finance Oversight Committee to review income and expenditure.

Risk Management

Trustees reviewed the Society's risk register and no changes were agreed.

Personnel

The Trustees re-affirmed their decision that the vacant Advocacy Officer post continue to be frozen until restricted funding for this post is secured.

Clinical Management

The Chief Executive appraised the Trustees of the current status and responses to the MPS Society and other LSD patient organisation's initiative to raise concerns relating to the Department of Health's plan under the new NHS reforms to disband AGNSS. Trustees were advised that the Prime Minister, Deputy Prime Minister and MPs had to date each received over 300 letters on this matter. It was agreed that the MPS Society send out an AGNSS e-news to all our members updating them on our activities around AGNSS. The Chief Executive reminded Trustees of the background to the Peninsula University Health Technology Assessment Study for MPS I, MPS II and Fabry disease and the basis of the involvement of the MPS Society. Incomplete patient summaries marked confidential were provided to the MPS Society in May 2012 but it had only just become evident that an updated version still marked confidential was sent to some of our MPS I, II and Fabry members who participated but not all. Trustee, Tim Summerton, agreed to carry out an analysis of the data for the three patient summaries as the results appear flawed and the study lacked rigour due to small numbers of participants. It was agreed the MPS Society along with the other LSD patient organisations call the study organisers to account.

Advocacy Support

Trustees noted that with all the changes to benefits the Advocacy team are experiencing a high volume of requests for help from member families and individuals.

Senior Management and CEO Succession Planning

The Trustees who participated in the Senior Management and CEO Succession Planning meeting fed back on the most constructive discussions to ensure the future direction of the MPS Society.

Fundraising

Trustees were informed that the Wicked Genes school packs have gone out to all MPS families with school age children. The Trustees confirmed that Wicked Genes is the fundraising initiative of the MPS Society. The Trustees agreed that Wicked Genes is a less complicated way of fundraising for MPS but agreed that the brand image needs some investment.

MPS Society continues to support those affected by Metachromatic Leukodystrophy

The Society for Mucopolysaccharide Diseases continues to welcome members with Metachromatic Leukodystrophy (MLD).

We have an Advocacy Support Officer who supports individuals and their families affected by MLD.

We have a Guide to Understanding MLD fact sheet which is available from the MPS office, and downloadable from the MPS website, plus a range of other information resources which cover issues related to those affected and their carers.

For further information please phone **0845 389 9901** or email **advocacy@mpssociety.co.uk**

Reduce paper consumption!

To cut down on postage and paper so we can put more of our resources into helping our members, we would like to become as paperless as possible.

If you would like to be informed by email of our events and activities, please email mps@mpssociety.co.uk with your current email address.

We will add this to our database to keep you informed and will not pass your details on to a third party.

We understand that this may not be possible or convenient for everyone and if, at any time, you feel you would rather receive information by post or be removed entirely from our mailing list you can email the above address or call **0845 389 9901**.

News from the MPS office

We have a new face to introduce you to. Many of you will have already spoken to Laura Burrows when you ring the MPS office but it's nice to put a face to a name!

Introducing **Laura Burrows**

Hi, my name is Laura Burrows, and I have recently joined the MPS Society as a Development Officer for Wicked Genes. Somewhat confusing as there are now two Wicked Development Officers called Laura!

My previous work experiences includes working in publishing, education and most important for my new role, in not-for-profit marketing and fundraising. I am very excited about developing the Wicked Genes brand and getting any and everyone on board raising money and awareness for the amazing work that the MPS Society does.

I am particularly hoping to engage with companies and businesses to promote the charity. (If you work for a company that might be interested in supporting us then give me a call on the Wicked phone 0845 389 9901)

When I am not working you can normally find me with my husband and children walking the dog somewhere in the Chilterns. I have a rather worrying fondness for all things chocolate, Downtown Abbey and (somewhat bizarrely) World War History!

I would like to thank everyone for making me feel so welcome. l.burrows@mpssociety.co.uk

Get Wicked at Work!

One of the BIGGEST ways we think you might be able to help is to get talking to your work colleagues. Get your brothers, sisters, mums, dads, friends, colleagues; team mates all talking in their businesses, companies, factories, tea shops, cake shops. Corporate donations to UK charities are estimated at around £1.6 billion annually. (NCVO, 2012). Our biggest problem is that unless you know of someone affected by an MPS or related disease you are unlikely to have heard of the Society for Mucopolysaccharide Diseases and so we aren't likely to get nominated for Charity of the Year relationships, Cause Related Marketing Partnerships or any kind of business support.

We need you to help us get a foot in the door. Go and hang round the desk of your CSR person (Corporate Social Responsibility) or HR person, leave our information with the right people. Find out if your organisation holds Charity of the Year elections, please nominate us. Perhaps you work in a small or medium sized business and there hasn't been a formal charity plan before – Now is your chance, seize the opportunity!. Any support great or small (Cake sales in the office to yearlong charity partnerships) we need it all.
In fact some companies will even match fund any money their employees raise, don't know? Go and ask…please!

Charitable activity, can improve brand and marketing efforts, and even increase sales and market share through association with a charitable causes. This competitive advantage has been proven in a number of research studies (BitC, 2008; Lev et al, 2008; Klein, 2011). Perhaps most importantly, a good CSR strategy can have positive benefits on staff morale, skills and even relationships with suppliers. Get your colleagues to get Wicked at work, they'll have fun, feel good and raise much needed funds.

We have some big fundraising targets and little resources to get there and we may be small, but we mean business! If you want to find out how to get Wicked at work, rest or play give us a bell on the Wicked line or send us a Wicked email. We are in this together.

0845 389 9901

www.wickedgenes.co.uk

wickedgenes@mpssociety.co.uk

Wicked Genes

WHAT'S ON!

CONFERENCES and REGIONAL EVENTS

Scottish Family Get Together: 3 March 2013

MPS National Weekend Conference: 28 -30 June 2013, Hilton Coventry

MPS REGIONAL CLINICS 2013

MPS IV clinic GOSH: 9 July

Manchester BMT clinic (under 6 years): 12 April, 12 July, 11 October

Manchester BMT clinic (over 6 years): 3 May, 5 July, 18 October

BMT clinic GOSH: 26 March, 24 September,

Fabry clinic GOSH: 9 April, 27 August

MPS III clinic GOSH: 26 February, 14 May, 13 August, 12 November

Birmingham MPS and Fabry clinic: 8 February, 14 June, 8 November

Adult Fabry Clinic Birmingham: 19 March, 11 June, 13 August

Bristol MPS Clinic: 27 March

MPS Awareness Day

15 May 2013

We know, we know, May is ages away but we are already getting our thinking caps on about a Wicked Wednesday in May. We want to make this year's MPS Awareness Day the biggest and best yet.

MPS Awareness day on 15th May is an international day to raise awareness of MPS and related Diseases and we want to get lots of activity going on across the country to fund the MPS Society's Support, Research and Awareness.

Whether you have a Wicked Wednesday at work and get everyone to pay a £1 and come in wearing blue in support of the MPS Society or you have a Wicked Weekend of sponsored Walks around some of Britain's nicest spots we want to hear from you. Can we help you coordinate an event? How can we support your fundraising?

We are putting our heads together here about what we can do to help, so keep checking our website for ideas and suggestions!

Visit **www.mpssociety.co.uk** for more information
or give us a call on **0845 389 9901** to find out how you can support us...

Congratulations!

On 16 December amid the Christmas decorations, Lucy Lavery and Paul McKay were married at Rudding Park, Harrogate, Yorkshire. Many of you will remember Lucy and Paul as regular volunteers for the MPS Society. Lucy, whose oldest brother died from Hunter disease 5 years before her birth, attended her first MPS conference in September 1987 as a baby. Lucy is now in her fourth year at the London and Barts medical school. Paul having graduated is now working at Proctor and Gamble.

Congratulations to Lucy and Paul and we look forward to seeing them in June at the MPS Conference as Mr & Mrs McKay!

Book Now!

MPS Society Conference Weekend

28 - 30 June 2013
Hilton Coventry

A programme and booking form is enclosed in this MPS Magazine. The full programme and booking form is also available from the Latest News section of our website **www.mpssociety.co.uk**

New members

Louise has recently been in contact with the Society. Her son Merlin has a diagnosis of Hunter disease. Merlin is four years old. The family live in South Wales.

Roger found out he had Fabry disease in 2009. He and his wife live in South Manchester. He leads a full and active life while receiving Enzyme Replacement Therapy (ERT).

Jemma and John have recently been in contact with the Society. Their daughter Grace has recently been diagnosed with Maroteaux Lamy disease. She is one year old. The family live in the North West.

Shaista has recently been in contact with the Society. She has a diagnosis of Fabry disease. The family live in West Yorkshire.

Births

Corey Jeffery has MPS I and is post transplant (2009).

After two previous affected pregnancies, we are delighted to welcome baby Joshua to our family. He arrived on 24th September 2012.

Corey is being a wonderful big brother and enjoys singing to Joshua and holding his hand!

Corey has also just started mainstream school, and has adapted extremely well to all the changes going on.
Teresa, Shane, Corey and Joshua Jeffery

Our trip to Orlando

We are the Harris family. I and all my children (aged 8, 25 and 26) have Fabry disease. We have been saving for a few years to go to Disneyworld Florida.

Early one October morning we woke Jack our 8 year old son up. He came downstairs bleary eyed thinking he was about to get ready to go to school. I handed him an envelope which he curiously opened. It was a Mickey Mouse card and it said, 'Jack shall we not go to school today? Shall we go to Disneyland instead?' He sat on the sofa and read it, then re-read it, then looked at us and said "What! Today! Now?" It was an amazing memory. He jumped up and down and hugged us and ran around like a mad thing for a bit!

We had packed already in secret, pills, potions, doctors' letters, tickets, passports. We headed off to the airport. I had previously spoken to the airline and explained that we had an existing illness and checked it was ok for us to have all our medication with us. They told me that as long as they were prescription medication that was fine and I would be able to board the plane with anyone who needed special assistance if I wanted too. We took advantage of this and it made the boarding process much easier. After a long 9 hour flight we arrived in Orlando. We settled in on that first day and unpacked.

The next day we headed off to Disney World. When we arrived we went straight to guest services with a doctor's letter explaining about our Fabry disease and they gave us special passes that allowed us to fast track all the queues. We did still have to queue a little but this made a huge difference to our visit. It meant no standing around in the heat in huge queues. If you take a doctors letter to all the parks like Disney and Universal Studios then you will be given a special guest services card to carry. This is so helpful if you have an illness that restricts you in any way. There is plenty of disabled parking, just take your UK disabled badge with you and show it to the person who takes the parking fee from you. Your disabled badge won't work anywhere outside of the parks unless you register it locally, but it's fine in the parks for disabled parking.

There is a massive amount of walking on this type of holiday. There were times I needed a wheelchair for Jack (as he is too big for the buggy hire). You can hire wheelchairs and buggies for the day in all the parks. I myself have Fabry and had to take a couple of days off during our trip as it is so tiring. When you have any long term illness it should never stop you from doing anything, you just need to pace yourselves, plan ahead and work round any issues you need to. We just needed to stop, rest lots and drink plenty of fluids! The water from the fountains in the parks tastes foul! Take bottled water with you, although there are many places to eat and drink).

We had a fantastic two weeks in Florida. October is a great time to go as it's not too hot but hot enough for flip flops and t-shirts. Another bonus to going in October is they make a huge deal of Halloween. There is lots to do for children in the parks, such as trick or treating, the children pick up goodie bags and then go to the trick or treat stops and are given treats. There is so much to see, and for the adults, the Universal park is closed off in the evening and they get it ready for adult Halloween! This is awesome! They have various horror mazes and scare zones within the park and zombies and all manner of gouls and ghosties hiding around every corner to jump out at you. I love a good scare so enjoyed it hugely! But, this is not for children though so we left our children at the villa with an adult.

All in all our holiday was amazing. In the parks you are given photo pass cards, get as many photos as you can stand being taken on that card and then you can pay a one off price (you can split the price of it if you have more than one family within your group). We are a family that believes memories are massively important, even if you have to save for years or have smaller trips or holidays. Photos capture those moments forever, so whatever life throws at you those moments are captured for you all to remember, so when you have the not so good bits, hospitals, medication, operations, pain, illness in general, those pictures and memories will always lift you up. We all wade through the bad bits knowing that the good bits are that much sweeter. **The Harris Family**

Sarah's Update

Sarah has Fabry. She writes a regular column for us about her experiences...

Hi again! So what's happened in the last few months?

It's been fairly busy actually. I'm settling in to my new job well. I love every minute of it although it's such a new experience. I am still finding doing 9-5 six days a week difficult but it's slowly getting easier. I am always grateful when it comes around to infusion time and I get a little boost, but all in all it's going really well.

I've got a few months left of my Open University degree for this year and I'm finding that extremely hard, when I have time off from work I have to do that and it's not easy. I think I might take a study break from it for a while to give myself a break. Now I've got a better job I don't need to worry so much and I can always come back to it later. We will see how it goes over the Christmas period and see where I end up.

The cold weather is arriving with a vengeance, I don't mind so much as it's the hot that gets to me but it does mean the likely onset of some kind of virus that I will find hard to shake so these next few months will be testing with work.

I hope you all had a healthy and happy Christmas and a brilliant start to the New Year. **Sarah**

A Happy Story

The beautiful dress has been bought and the venue is booked. Next year on 7th September 2013 at 2pm my beautiful daughter Samantha Brockie is getting married to Ashley Downey. Sam is 21 years old now and suffers with MPS I Hurler Scheie.

Sam has achieved so much and makes me proud with her fighting attitude (some who know me might say she is just like her mother). Sam moved into her flat in June 2011 and is happy being a domestic goddess and planning her wedding.

Sam was an original trial patient for Enzyme Replacement Therapy and we spent a lot of time travelling to Manchester from our home in Alcester Warwickshire for a lot of years, then to Birmingham Children's Hospital and thankfully now she has infusions at home with her dedicated friendly nursing team of Sandra, Kate and Jenny. Sandra has become Sam's main nurse and we have a great time putting the world to rights weekly whilst eating doughnuts!

With all the stress and tears shed with Sam's illness, obstacles that have to be passed and health issues we have had to concur there are so many positives in Sam's life too even on the worst days. **Vicki Brockie**

Get in touch...

If you would like to share your story and photos in the MPS Magazine please write to us, phone **0845 389 9901** or email **magazine@mpssociety.co.uk**

My Story

Susan Wilkinson shares her story of being diagnosied with Fabry and the impact it had on her life and that of her family...

I was seven years old, screaming, pains in my legs and hands, boiling hot feet every day. Nothing took the pains away; I could be in pain for minutes, hours and often days. I picked up every virus going around and needed so much sleep. My parents took me to see our local GP frequently, I had numerous out of hour's home visits to be told I had growing pains and I would grow out of them. I eventually went to see a child specialist who told my mum that he thought it was all in my head, blood tests were fine!

School was a nightmare, especially summer and PE lessons. I used to try and get out of every lesson, skiving if I had to, anything physical was torture and left me fatigued for days and in so much pain. I had no idea why. Then I would remember, it's all in my head! I pulled away from a lot of my school friends as they were all sporty, it was easier to do that than keep making excuses not to join them.

At the age of 15 the pains became unbearable, my legs felt like lead, I could hardly walk and my fingers felt like they were having electric shocks. Fainting, fatigue, other unexplained sickness and diarrhoea. My temperature was off the scale, I didn't sweat, no matter what I did it or tried, it continued. The doctors still said there was nothing wrong with me.

As I got older, people said I was lazy, I had no energy and never wanted to join in with a lot of activities, it was easier to stay at home and do what I knew I could manage. I was even told I was not a typical normal teenager.

I secretly lived off painkillers, drank cool drinks constantly and I also had acupuncture, it did not work.

Life continued and I learned to live with it. It was just how I was and I had to get on with it, thank God for buckets of ice cold water and co-codamol and my bedroom.

When I was 18 I developed a strange rash on both palms of my hands and also around my waist and upper body, even my belly button. I was told by a local GP "It's not cancer; get on with your life". So I did, well to the best I could. Then at 31 I developed high blood pressure, "it's hereditary in your family Susan", said the GP putting me on medication, no tests to get to the cause.

I met David in October 2002; David had two children who lived with him. When I was 36 years old David and I married and in 2008 I gave birth, 6 weeks early (due to pre-eclampsia), to our gorgeous son Adam.

During my pre natal checkups, of which I had many, my consultant suggested I had my kidneys checked out as she suspected, due to my high protein levels, that there could be a problem. I was sent to the Freeman Hospital, Newcastle upon Tyne. The Nephrologist suggested I had a biopsy but should wait till by body recovered fully from child birth.

I was back at work, working as a children's learning Mentor, David was running his own joinery business and Adam was in private nursery. Life was good.

In February 2009 my biopsy was done. I had an appointment to see the consultant soon after to discuss the findings. The day before my appointment I got a call from him asking me to take David with me to the appointment; obviously my first question in a panic state was WHY! His words stick in my head till this day: "Mrs Wilkinson, it's nothing to worry about but we have the results of your biopsy and found that you have a very rare genetic metabolic disorder. We need David with you as it's genetic and may involve your son too. The good news is that there is medication for it. Don't worry too much and we will have a good talk tomorrow". Immediately I googled rare Genetic Metabolic Disorders, Andersons Fabry's Disease flashed up. I read what follows....

What are the symptoms of Fabry disease?

Some women who carry the genetic mutation may have symptoms of the disease. Symptoms usually begin during childhood or adolescence and include: Burning sensations in the hands and feet that get worse with exercise and hot weather, small raised reddish-purple blemishes on the skin, fatigue, decreased sweating, fever, gastrointestinal difficulties, particularly after eating.

What is the prognosis for Fabry disease?

Patients with Fabry disease often survive into adulthood but are at increased risk of strokes, heart attack and heart disease, and kidney failure.

My first thought was "I'm going to die!" My life fell apart; this was me, my life on one page. I wasn't a hypochondriac, I wasn't lazy. I was normal. Normal, for a person with Fabry's.

The next day it was confirmed, I did have Fabry's, and my kidneys were slightly damaged. I was stunned but at the same time relieved that all the suffering I had been through in my life was real, not in my head. I cannot remember what was said in that meeting, a total blank apart from the word Fabry's. Luckily David did.

After many tears, sleepless nights and worries, life had to continue. I attended many appointments, went to see countless consultants and had numerous tests. My family had to be tested too; it had to be passed down from somewhere! Adam and my sister, Brenda, were tested first; waiting for the results was the longest 7 weeks and the worst time in my life. Deep down I knew Brenda didn't have it, she was energetic, walked for miles, and she was never poorly or in pain and loved hot summer days.

Adam, however, was a different story. Just to watch him made me feel physically sick, I couldn't sleep, I looked for signs (stupid I know at such a young age), cried every time I thought of my beautiful, perfect baby possibly having this awful disease. The guilt was inconceivable. Every day I waited for the phone to ring, every time it did my heart was in my mouth, my body shaking, was it Dr Brenan with the results. I knew that day had to come and it did, Adam and Brenda were fine, they did not have the defected gene. Thank god.

Within weeks, my Dad, Mam and Mam's side of the family were tested. Not one of them carried the Gene. I was apparently a spontaneous mutation, one in a million (faulty sperm). I was so thankful that no one had it but on the other hand wondered why me. I was, at the time, the only woman diagnosed because of pregnancy and the only woman to have no family link in the UK.

Medication for pains, gastrointestinal disorders, high blood pressure, kidneys, cholesterol, and depression/stress were prescribed and then Enzyme replacement therapy.

I gave up the job I loved and focussed on me. It's been three and a half years since diagnosis and two and a half years since starting ERT. I have good days, bad days and unbearable days, but I'm alive, living the life my body allows me to live.

Thank God for my Mam and Dad, who always believed me, David, my family and friends. The love, help and support they give me is second to none. The help, guidance and support from the MPS Society, especially Advocacy Support Officer Rebecca. My new GP, Consultants, nurses at Manchester, Healthcare at Home, without them I don't know how I would cope. Thanks too for medication and the scientists who are researching this awful disease every day.

Last, but by no way least, Adam, my little miracle baby boy. He makes me so happy, such an energetic, loveable and naughty (ha ha) little boy who I owe my life too, my life saver, my world.

I have learnt many things since my diagnosis and so have the people who didn't believe me. If there was one thing I would advise anyone, it would be... Never ignore your medical instincts and never let people tell you "It's all in your head".
Susan Wilkinson

Get in touch...
If you would like to share your story and photos in the MPS Magazine please write to us, phone **0845 389 9901** or email **magazine@mpssociety.co.uk**

Living with Morquio

Judith Evans is a Trustee of the MPS Society. She has a daughter, Joanne, who suffers from Morquio. Judith gives an account of their family's experience from diagnosis through to Joanne's ambitions and hopes for the future...

I have lived in Scotland with my husband Graham for the past 32 years and our daughter Joanne was born there in 1986; initially there was nothing to suggest that Joanne was anything other than a happy, healthy baby but when she was about 3 years old her growth started to slow down, although intellectually she was very bright. No parent wants to even contemplate that their child might not be perfect, but I had a niggling feeling that there was something not quite right; I was dismissed as a neurotic older Mother by the health visitor, our GP and even the paediatrician at our local hospital but finally a radiologist at the hospital made the diagnosis.

Like all parents receiving such a devastating diagnosis, we were left reeling and it took us some time to come to terms with being "new kids on the disability block" and the future we had envisaged for our daughter certainly changed dramatically.
We were given the contact details for the MPS Society but I couldn't bring myself to phone them which, with hindsight, was very silly. However, I was in complete denial and making that call would have meant admitting to myself that my only child had a life limiting condition and I wasn't ready for that… but I did get there in the end and, of course, the MPS Society has been a very important part of our lives.

Quite apart from the shock of getting the diagnosis, our next shock was to discover that there was no treatment and I think it was still a few years before ERT became even a pipe dream. Although, of course, the MPS Society was already funding the research that would eventually lead to treatments for the MPS diseases. However, the lack of treatment meant that Joanne was able to enjoy a relatively carefree and, that terrible word "normal" childhood with minimal medical intervention; and also that I didn't have to make the decision as to whether she should have to be involved in drug trials which have a considerable impact on the lives of the patients and their families.

So, we struggled through those early days until Joanne was referred to the wonderful Dr.Ed Wraith in Manchester. Ed helped us to see that Joanne's condition wasn't the end of the world, and we still have the diagrams drawn on scraps of paper which he used to explain the genetics and with which all his patients are familiar! It was also Ed who counselled us to contact the MPS Society which, of course, we did. Those two events were the turning points in our lives and I can honestly say, we've never looked back, although there have been many ups and downs along the way.

One of our first real battles was to get Joanne into our local mainstream school; thankfully things are very different now and it would be unthinkable that a child with Morquio would go anywhere else, but, way back then, our local education department were adamant that Joanne should go to a special school whilst we were equally adamant that she wouldn't. Suffice to say that Joanne went to the local primary school, then on to high school and eventually to Glasgow University from whence she graduated with a joint honours degree in 2009.

In her last months at university she began the frustrating search for a job - she applied for over 100 positions and attended interviews the length and breadth of the country and, whilst, she got superb feedback, there were no job offers. However, a couple of months after graduating, she began work for Cancer Research UK just on the other side of the business park here (and I think that whilst the job was a very menial admin one, which she found mind-numbing, she will always been grateful that they saw past the wheelchair and gave her the work experience she craved). She still lives with her boyfriend Phil, just along the road, although she now works as a Business Development Assistant for the Said Business School; for the past three years she has been studying in her spare time for a professional qualification with the Chartered Institute of Marketing and she is also an enthusiastic member of the Oxford Toastmasters Society. Her main love in life is tall ship sailing with the Jubilee Sailing Trust and it was when she took part in the Tall Ships Race in America seven years ago that she met her boyfriend Phil and they've been together ever since.

Joanne is a fiercely independent and ambitious young woman who copes with the many frustrations life throws at her with an admirable pragmatism. However, her desire for independence does sometimes stop her from asking for or accepting the help she needs, whereas an able-bodied person would have no such qualms. From being a relatively young child, Joanne has always been extremely single-minded and we, as her parents, were rarely consulted as she decided which particular path she would pursue; I partly blame Christine for this… when we were still very new members of the Society, Christine phoned me one day - Joanne was about 8 years old at the time - to ask if Joanne would like to go on a trip to Disneyworld with a group of other disabled children; my reaction was that she couldn't possibly go without me, but Christine gently persuaded me otherwise, and Joanne duly flew out to Florida for two and a half weeks and had the time of her life and an early taste of independence. It is a family joke that we decided, early on, that we would bring Joanne up to be as independent as possible and we often muse that we succeeded rather too well!

The past few years have been a very exciting time for all the Morquio families, with the advent of the drug trials for enzyme replacement therapy. Joanne took part in the trial for six months last year and had previously contributed to the natural history trial. She did this purely to help the research along but without any real thought of it benefiting from it herself.

She felt unable to continue on the ERT trial because she found the travelling to London every week tiring, and couldn't reconcile the time involved with pursuing her career. This was not a decision she took lightly but since she is fortunate to be reasonably healthy and felt no improvement during that six months, she decided that she wanted to get on with living her life while she's able to do so… time will tell whether this was the best decision… but it was her decision and, hers alone.

I think this is one of the aspects that, if she were talking to you today, she would want pharmaceutical companies to take account of when organising drug trials.

Joanne kept a blog of her experience during the ERT trial and at the end she said "that in years to come, when a family is told their child has Morquio disease, the doctor will be able to say to them "but there is a treatment" and I'll know I contributed to that and that's enough for me".

As her Mother, it's not enough for me and I really hope that, if, in the future, Joanne's health does deteriorate and she needs ERT, then it will be available to her. **Judith Evans**

The MPS Advocacy Service

The MPS Advocacy Support Service has been established since the Society was founded in 1982. At this time there were only 40 known families throughout the UK. The support provided was on a voluntary basis and depended heavily on individuals and parents to provide support to individuals diagnosed within their immediate and surrounding areas. However in 1991, the Society opened its first office and with this the advocacy service we know today was born.

The MPS Society provides, through a team of skilled staff, an individual advocacy support service to its members. The service is flexible and a wide range of support is offered on a needs led basis.

The rarity of these conditions means that in many cases, accurate assessments, support and advice are not given due to the vast majority of social care and health professionals knowing very little if anything about the diseases.

Support provided by the team

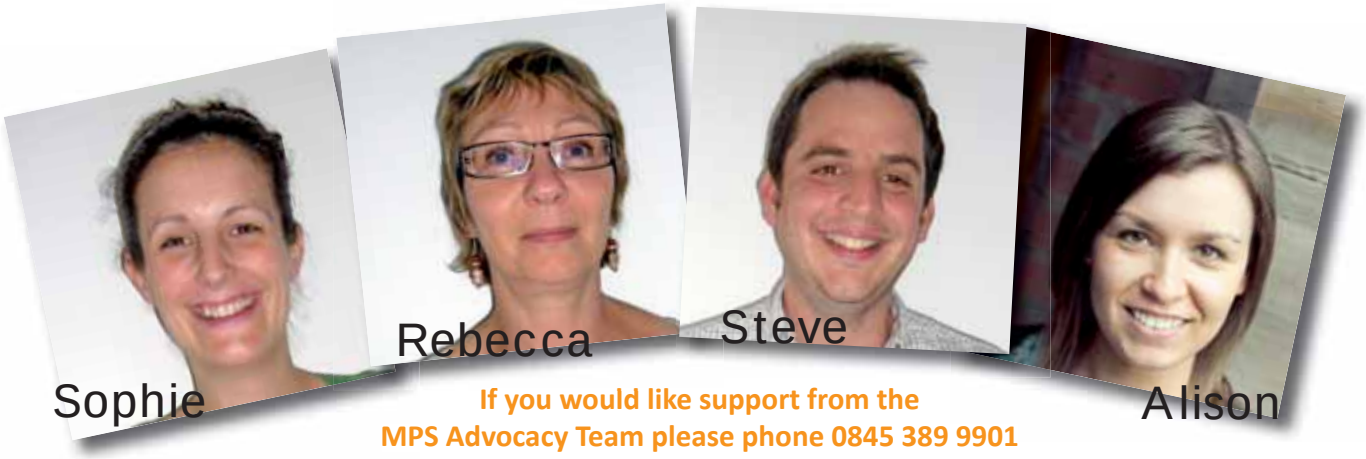
• Telephone Helpline – the Society provides an active listening service, information and support. This includes an out of hours service
• Disability Benefits – in understanding the complexities and difficulties individuals and families have in completing Disability Living Allowance claim forms, the Society continues to provide help and support in completing these forms and, where needed, will take a representative role in appeals and tribunals
• Housing and equipment – the Society continues to take a major role in supporting and advocating appropriate housing and home adaptations to enable the needs of an individual with an MPS or related disease to be met. Where requested, we can provide comprehensive and detailed housing reports based on individual need
• Education – the Society helps members to access appropriate education and adequate provision for its implementation. This is achieved through providing educational reports used to help inform and educate professionals, and in many instances, to inform Statements of Special Educational Need. Where requested, we also provide information days/talks to schools and relevant professionals
• Respite Care – the Society continues to work closely with a number of respite providers and, where appropriate can make individual referrals
• Independent Living/Transition – the Society provides advice, information and support through the transition from child to adult services. This could include access to independent living, learning to drive, further education and employment
• MPS Careplans – the Society undertakes a comprehensive assessment of the issues which need to be addressed when caring and providing support to a specific individual diagnosed with an MPS or related disease, as well as other family members through the writing of a careplan
• Befriender Service – the Society links individuals and families affected by MPS and related diseases for mutual benefit and support
• Bereavement support.

For more information on any of the above or if there is anything else that you would like to chat with the advocacy team about please contact us:
E: advocacy@mpssociety.co.uk
T: 0845 389 9901

Advocacy resources

The advocacy team have also developed a range of information resources focussing on particular issues which are available to download free of charge from the MPS website, **www.mpssociety.co.uk**

- Disability Living Allowance
- Life Insurance
- Travel Insurance
- Hospital Travel Costs
- Disabled Access Holidays
- Carers Legal Rights
- Carers Allowance
- Wheelchairs and Flights
- Guide to Housing and Disabilities Facilities Grant



If you would like support from the MPS Advocacy Team please phone 0845 389 9901 or email advocacy@mpssociety.co.uk

News from the Advocacy Team

A GUIDE TO CHANGES IN THE BENEFIT SYSTEM IN 2013

As many of you will already know, the government are introducing a number of changes to the benefits that individuals receive. Please find below a summary of the current changes and when these will come into force.

Council tax benefit

Council tax support is replacing the current council tax benefit, discounts exemptions and reductions, as of April 2013. Instead of the government paying bills for council benefits, discounts, exemptions, local councils will set up their own schemes and priority for who will receive this support and how much. The only exception to this is help for pensioners where the government will still set rules for this.

Housing benefit

The amount of housing benefit that households receive may be reduced from April 2013. If you live in a council or housing association home and you are deemed by the government to have more bedrooms that is needed, your housing benefit may be reduced. If under the new government rules you are deemed to have one "spare" bedroom then your benefit will be cut by 14%. If you have two or more "spare" bedrooms, you will lose 25%.

Community care grants and crisis loans

Funding for community care grants and crisis loans via the government are being abolished as of April 2013. Instead local council will take over responsibility for providing help in emergency situations. In respect of crisis loans, these will be replaced by a new national scheme of short term advances managed by the Department of Work and Pensions.

Benefits Cap

The government are putting a limit on the amount of money you can receive through benefits. This is coming into effect in April 2013. Some of the benefits included are; housing benefit, jobseeker's allowance, income support, employment support allowance, child benefit, child tax credits and carers allowance (the new universal credit will also be taken into consideration when this is enforced in 2013).

If you receive benefits such as pension credits, working tax credits, disability living allowance, attendance allowance and the support element of employment support allowance this will not apply to you.

From April 2013 the following maximum amounts will apply

- £500 per week for single parents
- £500 per week for couples with or without children
- £350 per week for single people without children

Universal credits

Universal credit is a new benefit system being introduced. It will replace individual benefits already in existence. The benefits included will be housing benefit, income support, jobseekers allowance, employment support allowance, child tax credits, working tax credits, budgeting and crisis loans.

The new system will be introduced in specific areas from April 2013 with a view to this going National from October 2013. Any new claims for benefits (mentioned above) at this point will come under the new universal credit scheme. From April 2014, all people of working age who are in receipt of tax credits will also transfer to universal credits if eligible. People already in receipt of certain benefits will be transferred to universal credit, between April 2014 – October 2017.

Personal independent Payment (Pip)

Disability Living Allowance (DLA) for Adults aged between 16 -64 years is being replaced by a new benefit called Personal independent Payment (PIP).

This is being introduced in some areas of the UK from April 2013 with a view to it being available in all areas from June 2013.

All new claims or renewals will be considered using the new PIP.

If you are currently in receipt of DLA it is envisaged that you will be written to between October 2013 and March 2016, inviting you to make a claim for PIP.

Further information...

For further information please contact the advocacy team on **0845 389 9901** or by email at **advocacy@mpssociety.co.uk**

Further information can also be found on our website www.mpssociety.co.uk under the Advocacy Resources and Downloads section. The advocacy team have developed a range of new advocacy fact sheets on each of these subjects.

New NHS reforms

I want to thank the hundreds of MPS members who wrote to the Prime Minister, Deputy Prime Minister and Member of Parliament regarding the forthcoming NHS reforms that will result in the Advisory Group for Specialised Services (AGNSS). Without so many of you making your concerns known I have no doubt the voice of the MPS Society and the LSD Collaborative would not have had the hearing it has to date.

On 26 October 2012 accompanied by four members who are his constituents we met with the Secretary of State for Health, Jeremy Hunt to take him through our concerns. Subsequently at his behest we wrote to Jeremy Hunt confirming our discussions and concerns that would be taken up by a Health Minister.

When we raised our concerns with you we were most concerned about the loss of the ring fenced LSD drug budget. After discussions with Commissioners and our meeting with Kate Caston, Head of Specialised Services, NHS Commissioning Board, on 7 December 2012 we have decided that in the immediate future Earl Howe's statement, below, is likely to hold. Indeed with approximately 50 new LSD patients a year suitable for Enzyme Replacement Therapy a frozen ring fenced LSD budget may not be in the interests of future members. I am well aware that those on clinical trials, in particular the MPS IVA ERT studies will be asking themselves where does this leave us? I want to assure you that we have raised this question at every opportunity and we will continue to do so.

EARLHOWE STATEMENT IN LETTER
'There are no plans to stop funding treatment for lysosomal storage disorders under the new arrangements for commissioning highly specialised services that will operate from April 2013. The Government hopes that the changes being made to the provision of specialised services through the establishment of the NHS Commissioning Board will lead to more consistency and better access to services for people with rare diseases. The Government's modernisation of the NHS recognises the needs of people with rare and very rare conditions and ensures that no-one should be unable to access treatment because of the rarity of their condition. The Health and Social Care Act 2012 reflects Government's view that specialised and very highly specialised services, which are currently commissioned either nationally or regionally, are best commissioned at a national level by the NHS Commissioning Board rather than locally by clinical commissioning groups.'

Another concern we raised was the Highly Specialised LSD service being dumbed down in the Metabolic Clinical Reference Group (CRF). During our meeting with Kate Caston we received the reassurances we had been looking for. The LSD service will be in a new CRF for Highly Specialised Services. We also understand the LSD Expert Advisory Group, which all the LSD Patient Organisations have a seat, will continue. If these developments are carried through then we can be reasonably confident that those who know the patients' needs best, clinicians and patient groups, will have an advisory role.

The appraisal of new high cost, low volume drugs for rare diseases will be transferred to the National Institute of Clinical Excellence (NICE) on 1 April 2013 and there is absolutely nothing we can do to change this. We are reassured that NICE are working on a new formula to appraise these products and to that end I have been invited to represent the LSD patients on the External Stakeholders reference Group.

So what are the challenges going forward? At the AGNSS LSD Expert meeting on 27 November 2012 the LSD patient organisation Collaborative and the LSD clinicians were told that substantial efficiencies have to be made. A quick gain is to standardise the number of ERT infusions patients receive in hospital before transferring to Home Care. Reducing hospital infusions saves 20% VAT as VAT is only paid on products used in hospital.

Of course if home care is not clinically indicated infusions will continue in the hospital. Currently clinicians are being asked to make other efficiencies across a number of the Lysosomal Storage Diseases and once decisions are finalised we will share them with you.

In the meantime if there is any decision regarding your clinical management or treatment that worries you please do discuss this with your LSD doctor. You are also welcome to contact the Society's Advocacy Team.
Christine Lavery Chief Executive
c.lavery@mpssociety.co.uk

A New National Homecare Specification for Lysosomal Storage Disorders Service

In England representatives of the disease specific patient associations (Battens, Gaucher, Fabry, MPS, Niemann-Pick, Pompe and Krabbe) work together as a Collaborative on many national projects including the recent review of homecare services for patients with a lysosomal storage disorder.

As a family with a child with an LSD, through the national specialised service for lysosomal storage disorders, your child is able to receive care that allows them to receive their ERT infusion in your own home. In the past this set up was managed through each of the eight hospital trusts having their own contract with one or two homecare companies.

However last year, in 2011, the National Specialised Commissioning Team (NSCT) who manage the specialised service for LSDs in England announced that they intended to develop one single homecare contract for the service which would be managed centrally by the Department of Health's Commercial Medicines Unit (CMU). The rationale behind this decision was to a) introduce new companies into the market and b) drive up the quality of service to patients.

A representative of the patient associations was invited to sit on the national project team for the development of the national homecare tender. Over the past twelve months the project team met monthly to develop the tender and worked closely with representatives from each of the eight LSD centres.

Homecare Companies interested in delivering the service were invited to tender for the contract and their bids were adjudicated by representatives from the NSCT, the Commercial Medicines Unit, Patient Group representatives and clinicians and nurse specialists from the eight LSD centres. The successful companies that met the demanding levels of quality required by the service specification were awarded a place on the new framework and are now eligible to work with the eight LSD centres to deliver homecare to their patients. The four companies are Bupa, Central, Healthcare at Home and Medco. The individual LSD centres will now work with the homecare companies to deliver homecare to their patients.

For some families it may be necessary to change homecare provider, this decision will be made by the hospitals taking into account the individual circumstances of their patients. It is recognised that for some families this change will mean a new nurse, driver and customer care support voice on the end of the phone which may take some time to adjust to. However please be reassured that the Homecare Companies are very aware of the impact of these changes and will work with you to develop a good relationship whilst delivering a high quality service. Going forward, if patients and their families feel that they are not happy with the level of service they are receiving through their homecare provider, it is essential to report this to their treating centre and their patient association, as it is only through raising awareness of issues that these services will improve.

Patient Charter

As part of the national homecare specification, the Patient Collaborative developed a 'Patient Charter' which is an integral part of the tender documentation and outlines what you as the customer can expect from your homecare provider. If you would like a copy of the charter please ask your Clinical Nurse Specialist.

Palliative care and end of life

The MPS Advocacy Team is developing a new resource focussing on palliative care, end of life and bereavement. This will be available from the MPS website or by contacting the MPS office. Here we bring you a summary of some of the most important points which may need to be considered…

The new resource has been written to try to provide you with information and help with both practical and emotional issues surrounding end of life care, loss and bereavement. It has been written primarily for families facing the death of a child, young or old, but also for partners facing the loss of a loved one.

Knowing when a child will die is an impossible task. For many children with life limiting conditions their condition can decline at any time. For some death can be quite sudden for others it can be slower, allowing for better acceptance, planning and goodbyes. For some when you think that death is imminent they make a miraculous recovery and can be well for a long period of time afterwards.

As a parent you only want the best for your child to know they haven't suffered and were not in pain. Good practice on behalf of the professionals involved is vital as well as the gathering of information, building relationships, trust and confidence. It is important that families receive full care and support during this time that they are in full receipt of the facts and information in order that informed choices and plans can be put in place without any regrets or questions.
End of life planning

Communication and planning is key, as well as support from professionals experienced in end of life care. A multi-agency meeting should be convened to allow the family to meet the professionals involved and to draw up a care plan, listing individual care needs, agreed medical intervention, management, emergency procedures and intervention, family wishes, expectations and allow for on-going review and changes as directed by either the professional involved or the family. This should include symptom management and issues relating to resuscitation. Twenty –four hour access to a paediatric palliative care team / doctor should be made available to the family especially if the child is cared for at home.

Some children and young people suffering an MPS or related disease may become ill and die very suddenly – often in a matter of days – while for other children, with palliative care needs, this process may happen over a period of many years.

Most parents will have been aware of their child's deterioration for some time and in some cases, will be first to recognise that their precious child is entering the final stage of life. In other cases, it may be the judgement of the healthcare team that are responsible for the care of your child, which breaks the news to you.

The approaching death of your child is likely to be the most difficult time in your life. Some families might feel that acknowledging that their child is dying is a sign that they are giving up and, somehow, letting their child down. Others want and need to be prepared. The desire to make everything perfect is so strong that this in itself can cause extra pressure for the family.

Many people feel that difficult times can bring families together but it can also create divisions. Parents may be at different stages of understanding, acceptance and preparation and it is important to realise that it puts a tremendous strain on relationships. Try to see what is happening, not only from your own point of you but your partner, other children and even grandparents.

The desire to make everything perfect is so strong and can be likened to the period before a birth; this is natural and should be recognised and accepted.

Some parents may find it useful to ask for outside help and this can be sought from your hospice, Palliative Care teams, specialist centres or the MPS Society.

Most parents, at this stage, want to know where their child can die and how they will die. Where your child dies is very much up to you as a family and what you feel is best, you can always change your mind. It is worth having a plan of care in place, which can be continuously reviewed; this should involve all relevant agencies and allow for continuity of care, teamwork and advanced planning (including out-of-hours cover). Include family wishes, favourite pieces of music, poetry etc.
It is worth considering how much you would like any siblings involved and what practical and emotional support they will require. Brothers and sisters, who are involved at this time, generally cope better in the months following the death of their brother or sister but their wishes obviously need to be respected and will allow them to feel more in control.

It is one of the most difficult series of decisions you will make but once completed, it can be reassuring to know that everything is in place.
Parents obviously do worry how they will cope at this time and some families, when looking back, are surprised at the strength they found and how well they got through it.

On becoming aware or being informed that your child is in the final stages of life, it is time to consider the options, discuss these and any concerns with the care team who will be able to reassure you.

Our factsheet provides further information on the different options and choices available to you and covers a number of other topics including: What happens after my child dies?, Who do I need to contact?, Funeral preparations and choices, Keepsakes and memories, How we grieve plus a range of useful support resources and books for both children and adults.

Deaths

We wish to extend our deepest sympathies to the family and friends of:

Benjamin Illingworth who suffered from Sanfilippo and who passed away on 25 October 2012 aged 9 years.

Jamie Moxon who suffered from Hunter and who passed away on 17 November 2012 aged 19 years.

Anabelle Shepherd who suffered from Hurler and who passed away on 25 December 2012 aged 1 year.

The Childhood Wood Planting Day 2012 will be featured in the next edition of the MPS Magazine.

In memory

Benjamin Illingworth

20 November 2002 - 25 October 2012

Our beautiful and loving son Benji who suffered from Sanfilippo disease passed away very suddenly and unexpectedly on 25 October, aged 9.

It was such a shock that our special little man passed away peacefully in his sleep at home, as we hoped that we had him for several more years to come, but we take comfort in that he did not suffer and is now at peace surrounded by those who love him in heaven.

We may only have had a short time with you, but what a great 9 years of happy memories we have of you. You touched so many lives and were deeply loved by all of your family and friends. I hope that you know how much you touched our hearts and how much we all loved you. You gave us unconditional love and made us appreciate the things we had, and that the simple things in life are the best.

The ladies remember you as a bum slapper as they walked past you, for a little hand would always come out to give them a pat as they passed you! Everyone talks of your lovely cuddles, your endless supply of big kisses, your amazing smile which could light up a room and your infectious laugh - these are the things we will miss the most about you.

They say memories are
golden, well, maybe that is true.
I never wanted memories, I only wanted you.
A million times I cried.
If love alone could have saved you, you never would have died.
In life I loved you dearly, in death I love you still.
In my heart you hold a place no one else could fill.
If tears could build a stairway and heartache make a lane.
I'd walk the path to Heaven and bring you back again.
Our family chain is broken, and nothing seems the same.
But as God calls us back one by one, the chain will link again.

Sleep tight our little angel, we cannot wait to see you again and feel the warmth of your touch and kiss as we share a big hug again. Forever in our hearts and thoughts your remain, never shall we be parted. God bless you Benji, we will love and miss you forever.

All our love, Mummy, Daddy, Ollie
and all of your family and friends xxx

All Ireland Advocacy Support

The All Ireland Advocacy Support Service is continuing to grow. Alison Wilson, All Ireland MPS Advocacy Support Officer is delighted to have been able to support many more families since she last updated you on this service. As usual she brings just a brief update on the work we do in Ireland and what we hope to do in the future.

All Ireland Advocacy Support

It's been a busy but inspiring 2012 for the All Ireland Advocacy and Support Service. I am delighted to have been able to provide practical and emotional support to many families across the Island of Ireland.

I am always amazed by the courage and determination of families affected by MPS and related diseases - when times get tough our families get tougher. It has been my pleasure to support families through some tough times but also to join with them in celebrating all the little victories they have had over the last year.

The current economic climate, both in Ireland and the UK, has resulted in families being put under additional financial pressures. These pressures can have a huge impact on how families cope on a day to day basis and in some cases life can start to feel unmanageable. Our hope is that through the support of the Advocacy Support Team we can support our families to face these financial difficulties with courage and confidence.

I'm always delighted to hear a new voice on the end of the phone and would encourage you to spread the word about the All Ireland Advocacy Support Service to your friends in Ireland.

Northern Ireland MPS Clinic

The last Northern Ireland Winter MPS Clinic was held on Friday 30th November 2012.

This was another very busy and productive clinic! The medical team - Dr Fiona Stewart, Dr Simon Jones, Dr Siobhan O'Sullivan, Mrs Jean Mercer, Mrs Siobhan Harding-Lester (Genetic Counsellor) - had appointments with 15 of our members and I was able to catch up with all of the families who attended.

As usual the clinic proved to be the perfect place to discuss family support. In the months following the clinic I will be meeting with families in their homes to address any support needs that were identified.

We were delighted to welcome a new member to our team in Belfast. Dr Siobhan O'Sullivan is the new Metabolic Paediatrician based in the Royal Victoria Hospital in Belfast. I look forward to working closely with Dr O'Sullivan in supporting our families in the future.

Fabry Clinic

The last Joint Cardiac and Genetics Fabry clinic was held on 27th November 2012. This clinic is evolving into a 'one stop shop' for Fabry patients in Northern Ireland. By including as many investigations as possible in this clinic we have cut down the number of hospital trips for our patients.

As a relatively new clinic we are always keen to hear any feedback you have from the clinic.

Again, we were delighted to welcome Dr Siobhan O'Sullivan to our team. Dr O'Sullivan will be looking after all children who attend our clinic.

Southern Ireland

In the last few months I have had an influx of requests to visit schools in Southern Ireland to educate school staff and other associated professionals about MPS.

An MPS Society School Education Session is an excellent way to explain your child's needs to school staff and to provide an opportunity for staff to ask questions about their condition and any potential adaptations and/or accommodations that may need to be made. It is essential that schools understand the needs of your child and take steps to ensure that the school environment is suitable to support your child in reaching their academic and social potential.

In the last few months I have given education sessions in relation to MPS I, II and IV and have found that the schools really valued the information they received. If this is something you feel would be of benefit to your child and their school please do get in touch. As well as providing general education sessions we can also draw up more detailed care plans to support schools in meeting your child's needs.

MPS I clinic
On 8th and 9th November 2012 I attended the MPS I specialist clinic at Our Lady's Children's Hospital in Dublin. At this clinic I was able to meet with 35 individuals with MPS I and their families.

This was a very busy clinic with input from the Bone Marrow Transplant Team, the Metabolic Team from Temple Street Metabolic Unit and the Orthopaedic Team. Other specialties (dental, ophthalmology, physiotherapy and others) were also available on site for those who require any follow-up. As usual I was available to provide support and advice in relation to social welfare, housing, education and access to services. I have a very long 'to do' list from the clinic and look forward to meeting many of the families again in the coming months to support them in any way that I can.

There are big changes ahead for this clinic as Dr Anne O'Meara (Consultant Paediatric Oncologist) works towards her retirement. I hope to bring you an update of how this service will move forward in our next magazine.

Advocacy and Support for the Gaucher families in Ireland
In the new year my post will be extended to include an Advocacy Support Service to individuals and families affected by Gaucher Disease in Ireland. Gaucher Disease is another of the Lysosomal Storage Disorders. I hope that my experience in supporting MPS patients in Ireland will help me in supporting this new group of patients.

Get in touch...
If you would like support from the MPS Advocacy Team please phone **0845 389 9901** or email **advocacy@mpssociety.co.uk**

Shared experiences from our members...

"I'd love to hear how other parents deal with the issue of toe-walking children with MPS and suitable footwear. My little girl is 11 and requires orthotics. She is also a toe-walker as she has MPS1 H/S. She hates all the specialist/ practical footwear out there and wants to wear trendy footwear like her friends, but unless it's a boot or high-top trainer...nothing works with the orthotic. I'd love some advice from other teenage/adult female MPS sufferers on how they cope with the issue of footwear with orthotics or if they can recommend any particular shops. Many thanks Helen Lever."

MPS Regional Specialist Clinics

The MPS regional clinic programme involves centres including Manchester, Belfast, Birmingham, Bristol, Cardiff, London and Newcastle.

The programme aims to enable individuals affected by MPS and Related Diseases to access a consultation with a tertiary centre consultant without having long distances to travel to their designated specialist centre. The clinics also aim to increase expertise in the regional centres on MPS and related diseases in the clinical management of those affected.

The MPS Society supports the regional clinic programme, which can include arranging and co-ordinating dates of the annual programme and funding the tertiary centre doctors' travel and subsistence costs to the regional centres.

Specific arrangements for each clinic and for organising appointments differ for each of the centres. At least one member of the Society's advocacy support team meets individuals and families face to face to offer individual advocacy support.

You can read more about recent clinics on these pages...

Bristol Clinic

10th October 2012

We certainly know that autumn is on the way! Being dark outside means I should be asleep! Luckily Steve was awake… just as well as he was driving; the newest addition to his family had woken up at 3am.

The traffic was ok till we got to Bristol; luckily the families were also running late. Dr Pierre and Dr Jones (from Manchester) were ready and waiting to greet the families.

Farhaan arrived with his mum and had been getting over a cold but seemed happy enough, probably due to not having to go to school that morning.

Terry, his mum and little sister had only a short journey so didn't have the issues with the traffic; Terry was content to listen to his music while he waited.

Tara decided that she wasn't going to have her photo taken and was not impressed that mum had not brought her a change of clothes so she could get out of school uniform.

Merlin was next on the starting blocks, he had a lovely time waving his 'Dora the Explorer' balloon around, and a few consultants may have had a few near misses as she 'explored' the clinic room.

Oliver was on fine form although mum has decided to jump out of a plane to raise some money and I think is beginning to think it may not be such a good idea, hopefully it will be plain sailing…

We also had a chance to meet some of our adult members, before we hit the road home.
We look forward to seeing you all again soon.
Rebecca Brandon & Steve Cotterell



Bristol Clinic photos from left to right: Farhaan Ahmed (Fucosidosis), Merlin Penny Smith (Hunter) ; Terry Butler (Hurler)

Bone Marrow Transplant Clinic

12th October 2012

I arrived at Manchester Children's Hospital and soon people joined me in the waiting room. I had a chat with Callum and heard of his planned trip to Lapland this winter, after hearing the tales of the lucky few who went last year with the MPS Society I advised that he takes his thermals!

Melissa too was excited as she and her family were off to Australia to visit family. As ever it was a pleasure to see Rachel and Cody.

I would like to thank the team at the Willink for making me feel welcome and we look forward to the next clinic.
Steve Cotterell, Advocacy Officer
steve.cotterell@mpssociety.co.uk

Bone Marrow Transplant Clinic

19th October 2012

Well it was another early start to get to the clinic and this time it was still dark when I got to the train station, winter is on the way.

Arrived at clinic but they were running late. Sonny arrived with his family and was straight into playing in the little house, I think he was glad to stretch his legs after the four hour car journey. Talking of stretching legs, Sonny's mum had just completed a half marathon in aid of the MPS Society, well done to her. I think she was still suffering…

Charlie arrived with his brand new red wheelchair, Charlie is a Liverpool supporter hence the red wheelchair.

Well there were lots of comings and goings and soon it was time to head back home.

Thank you to the team and we look forward to seeing you all again soon.

Rebecca Brandon, Advocacy Officer
r.brandon@mpssociety.co.uk

Photos clockwise from top left - BMT clinic 12 October: Melissa McKie; Rachel Rothwell; Cody Taylor; Callum Pollock and family.
BMT clinic 19 October - Carlette and Charlie Escalonilla , Sonny Gibbard

Great Ormond Street MPS III Clinic

13th November 2012

The MPS III clinic was held within the busy outpatients department, Niamh, Victoria and the multidisciplinary team were ready and waiting. Fortunately I managed to secure some seats near the TV and Mister Maker was distraction enough to keep the children entertained and in one place long enough for a photo or two. I think the only people I missed were Roshani and her family, Roshani was giving a guided tour of the hospital at high speed, I'll catch her for a photo next time. With thanks again to the team at GOSH.

Steve Cotterell, Advocacy Officer

GOSH MPS III Clinic photos clockwise from top left - Anna Jabeen, Daniel Zaldua, Luke Bown and Lily Brooker

Birmimgham Clinic

23rd November 2012

It was a foggy start for Steve and myself, luckily we only had a short journey to the train station and then 'let the train take the strain'.

The clinic was in the research centre which is tucked away from the mayhem of the children's outpatients.

Thomas had arrived and been seen and was on his way out when we got there. He managed a lovely smile and made a gruffalo growl at Louise which made him giggle. Jensen came with his mum and gran and I have a sneaky feeling he likes his visits to the hospital as he has a chance for some shopping and going to his favourite restaurant. He also beat Steve in the interactive football not once, but 3 times, Steve tried to blame it on a gammy leg! Mohammed had a great adidas t-shirt and would much preferred to haev been at school rather than have the pleasure of our company.

Fahim looked very smart in his waistcoat and was very content just waiting to be seen. Charlie arrived full of beans and then proceeded to beat me in the football, unlike Steve I am blaming it on wearing heels… This is beginning to sound like a fashion article!

Well we saw lots of other families and children and then caught the train home, just a bit worried at one stage as one of the tunnels was flooded and we had to slow right down, luckily we didn't get stuck, that would not have been a good start to the weekend. Thank you to all the team.

Rebecca Brandon & Steve Cotterell

Photos clockwise from top left:
Thomas Kynaston (Maroteaux Lamy),
Mohammed Gul (Morquio),
Fahim Hussain (Sanfilippo),
Charlie Warren (Fabry),
Jensen Bechemin (Fabry)

Volunteering opportunities at MPS

Can you volunteer your time caring for MPS children and their siblings at events run by the MPS Society?
Volunteering is fun and rewarding. It could also help you learn new skills and gain valuable work experience.

We are always looking for new volunteers to help out with events and conferences that we run throughout the year. The MPS Society relies on volunteers to assist in the care needed for children and young adults affected by MPS and Related Diseases. All of our volunteers undertake training in moving and handling and are fully briefed prior to the event.

Becoming a volunteer: The MPS Society accepts volunteers from the age of 16 years on a trainee basis. We will require you to undergo an enhanced Criminal Records Bureau check as the Society supports children and vulnerable adults. If you are a new volunteer we also require two references and ask you to attend a compulsory training day at MPS House in Amersham. All new volunteers will be mentored by an experienced volunteer.

Please can you help us? Email: mps@mpssociety.co.uk or phone 0845 389 9901

Alton Towers Fun Weekend

On 14 – 16 September, Sophie, Laura and Antonia from the MPS office travelled to Alton Towers for a weekend of fun for all the family funded by the Gosling Foundation and the Clover Trust. We were joined by many MPS families. The feedback we had from many of the families was that the weekend was a fantastic success. Many of you enjoyed the laid back, informal atmosphere and it was a great opportunity for all the family to spend some time together and meet up with others.

The Brentnall Family

Thank you to everyone at the MPS Society and MPS Supporters for organising and contributing to the weekend trip to Alton Towers. The Splash Landings Hotel and Waterpark were fantastic and we all had a great time together going on the rides and attractions.

Jacob had lots of fun playing in the waterpark on the Friday but drank too much pool water and made himself ill later that evening (a very big thank you to the hotel staff for quickly mopping up the floor in the bar!). Jacob soon bounced back though and we had two wonderful days on Saturday and Sunday in the park. Both Jacob and Samuel especially enjoyed 'The Blade' and 'The Flume' rides.

It was nice to meet some new faces as well as catch up with other families that we have met at previous MPS events.

Chris & Hannah Brentnall
(Jacob has MPS III type B)

The Bradshaw Family

We had a lovely day at the park - we didnt get there until lunch time but we still managed to get to see a few of the attractions. We were so pleased the weather was nice and sunny. Emily just loved the pirate section - she kept pointing to the pirates walking around saying they were her friends!

We took her on the squirrel nutty ride which she enjoyed and then went towards the Ice Age 4D section - she really wanted to go on the cars but unfortunately she was just too short. We took her in the Ice Age 4D attraction - she had never seen Ice Age before but seemed to love it - especially the snow (although she wouldn't put the glasses on).

After this we made our way back to the pirate section where we were going to go on the boat ride but ended up firing water at all of the other people on the boats - at which point we realised we would get drenched if we went on the boats so instead we decided to drench everyone else - Emily was squealing with laughter at this point.

Once the park had closed we made our way to the Splash Landings Hotel where Emily was able to sit and do colouring with the entertainers whilst myself and Nev had a cuppa and a rest. She also got to meet the Ice Age characters which were walking around - we thought she may have been a little scared of them as they were so big but she was so excited to see them she was showing them her pictures she had coloured.

The best part of the day for Nev and I was the evening where we got to meet some lovely families - this was the first time we had met any other families who had experience of MPS. Helen, Nigel and their daughters sat on our table and we were so please to be introduced to them - they are such a lovely family. We are now in regular contact with them.

Emily really enjoyed the pass the parcel and the disco - it was the first time we had seen her dancing away - such a lovely memory to take away.

We definitely want to go on the next MPS event as we found it so reassuring meeting other families who have gone through the same as ourselves and also seeing how well their children are getting on.

Thank you so much MPS Society for organising the event - it was such a memorable day.

We were so impressed with the hotel and really wished we had booked accommodation - we have now booked a Santa sleepover for Emily this Christmas which we are all really excited about. **Gayle & Nev Bradshaw**

UK Genetic Testing Network Conference

To celebrate its 10th Anniversary the UK Genetic Testing Network (UKGTN) held a conference on 22 November 2012 in London looking at Changes to Patient Care attended by clinicians, scientists, commissioners and patient organisations.

The UKGTN focuses on promoting equity of access to genetic tests for patients and their families and the quality of laboratory services. The evaluation of new tests is a key component of UKGTN's work to drive effective and efficient diagnostic care pathways that are coming about as a result of genomic advances.

During the meeting Alastair Kent, Director of the Genetic Alliance talked about UKGTN's work in developing a set of quality assured genetic tests with clear commissioning criteria and for which information is easily accessible to patients, the public, clinicians and commissioners understand what can be expected and from whom to expect it. It is vital that patients and their families know what services they can access or receive and what the benefit of these tests will provide.

As a member of the UK Genetic Testing Network it was my pleasure to acknowledge the importance of the work carried out by the UKGTN as it goes forward.

It has been really important to the Society for Mucopolysaccharide Diseases (MPS) to be working with the UKGTN as a member of the Clinical and Scientific Advisory Group. The MPS Society supports over 1200 families across 24 lysosomal storage diseases. Strong relationships with accredited diagnostic laboratories and genetic centres across the UK are vital to our members through our advocacy support service. The UKGTN website provides our members and those of other rare disease groups with a unique and reliable information resource as to the availability of genetic tests and their indications. Whilst one immediately thinks of genetic testing and genetic services as running hand in hand with the prima facia case of a genetic disease diagnosis in fact in the MPS Society the greater number of enquiries come years down the line from siblings of those affected by genetic disease. Some families come recalling results of 'hair root tests' the science for carrier testing 15 – 20 years ago and are taken aback when we are able to tell them of advances and refer them to the UKGTN website.

Without a doubt new technologies have provided a much greater understanding of genetic diseases and the work of the UKGTN has been pivotal in ensuring even in the case of patients with the most complex clinical needs and their families that they have equitable access to the advantages resulting from the UKGTN genetic science programme. In the same vein the MPS Society and many other rare disease patient organisations continue to encourage the UKGTN to build on its innovative plans and in particular supporting the use of genetic tests by relevant specialities as part of agreed clinical and diagnostic pathways as well as strengthening existing good models of care with greater involvement of genetic services.

Christine Lavery MBE, Chief Executive
c.lavery@mpssociety.co.uk

Reliable Accurate Prenatal Non-invasive Diagnosis (RAPID)

New cell-free fetal DNA (cffDNA) testing is now available during pregnancy to women who are carriers of sex-linked genetic conditions. Sex-linked conditions either affect only males or only females. The cffDNA test can identify the sex of the baby early in pregnancy.

Why does the test work?

Genes contain the DNA which determine many of our characteristics including whether we are male or female. We now know that some of the baby's DNA circulates in the mother's blood during pregnancy. We call this cell-free fetal DNA (cffDNA). This cffDNA can be tested and the baby's sex determined by taking a sample of the mother's blood in pregnancy. At the moment the test can only tell us the baby's sex, not whether the baby is affected by a particular condition.

How is the test done?

The timing of the cffDNA test is important as it works best after 7 weeks of pregnancy. You will need to have an ultrasound scan first to find out exactly how many weeks pregnant you are and whether there is more than one baby in the womb, as the test works best when there is only one baby. The cffDNA test is performed on a sample of the mother's blood. About 10mls or roughly one table spoon, is taken like a normal blood test. The blood is then sent to the genetic laboratory for testing to see whether there is a 'male' (also known as Y) chromosome in the blood sample. If there is, this shows the baby is a boy because females do not have this male chromosome. If no Y chromosome is present the baby is female. The cffDNA is lost from the maternal blood stream within a few hours of delivery and so testing is specific to the baby in that pregnancy.

How accurate is the test?

Cell-free fetal DNA testing is 99.5% accurate, however sometimes the laboratory is unable to give a result. This might be because there was not enough fetal DNA present in the blood sample to perform the test. If this happens the test can be repeated. Due to the very small chance of an incorrect result, the baby's sex should always be checked by ultrasound scan.

How safe is the test?

As this is a blood test taken from your arm, the test carries no significant risk to you or your baby.

How long does it take to get the test results and how will I get them?

It usually takes about a week to get the results. Your doctor or healthcare specialist will discuss this with you, including how you will get your test results.

What happens next?

Depending on the results of the cffDNA test, you might decide to have an invasive test to determine if the baby has the condition you are concerned about. Alternatively you might decide to continue with the pregnancy without any further testing. Your doctor or healthcare specialist will discuss these options fully with you.

http://www.rapid.nhs.uk/uk-testing-centres/fetal-sex-determination

Information and Education Resources available from the MPS Society

Resources: We publish a number of resources aimed at different audiences – children, parents and carers, grandparents and professionals. They cover a whole range of different topics, including detailed information on each of the MPS and related diseases including Fabry, particular issues covered by the advocacy team such as housing, benefits and travel insurance to guidance on fundraising. There are also general guides on the management of anaesthesia, the pattern of inheritance and challenging behaviour.

Some of these resources are available to download free from the MPS website under Resources and Downloads, or you can order them for a small charge online from our MPS online shop. Check out **www.mpssociety.co.uk**. Alternatively, place your order over the phone on **0845 389 9901** or ask us for a publication order form to be sent to you in the post.

Website: Thank you to everyone who took part in our survey on the MPS website. The majority of you found it easy to navigate and were able to find the information you were looking for. If you have any further thoughts on how we can improve the site, please do email us at **website@mpssociety.co.uk**. Don't forget you can download many of our resources and materials directly from the site. There is plenty of information on our different activities and how you can get involved, either by volunteering or helping us with fundraising. We also update our latest news regularly so don't forget to have a look to see what's new.

MPS Magazine: The MPS Magazine goes out quarterly to all our members, supporters and subscribers. It is one of our main ways of communicating with you, telling you all about what's been happening with our events, research, treatment updates, fundraising, and for you to share your stories and news. You can subscribe to the MPS Magazine by completing the relevant forms online.

We do get a few people suggesting we should send the magazine out by email, rather than wasting money on printing and postage and to be kinder to the environment and this is something we continue to consider. However, our membership is very diverse and the majority still like to receive their copy in the post to pass on to our family, friends, colleagues etc. We obtain very competitive quotes which we review each edition and the grants we receive for the printing are restricted for that particular purpose so can't be used for anything else. Again, if you have any suggestions for how we can improve the MPS magazine, or would like to submit an article please email **magazine@mpssociety.co.uk**.

Lysosomal Storage Diseases: A Practical Guide

This new book co-edited by Atul Mehta (Royal Free Hospital, London) and Bryan Winchester (a Trustee of the Society and formerly of the Institute of Child Health at Great Ormond Street Hospital) is the first to bring together all aspects of lysosomal storage diseases in a single volume. It is aimed at clinicians and scientists entering the field but will be equally useful for health care workers and administrators, those working in the pharmaceutical industry and patients and their families and organisations.

The first part of the book reviews our current understanding of the scientific, clinical and genetic background of the diseases. This is followed by succinct accounts of the individual diseases. The book concludes with a discussion of current and potential therapies, screening and the patient perspective, to which Christine Lavery contributed. Each of the 24 chapters is written by an acknowledged expert in the field and there is a comprehensive classification of the diseases and index.

This publication has been made possible by an unrestricted educational grant to The MPS Society from Shire HGT. In consequence the editors will be very happy to send a complimentary copy to anybody who feels that the book will help them in their professional work or support for families affected by LSD. Please send your name and postal address to the MPS Society (t.ellerton@mpssociety.co.uk). The editors hope that this book will increase awareness of the LSDs and stimulate further research and support for patients.

The guide is Published by Wiley-Blackwell. (ISBN 978-0-470-67087-3; 197 pages) and is also available for purchase on Amazon.

Amicus Therapeutics Achieves Target Enrollment in Second Phase 3 Fabry Monotherapy Study

56 Patients Now Randomized to Switch from Enzyme Replacement Therapy (ERT) to Migalastat HCl or to Remain on ERT Final Enrollment Expected Ahead of Year-End Target

CRANBURY, NJ, US, October 22, 2012 – Amicus Therapeutics (Nasdaq: FOLD), a biopharmaceutical company at the forefront of developing therapies for rare and orphan diseases, today announced that it has now achieved target enrollment and has 56 patients in its second Phase 3 study (Study 012) of migalastat HCl monotherapy for Fabry disease. Screening is now closed at participating sites, and final enrollment is anticipated by year-end 2012. Amicus in collaboration with GlaxoSmithKline (GSK) is developing the investigational pharmacological chaperone Migalastat HCl for the treatment of Fabry disease.

Study 012 (The ATTRACT, or FAB-AT1001-012 Study) Highlights:

- First clinical study to compare oral migalastat HCl to standard-of-care ERTs (Fabrazyme® and Replagal®)
- Enrolled males and females with Fabry disease, who had genetic mutations amenable to migalastat HCl as a monotherapy, and were on ERT for a minimum of 12 months
- Primary outcome measure is renal function assessed by Glomerular Filtration Rate (GFR) at 18 months.

John F. Crowley, Chairman and Chief Executive Officer of Amicus, stated, "We are very pleased to have met the enrollment objectives for this important Phase 3 Fabry monotherapy study ahead of the year-end target. The willingness of these Fabry patients to switch from an approved ERT to migalastat HCl to participate in this study highlights the unmet medical needs that persist in this community. We look forward to evaluating the effects of migalastat HCl, as well as currently used ERTs, on renal function over the course of 18 months of treatment in this study."

Amicus and GSK are co-developing all formulations of migalastat HCl under a global Fabry collaboration. Migalastat HCl monotherapy is in Phase 3 development (Study 011 and Study 012) for Fabry patients with genetic mutations that are amenable to this chaperone monotherapy, as determined by a cell-based assay. Study 011 is a placebo-controlled study intended primarily to support U.S. registration, and Study 012 compares migalastat HCl to ERT to primarily support global registration. Migalastat HCl coadministered with ERT is in Phase 2 (Study 013) and migalastat HCl co-formulated with JCR Pharmaceutical Co. Ltd's proprietary ERT (JR-051, recombinant human alpha-Gal A enzyme) is in preclinical development. Amicus has commercial rights to all Fabry products in the United States and GSK has commercial rights to all of these products in the rest of world.

Genet Med. 2012 Sep;14(9):779-86.

Clinical observation of patients with Fabry disease after switching from agalsidase beta (Fabrazyme) to agalsidase alfa (Replagal).

Tsuboi K, Yamamoto H.

Source: Department of Hematology, Nagoya Central Hospital, Nagoya, Japan.

ABSTRACT

Purpose: Fabry disease is a rare, X-linked, inherited lysosomal storage disorder that can be treated with the enzymes agalsidase alfa (Replagal) and agalsidase beta (Fabrazyme). Currently, there is a global shortage of agalsidase beta, and this has increased global demand for agalsidase alfa. We assess the feasibility of switching patients on agalsidase beta treatment to agalsidase alfa instead.

Methods: This analysis is part of an ongoing observational study involving 11 patients with Fabry disease in whom the treatment was switched from agalsidase beta (1 mg/kg every other week) to agalsidase alfa (0.2 mg/kg every other week). Data were collected for a minimum of 36 months: 24 months before and 12 months after the switch. Serial data were evaluated with respect to renal function, cardiac mass, pain, quality of life, and tolerability/safety.

Results: Indexes of renal function (estimated glomerular filtration rate) and cardiac mass (left-ventricular mass index), pain (Brief Pain Inventory), and quality of life (EuroQoL-Dimensions) clearly showed that, in patients switched to agalsidase alfa, Fabry disease stabilized during the 12 months of follow-up.

Conclusion: Despite the limitations of this preliminary observational study, it was found that all the patients maintained disease stability when treated with agalsidase alfa, as evidenced by estimated glomerular filtration rate, left-ventricular mass index, pain scores, and quality-of-life indexes, throughout 12 months of follow-up.Genet Med 2012:14(9):779-786.

BioMarin Phase 3 Study of GALNS for the Treatment of MPS IVA Meets Primary Endpoint

SAN RAFAEL, Calif., November 5, 2012 - BioMarin Pharmaceutical Inc. (Nasdaq: BMRN) announced today that the pivotal Phase 3 study of GALNS met the primary endpoint of change in six-minute walk distance compared with placebo at 24 weeks in subjects receiving weekly infusions of GALNS at the dose of 2 mg/kg (p=0.0174).

MOR-004 was a randomized, double-blind, placebo-controlled study evaluating two doses of GALNS (BMN-110, N- acetylgalactosamine-6-sulfatase) for the treatment of patients with the rare lysosomal storage disorder Mucopolysaccharidosis Type IVA (MPS IVA), also called Morquio A Syndrome. Patients dosed with GALNS at 2 mg/kg every other week did not show a meaningful or statistically significant change from baseline compared to placebo. The company also announced preliminary data from the MOR-005 extension study which suggests that clinical benefits continue to improve with further dosing with GALNS. Only a limited number of patients have reached the 36 or 48 week points of total time on treatment in the extension study, and the results will be updated when the study is completed. The company confirmed that based on the results of MOR-004, and following planned discussions with regulatory authorities, it expects to submit marketing applications starting in the first quarter of 2013.

Treatment with GALNS Significantly Improves Primary Endpoint

The primary endpoint of the study, change in six-minute walk distance at 24 weeks, was statistically significant in patients dosed with GALNS at 2 mg/kg every week with a mean increase of 22.5 meters (p=0.0174) over placebo. In MOR-004, patients dosed at 2 mg/kg every week showed an improvement in six-minute walk distance at week 12 compared to baseline and showed continued improvement at week 24. Preliminary analysis of a subset of the patients in the MOR-005 extension study who have reached the 36 week and 48 week timepoints in the study also showed further improvement at weeks 36 and 48.

Treatment with GALNS Improves Both Secondary Endpoints

On the secondary endpoint of three minute stair climb, patients dosed with GALNS at 2 mg/kg every week showed a trend toward improvement at 24 weeks of 1.1 additional stairs per minute over placebo. In MOR-004, patients dosed with GALNS at 2 mg/kg every week showed an improvement in three-minute stair climb performance at week 12 compared to baseline and showed continued improvement at week 24. Preliminary analysis of a subset of patients in the extension study (MOR-005) who have reached the 36 weeks and 48 week timepoints in the study also showed further improvement in three-minute stair climb performance. In the other secondary endpoint, urinary keratan sulfate (KS) levels, patients dosed with GALNS at 2 mg/kg every week showed consistent and robust reduction in urinary KS with a mean difference from baseline as compared to placebo of 40.7 percent (p<0.0001). Preliminary analysis of a subset of patients in the extension study (MOR-005) who have reached the 36 weeks and 48 week timepoints in the study showed this level of reduction was maintained.

Treatment with GALNS Improves Pulmonary Function

Pulmonary function, as defined by maximum voluntary ventilation (MVV) was measured at 24 weeks. In MOR-004, patients dosed with GALNS at 2 mg/kg every week showed a trend towards improvement from baseline of 10.3 percent over placebo. Preliminary analysis of the subset of patients who reached the 48 week timepoint showed a reduction in the improvement, though an increase over baseline was maintained.

Pulmonary function, as defined by forced vital capacity (FVC) was measured at 24 weeks. In MOR-004, patients dosed with GALNS at 2 mg/kg every week showed a trend towards improvement from baseline of 3.3 percent over placebo. Preliminary analysis of the subset of patients who reached the 48 week timepoint showed continued improvement.

Safety Summary

In MOR-004, GALNS was generally well-tolerated and adverse events were similar to those seen in clinical trials of other enzyme replacement therapies. The most common adverse events occurring in more than 25 percent of treated patients included vomiting, pyrexia, headache, nausea and cough. Serious adverse events that were thought to be related to study drug occurred in 3.4 percent of the weekly group, 1.7 percent of the every other week group and 0 percent in the placebo group. There were no deaths and no patients withdrew from the study due to an adverse event.

Infusion-associated reactions were generally mild to moderate and manageable with symptomatic treatment and/or infusion rate modification. Of the 1,345 total number of infusions in the weekly dose group, 17 infusions (1.3 percent) were interrupted or discontinued due to an adverse event. All patients subsequently resumed dosing.

“The positive results from this pivotal study will help support GALNS as the first therapy available to help the approximate 3,000 people worldwide suffering from MPS IVA -- a rare, degenerative, life-threatening genetic condition with no available therapy,” said Hank Fuchs, M.D., Chief Medical Officer at BioMarin. “We are very pleased with the clarity that the MOR-004 study has provided us with respect to the appropriate dosing of GALNS. The weekly 2 mg/kg dose provided a statistically significant and clinically meaningful improvement in the study’s primary endpoint, and positive trends toward improvement in other clinically meaningful endpoints, including three-minute stair climb and pulmonary function tests. By contrast, the 2 mg/kg every other week dose was shown to be similar to placebo on the primary and clinical secondary and tertiary endpoints. We look forward to reviewing the results of this study with regulatory authorities, and applying for marketing authorizations starting in the first quarter of 2013.”

“The GALNS clinical program is currently the highest development priority at BioMarin, and this positive Phase 3 study serves as a potentially transformative milestone for the company,” said Jean-Jacques Bienaimé, CEO of BioMarin. “We are applying our track record of success in developing novel treatments for orphan diseases and our existing commercial infrastructure for Naglazyme to bring GALNS to patients as rapidly as we can.”

Shire

Shire Enters into Novel Research Collaboration with Italian-based Telethon's Institute of Genetics and Medicine for Rare Diseases

Nyon, Switzerland – October 24, 2012 – Shire plc (LSE: SHP, NASDAQ: SHPG), today announced a long-term, broad based, multi-indication research collaboration in rare diseases with Fondazione Telethon, a major Italian biomedical charitable foundation, for research carried out at the Telethon Institute of Genetics and Medicine (TIGEM). This alliance will facilitate research on 13 undisclosed rare disease indications and has the potential to add multiple, novel therapeutic candidates into Shire's early stage pipeline. The partnership underscores Shire's long-term commitment to bring innovative therapies to patients with rare diseases worldwide.

The collaboration brings together Shire's established capabilities in developing and distributing effective, life-altering therapies for patients with rare diseases and TIGEM's world renowned research expertise in gene therapy and other novel therapeutics. Under the terms of the agreement, Shire will provide $22 million (€17 million) funding over five years for several research projects that collectively address a number of different lysosomal storage disorders and neurodegenerative diseases. The majority of the research will be conducted in TIGEM's facility in Naples, Italy under the direction of Andrea Ballabio. Successful projects that arise from this research will be incorporated into Shire's development pipeline and benefit from additional Shire investment and resources.

"As a leader in rare diseases, Shire's partnership with Fondazione Telethon is another way for us to ensure that we expand into new disease areas and enhance our collaborative relationships with academic institutions," said Philip J. Vickers, Senior Vice President, Research and Development, Shire HGT. "Shire's developmental expertise combined with TIGEM's early stage research capabilities will enable us to accelerate our discovery and development efforts. This research collaboration is evidence of Shire's commitment to patients with rare diseases and our intent to work with the best institutions to achieve this."

"The collaboration with Shire proves yet again the value of Telethon-funded research which is able to attract interest in the pharmaceutical market, even in a critical phase like the current one. We are particularly pleased because alliances like this one represent, for a not for profit organisation like ours, an actual opportunity to fulfill the promise made to patients and donors who have been supporting us; translating the excellent results of scientific research into therapies that are accessible to people suffering from genetic diseases still lacking a cure. Those are the people for whom Telethon was born and still exists and keeps working," said Francesca Pasinelli, General Manager of Fondazione Telethon.

Ultragenyx In-Licenses Therapeutic Program for Rare Genetic Disease Mucopolysaccharidosis Type VII from St Louis University: MPS VII Patients may at last have an opportunity for Enzyme Replacement Therapy

Novato, CA – January 5 2012 – Ultragenyx Pharmaceutical Inc., a biotechnology company focused on developing treatments for rare and ultra-rare genetic disorders, today announced it has in-licensed an enzyme replacement therapy program from St. Louis University to treat MPS Type VII. The in-licensed programme is a treatment for an ultra-rare genetic, metabolic disorder that results from the deficiency of the beta –glucuronidase (GUS) enzyme.

Also known as Sly syndrome, the disorder was first identified in 1973 by William S. Sly, MD, a world-renowned researcher in inherited diseases, who is currently Professor and Chairman Emeritus, Department of Biochemistry and Molecular Biology, at St Louis University School of Medicine. Dr Sly will collaborate with Ultragenyx on the MPS VII development program.

"We are pleased to have the opportunity to develop this treatment for MPS VII which has been in the research stage for a long time and has yet to be made available to patients. We look forward to working in collaboration with Dr Sly and the MPS community on this program," said Emil Kakkis, MD, Chief Executive Officer of Ultragenyx.

Dr Sly noted, "After so many years of research by my laboratory and my research colleagues, I am pleased to finally have the chance to see if MPS VII patients can be successfully treated with enzyme replacement therapy. I have confidence in Ultragenyx's ability to advance the MPS VII program through the development process and fulfil our shared goal of bringing this potentially life-changing therapy to patients. We look forward to working closely with the Ultragenyx team on this program.

Transforming good science into great medicine for rare genetic disease. www.ultragenyx.com

2nd European Fabry Expert Lounge Meeting

This meeting held on 12-13th October 2012 in Munich, Germany, was hosted by Genzyme Europe and was attended by 110 Fabry experts and three representatives of the Fabry International Network (FIN); Nawel Van Lin, Global Development Officer; Lut De Baere, FIN Director and Christine Lavery, FIN Director and Chief Executive of the MPS Society.

Following an early buffet lunch the meeting was addressed by Dr David Meeker, CEO of Genzyme. The first session featured three presentations relating to the Neurology of Fabry Disease. I was particularly interested in Professor Hilz talk on 'Autonomic verve system involvement – possible implications for morality? It would appear that further investigation of the autonomic dysfunction may be indicated. Certainly as we put the MPS Fabry Conference programme together for June 28th - 30th 2013 at the Coventry Hilton this is a subject you may find of interest.

The next session explored the risks and benefits of adjunctive therapies, in particular the question 'should statins be used in all patients with Fabry Disease and the role of Cardiac Arrhymia Devices (CAD)? The key messages were that dosage of Statins is very important and that the insertion of a CAD is not without considerable risk.

During session three chaired by Professor Robert Desnick we learnt about Mutations and Polymorphisms in Fabry disease. Professor Desnick demonstrated most graphically how usually polymorphisms are not disease causing in Fabry disease although they may have health impacts in the future. The last session of the first day chaired by Dr Stephen Waldek addressed Individualising the ERT dose in Fabry disease. Dr Waldek spoke on registry event data in female patients. This included the first renal event; first cardiac event; stroke and non-cardiac death. 17% of women reported a clinical event prior to ERT and 13% reported events during treatment. Out of 58 events during treatment there were 14 Stroke, 32 Cardiac, 11 Renal and 6 Non Cardiac events included malignancy and dialysis complications.

Dr Camilla Tondel spoke on clinical trials in children and ERT in children and spoke of a low incidence of complications in kidney biopsy. In Fabry disease the glomeruli are white normal glomeruli are red. A five year study of 12 young Fabry patients with baseline biopsy and annual biopsy, found correlation in dose and clearance of GL3 in Fabrazyme. Dr Tondel concluded that kidney function stabilised and endothelial cells cleared after long term ERT in young Fabry patients. She further concluded that kidney biopsies are in important and state of the art in the treatment of young abry patients. Professor Warnock finished the session by speaking on ERT dosing and progression of Fabry nephropathy in adults and described the FAACET study...

FAACET Study (The Fabrazyme and Arbs and ACE Inhibiter Treatment)

The purpose of this study is to prove the hypothesis that titration of the ACE inhibiter and Angiotensin Receptor Blockers (ARB's) reduce urine protein excretion to less than 500mg per day in Fabry patients receiving Fabrazyme ERT at 1mg/kg every two weeks and slow the progression rate of decline of glomerular Filtration rate (GFR) compared to case controls drawn from the Genzyme-sponsored Phase III extension study or the Phase IV study.

He spoke about .03mg/kg for Fabrazyme stabilising patients with clearance in the kidney taking place after some years.

After a working dinner and a welcome opportunity to meet up with some of the UK LSD laboratory scientists it was a not so early night ready for day 2 of the Expert Lounge.

The first session was the 'Laboratory in Fabry Disease'. This was chaired by Professor Simon Heales, Institute of Child Health, London. Dr Heather Church, Willink Laboratory, Manchester spoke about the Willink's dry blood spot screening programme and data from women being tested for Fabry disease. Dr Derralynn Hughes, Royal Free Hospital, London described her work looking at disease causing mutations and their utility in diagnostic laboratories. The strength of the UK contribution to the science of Fabry disease was concluded with a talk on 21 Biomarkers discovered for Fabry disease and how to turn them into diagnostic tests.

This next session focussed on Fibrosis and the clinical impact for the prognosis of the patient and it was agreed that better understanding of the pathology of Fibrosis and improved imaging is needed.

After lunch, attention turned to new data and late breaking data. Dr Uma Ramaswami, Manchester Children's Hospital, spoke on the objective assessment in paediatric patients. Dr Ramaswami described early cardiac changes in children with classic and cardiac variant of Fabry in children concluding there needs to be regular follow-up in children to ensure a full understanding of the progression of Fabry diseases and treatment options. Dr Partick Deegan presented data on the Lysosomal delivery of therapeutic enzymes in cell models of Fabry disease. 95% of enzyme is taken up by the liver which is not affected in Fabry disease. **Christine Lavery**
c.lavery@mpssociety.co.uk

Genzyme EU Patient Group Forum: Market Access for Rare Diseases

I was honoured to be invited to be a member of a forum, held on 26 November 2012 in Amsterdam, to examine Market access for rare diseases in Europe organised by Genzyme. I was joined by two members of the LSD UK Patient Collaborative, Tanya Collin-Histed from the Gaucher Association and Toni Mathieson from The Niemann-Pick Association for this one day meeting in Amsterdam where we were also joined by Nawel Van Lin of Fabry International Network (FIN) and delegates from Portugal, Romania, Netherlands, Bulgaria and Spain.

The meeting started with two presentations on Health Technology Assessment (HTA). One, looking at changes in Healthcare and reimbursement landscapes in Europe and the impact on patients with rare diseases. The second by Henry Featherstone was on the United Kingdom objective to introduce a value-based pricing (VBP) approach to pricing of branded pharmaceutical products. During this discussion the LSD UK Patient Collaborative representatively received confirmation that the LSD ring fenced budget post April 2013 will disappear and any efficiencies to be found going forward now into 2013 and beyond will go back into the Treasury pot. Clearly the UK LSD Patient Organisations have a lot more to do to change hearts and minds at the new NHS Commissioning Board to ensure any efficiencies found in the LSD Service are led back into service to enable new patients to receive reimbursed ERT.

The next session looked at evolving the role of patient groups in access now and in the future. First Ria Broekgaarden from the Pompe group in the Netherlands spoke of the dark summer of 2012 when the Dutch Government were advised not to reimburse the cost of ERT for Pompe and Fabry patents and the campaign by patients to reverse this decision. I then spoke on the role of the LSD Patient collaborative in the UK and its work to influence government in the light of the disbanding of AGNSS as of April 2013.

Other subjects discussed included examples of on-going HTA projects in Europe and European initiatives on rare diseases and orphan medicinal products from an industry perspective.

Finally Genzyme talked of its charitable access programme including the role of the Medical Advisory Board. ICAP enables over 280 patients around the world to receive treatment. INCAP in India is for all Genzyme products (80 patients) and CHINACAP is only for gaucher disease. The key to success is understanding the local healthcare system, partnerships with clinicians, involvement of patient organistation and dialogue with authorities and government. Today Genzyme have 600 patients on the charitable access programme and 500 more have transferred to sustainable treatment access.

Tanya Collin-Histed, to conclude the day spoke on the European Gaucher Alliance global pathway for humanitarian aid. Her take home message was, 'it is hard, we can't save the world, but I hope we can make a difference!' **Christine Lavery**
c.lavery@mpssociety.co.uk

Kirsten Harkins receives the Queen's Diamond Jubilee Medal

The Queen's Diamond Jubilee Medal is a commemorative medal created in 2011 to mark the 60th anniversary of the accession to the throne of Queen Elizabeth II. Commonwealth citizens alive on 6 February 2012 were eligible to receive the award which recognises honourable service in military, police, prison, and emergency forces, or for outstanding achievement or public service.

Kirsten Harkins has shown the most exemplary leadership over the last fifteen years in the support of children and adults across Canada with an MPS or related Lysosomal Storage Diseases and their families and carers. Kirsten is Executive Director of the Canadian MPS Society and the mother to Niklas who has MPS I.

Well Done Kirsten, no one could be more deserving of this award.

A fresh new logo for the Fabry International Network

Fabry International Network is delighted to announce the launch of its new logo which reflects its continuous commitment to its Fabry Patient Organisations around the world, professionalism and growth.

Fabry International Network (FIN) is the only global organisation that connects all parties with an interest in Fabry disease with a purpose of collaborating, communicating and promoting best practices to support those affected by the disease.

The stakeholders are the Fabry Patient Organisations around the world; Industry Partners like GSK, Genzyme, Shire and Amicus Therapeutics; The Medical Advisory Board and any organisation that is linked to Fabry disease. Through this unique partnership, FIN is able to look after the interests of over 10,000 Fabry patients worldwide.

Today, FIN has a network of 40 Fabry Patient Organisations in 30 countries. FIN was established in 2005 with a vision of a 'world where every person affected by Fabry disease has the best quality of life possible through early diagnosis, treatment and cure'.

Why would a charity like FIN need an updated, new logo? All around us successful non-profit organisations are run with attention to detail and professionalism. Their dedication to their cause is taken seriously. As such, even charities need to work on their public relations and create a positive image that is contemporary and represented by a logo that reflects its cause and core values. Here are some recognisable logo designs for other charities.

UNICEF was created to provide emergency supplies such as food and water to children who are affected by war and natural disasters. It is only appropriate that this creative logo design features the United Nations logo with an adult holding up a small child. As this is a United Nations program in which adults support children through difficult situations, the logo is more than appropriate. It is recognised around the world and will continue to be a force in children's rights in the foreseeable future.

The World Wide Fund for Nature, formerly the World Wildlife Fund, is a conservation-oriented charity. It features, simply, a stylized and easily recognizable panda. Pandas are a nearly extinct animal that has been rescued by the WWF and other concerned organisations; there could be no better mascot for this creative logo design.

This international children's charity focuses on improving the lives of children worldwide through a variety of means, including by providing education and healthcare to many young people who otherwise would have no access. The red colour seeks attention and urgency, while the round shape makes people feel included. The small child shape in the centre of the circle holds out its arms, reaching out to prospective donors. The message of this creative logo design is that these children need help immediately, and that you can be included in the effort.

FIN's new logo is made up of three hands coming together to create a strong unit into a caring and protective cup-shape. This intends to portray the collaboration of the Organisations' key Fabry stakeholders with a common purpose of finding a cure for Fabry disease.

The three interlinking hands represent the bringing together of a Fabry patient, Industry Partners & Medical Advisors and FIN. The word FABRY has been highlighted as this is what the organisation is all about.

The logo is a small part of the communications strategy that FIN Directors have been developing over the last twelve months.
May I take this opportunity to invite you to contact FIN if you wish to:

- Connect with the Fabry community: Please use the Patient Organisations' links on our website www.fabrynetwork.org should you wish to connect with other Associations.
- Communicate your latest news: We publish a quarterly e-newsletter that is sent to all our members and partners.
- Connect with our Industry Partners: We will communicate to all our members the latest news from the Pharmaceutical Industry.
- Attend teleconferences and face-to-face meetings: With FIN Board of Directors, Industry Partners and the Medical Advisory Board. FIN will be hosting a membership meeting in Frankfurt on 26-28th April 2013. For more information, contact Nawel Van Lin on +44 1494 766 633.

The Fabry International Network is governed by a group of trustees: Megan Fookes (Australia), Jack Johnson (USA), Lut De Baere (Belgium), Anna Meriluoto (Finland) and Christine Lavery (UK). The FIN Office is based at MPS House, Amersham, Buckinghamshire. We hope that you will like the new changes and welcome your feedback by emailing Nawel on n.vanlin@mpssociety.co.uk.
Nawel Van Lin, Global Development Officer, In post since July 2012

Fundraising for the MPS Society with **Wicked Genes**

Welcome to a new year and what a year it's going to be!

Fundraising

In the next few pages we want to share your wonderful fundraising stories, whether you have been baking cakes, running or fundraising at work you are the reason the MPS Society can keep supporting you and keep funding vital research for treatments. Without your efforts the only Charity for MPS sufferers and their families in the UK simply wouldn't be here…which is why we need you to keep doing what you do best…. and we want to help.

What's New?

Look out shortly for our updated Fundraising packs and website. We appreciate if you are trying to get people to fundraise you need it to be made as easy as possible, so we have listened to you and reacted.

As Wicked Genes is now THE fundraising arm of the MPS Society we will have just one MPS website where you can get all the information you need (No more having to flick between two sites!) You can find out how the charity can support you, where clinics are, what trials are running and in the same place you can get all the Wicked fundraising information you need. Follow us on Twitter and Facebook and get up to the minute information about benefits, treatments and trials as well as finding out about fundraising across the country.

An MPS Awareness Event you say!

We love to get your stories and pictures from your events, so much so we have decided to have a go at running our own for you to join in. Lots of charities across the UK run coffee mornings, runs, cycles and we thought it be great to run an MPS event to coincide with MPS Awareness Day in May. We are just ironing out the details so stay tuned and we will be online and on twitter to let you know what the plan is. If you have got any ideas, or want to help we would love to hear from you.

E: wickedgenes@mpssociety.co.uk
T: 0845 389 9901

‘Oh, What a night!’

During the year the Murphy family decided it was time that we put our heads together to organise a fundraising event in aid of the MPS Society. After much debate, we decided upon a night for family and friends to gather together and what a night it turned out to be!

In order to gather prizes for the evening, we wrote to local businesses for donations and together with the huge generosity from friends and family members we were able to accumulate a fantastic array of prizes ranging from a 48” TV to a signed Liverpool football. In addition to this, the Sartan Club provided the venue free of charge and a wonderful singer and magician provided the evening’s entertainment also free of charge which enabled us to keep out outgoings to the minimum.

The evening was a total sell out with special appearances from two lovely MPS families. Jackie and Jordan Mount flew down from Glasgow while Rhian, Roswyn and Sarah McKnight traveled from Wales to be with us for the evening. As you can see, Jordan had a wonderful time showing off his groovy dance moves while Sarah knitted three scarves that went to auction. Rhian (Sarah’s Mum) obviously showed that she had missed her vocation in life as she took to the stage to auction Sarah’s scarves. She was superb at cajoling people to bid and raised £40 per scarf, a total of £120. She then went on to auction a signed Liverpool football which was bought by a neighbour, who kindly gave it to Tara as he knew she was an avid Liverpool supporter. I must say that if anyone is thinking of doing an auction in the future, I would recommend that you invite Rhian, you won’t be disappointed!

All in all, the evening was a huge success and I would like to thank everyone involved. I would like to say a special thank you to my sister Cath and her husband Mark who helped us tremendously. Mark’s skills on the microphone during the raffle prizes was unmissable! I would also like to thank the MPS Society for all the help and support provided over the years and we are pleased to announce that we raised a grand total of £3270.

A wonderful way to raise money, a brilliant night in the company of fantastic people. Thank you!
Maria, Ivan, Kate & Tara Murphy

Donation from Kevin and Margaret Boyle

We were delighted to receive donations totalling £800 from Kevin and Margaret Boyle. The couple recently celebrated their Golden Wedding Anniversary by hosting a party for friends and family at Arnos Court Manor, Bristol. Some of the guests travelled from Ireland to be with them. In lieu of presents, they requested donations for the MPS Society.

Kevin and Margaret are regular customers of 'Marina and Friends' Charity Shop in Sandy Park. Over the years they have been touched by Marina Foster's tireless fundraising, and wanted to make a contribution to her work. Therefore, the donation is for research into Sanfilippo disease. Here is a photo of the lovely couple. **Chris and Julie Kembrey**

Simply Baked

I first saw Tillie-Mae's story from a friend and I really wanted to help! I have a nearly 3 year old daughter myself, and I just thought if that was my daughter I would want anyone and everyone to help her. I have previously seen other crafty friends do various sale nights and market nights through their Facebook business pages, so I thought that would be a fantastic way to raise some money.
I appealed to any cakers, crafters and businesses for donations to the market night. The idea was, people would bid on the items, the highest bidder would win the item, and all funds would go to the Genistein trial. Over 4 days we managed to raise a fantastic £1,000 on Tillie Mae's ShareAGift page!

Bidders were super generous, often bidding more than the item was worth, and donating more than they bid anyway.

Support from the Draynes

Keiran Drayne donated £450 as a contribution to the work of the MPS Society. He writes: 'I am aware of the MPS Society's work as my neice, Roma Drayne, has Morquio. Roma is now 19 years old and has coped with many adversities in her life, through her positive attitude and wit has been a constant inspiration. Her Mum and Dad, Bernie and Kevin are equally inspirational and their 'just get on with it' attitude has also been uplifting. We raised the donation through a group of us; Kevin, Bernie, Dinah, Damien, Eamonn, Angela, James, Philomena and myself attending a Kevin Bridges gig in the Belfast Odyssey Arena and making a donation. This was a superb night of comedy and we all enjoyed the insightful wit of this Glaswegian comic.' **Kieran Drayne**

Athena Network supports MPS

As a result of meeting the very passionate Laura Burrows, I felt compelled to help the MPS Society after hearing more about the wonderful children this charity helps. As a mother myself, I always try and put myself in the situation of others and as I am able and willing then I will do all I can.

I belong to the Athena Networking group for women, a remarkable network of women all supporting each other at the core of how we work. We had our regional area Christmas lunch on Tuesday 4th December and Jacqueline Rogers, the CEO of this amazing network, asked for a local charity that our Christmas raffle could benefit from. I immediately thought of Laura at the MPS Society. We raised £150 and all our prizes were generously donated by members from the network.

Thank you Laura for bringing to our attention this wonderful charity and we will continue to do what we can to help your very special children.
Vanessa Stottor, Mary Kay Cosmetics and Skincare

Thank you to Vanessa's father, Michael Moore for his kind donation of £250 to the MPS Society.

Sandra Silcock donated £30 to Sanfilippo research being the proceeds from her MPS piggy bank.

Marina Foster has raised a cumulative total of £91,817.80 from the sale of second hand goods in her charity shop.

Susan Swayne donated £50 in her MPS collection box.

Steve Cotterell did a school talk at **St Patrick's RC Primary School** and MPS received their donation in lieu of £250.

J Langford Stacey donated £50 being the proceeds from the sale of greetings cards.

Rick and Ann Coleman held a fundraising garden party on Bank Holiday Monday and donated £100 of the proceeds to the MPS Society.

We would like to thank the following donors who kindly donated to MPS for the marriage of Emma Herod and Damion Westland: Steven and Alison Hedges, Lesley Seymour, Daniel Knight, Kris Newstead, Graeme and Nicola Fish; Mr and Mrs A Graley, Mrs C Oakley, Debbie Winfield

Ann Parson has donated £105 being the proceeds from the sale of MPS trolley key rings.

Sandra Bates donated £70 being the proceeds from the raffle of an e-book at Randalstown Medical Practice.

Brenda Weston donated £235 being the proceeds of a raffle and donations at Greenfield Methodist Church.

Alison and Doug Gunary donated £500 in lieu of 55th wedding anniversary presents. This donation is in memory of Paul Gunary who had MPS II, Hunter.

Students on the vocational skills course at **Wiltshire College** held a sponsored walk and donated £64 in sponsorship monies raised.

The **Greggs Foundation** NE regional charity committee awarded £500 to MPS following Jodie McNally's parachute jump on behalf of the MPS Society.

MPS received a donation from the Medic Society at **Dr Challoners Grammar School** in the name of Christine Lavery. Christine gave a lecture to the young medical students and requested her fee be paid to the MPS Society instead.

Dorothy Robinson (nanny to Hannah Shannon MPS III) donated £170 in respect of carpentry and card-making projects undertaken by her husband Colin and herself.

Kevin Baker's niece has Hurlers and he has raised £35 at work for the MPS Society.

NATS IS continue to support the Society and have donated £500 to MPS.

St Ivo Lodge No. 2684 held a ladies night and raffle and raised £1252.34 for the MPS Society.

Jenny Quant donated £55 to the MPS Society being donations made by family when buying Christmas cards.

Louise Shaw donated £20 in lieu of Christmas cards because of a friend of her being recently diagnosed with MPS.

Dorothy Robinson, nanny to Hannah Shannon (MPS III) recently held a coffee morning and sale raising £275 for MPS.

The **Sudbury Methodist Church Badminton Club** kindly donated £27.50 to MPS.

Kathy Wilton and Dave Weston at HSBC Bank plc in Cheltenham sent in used stamps, cartridges and phones in memory of Ben Wilton. They are hoping to get this match funded by HSBC.

The **Norton Rose Charitable Foundation** kindly donated £5000 to the Genistein Appeal with reference to one of their colleagues, Liz Gill, whose son Bobby has Sanfilippo.

Air Salvage International kindly donated £50 in lieu of sending Christmas cards to their customers.

Zoe Parham sent in a CAF voucher for £250.

Keith and Shirley Bown sent in a cheque of £25 donated by **Auntie Peggy** in memory of Shirley's sister, Linda, who sadly passed away this year.

Joan Crespin donated £25 to MPS towards research into Sanfilippo.

The **Caerdydd Lodge of Freemasons** have kindly donated £1500 as a result of support given to the family of Mr and Mrs Royston Plummer, and in particular, in memory of their grandson, James Bernard Edwards.

The **Gill family** have kindly donation £20 in memory of Ben Illingworth.

Pam and Ken Ballard wrote to MPS enclosing a cheque for £86.61 being the proceeds from the exchange of foreign coins.

Karen and Andrew Weedall raised £43 through sales from the Christmas Webb Ivory catalogue.

Members of the **Bowen Lodge of Beaconsfield** kindly donated £100 to the MPS Society.

The **Thursday morning riding for disabled helpers** donated £37.50 in memory of Ben Illingworth.

Peter Levy OBE kindly donated £250 following a pleasant day out at St George's Hill Golf Course via Heather Gordon.

Mr Philip Noble of St Thomas More Chambers donated £500 to the Genistein appeal after reading the article about Bobby Gill in the London Evening Standard and Roll On Friday.

Donna Bown held a table top sale at Asda Eastbourne which raised £25. Donna has now left Asda but Asda have kindly said they would continue supporting MPS through the sale of trolley key rings and collection of used stamps.

Claire Knox of Pro-Co raised £90 for a company dress down day.

Paul Kelly has raised £810 on his justgiving page, www.justgiving.com/paul-kelly12, Paul's 5 for 50.

The Lions Club of Nailsea kindly donated £409 to the MPS Society and the Pearson family donated £100, both of which were the result of an annual tea party in support of MPS held by Ian Pearson, grandfather of Archie.

The Members of Stayley Lodge No. 3370 kindly donated £500 to the MPS Society. This money is to be put towards research into Fabry disease following a specific request from Joseph and Pauline Green.

The students of **Farringdon Community Sports College** raised £250 from a non-uniform day held in July. The Assistant Headteacher is Derek Wright who has Fabry.

Jennifer Barclay donated £200 online being the proceeds of wedding favours in the name of Aiden Brown.

Ian Evans raised £70 through the sale of MPS lanyards at his place of work, Arriva Shires.

Norton Rose LLP kindly donated £1003.67 to the MPS Society's Genistein Appeal.

The **Celtic Charity Fund** donated £1000 for the Genistein trial helping children like Jack Baird.

Linda Allen donated £60 online being the proceeds of fundraising for the MPS Society.

Iris Hitter sent in donations totalling £176.66 being the proceeds from a quiz night and loose change collected by Iris's daughter, a dinner lady at a high school.

Damion and Emma Westland were married on 20th October 2012 and instead of gifts they asked for donations to the MPS Society as Damion's brother, Darryll, was diagnosed with MPS in the early 1970's when he was very young. £820 was received in cheques and MPS also received a number of online donations.

Anne Cotton and Heather Gordon organised a golf day at St George's Hill Golf Club in Weybridge raising a wonderful £7,536.19 for MPS.

Jenny and Andy Hardy recently held an MPS Christmas card sale. They wrote to tell us: "We sold all our cards, wrapping paper etc at our recent sale. We had a steady flow of people during the morning, the afternoon picked up and our house was full of friends by the evening. It's strange how the pull of mulled wine seems to be stronger than that of tea or coffee! On top of selling goods to the value of £448.15 we also received donations of £230.72."

Lisa Burr, Contract Manager at **ISS Facility Services**, Newcastle Site, recently wrote to tell us: "As part of our local corporate social responsibility commitment we recently held a coffee morning to raise money for charity. I have over 150 staff on the site and we all got together to have a little break from our busy cleaning schedules at the college, we had a cake and coffee sale and raised £60.00. My staff chose to donate to the MPS Society because we believe what you do is outstanding and we want to show that we really appreciate it. Please keep up the good work and we are proud to help and support."

Sue Jenkins writes: 'Earlier in the year I decided to help raise funds for the MPS Society so I approached our local Resident's Association fete committee who kindly agreed to let me have a stall free of charge at this year's fete. Sadly, due to bad weather conditions and flooding of the village green, the fete was cancelled for the first time in over 40 years. Kindly, the local community market offered me a free stall at two of their meetings so I'm pleased to be able to donate the proceeds from sales of £141.10 and a further £10 in merchandise sales. I would like to take this opportunity to thank everybody who donated goods for me to sell, especially oypla.com, ticracing.com, Elliots Engineering and also the Bar Hill Residents Assocation and Bar Hill Community Market.'

The MPS Society recently received a cheque for £1070 towards the Genistein Trial from **David Cook**. David writes: "I work for a company called UKAR in the North East and Jack (who was one of the faces of the MPS Society's Genistein Appeal) is local to where we are based and relative of a family friend. I organised a charity football match to raise funds. The generosity and time from all the players and co-workers who have made this happen has been invaluable. UKAR is a company that actively promotes charity work and they have also stated that they will match the funds up to £1000 so a total of £2070 will be donated. It has been a pleasure to help such a worthwhile cause and we hope that all of the children affected by this terrible disease receive all the help they need and hopefully this can help contribute to that cause.

Congratulations to **Ian Evans** who has been awarded the Gold Award from the Arriva Community Action Awards in recognition of his valuable contribution to Society. It is wonderful that Ian's fundraising and support of the MPS Society which included organising for Santa to drive a bus round Aylesbury this Christmas (Who needs a sleigh!) has been recognised in this way.

Chess Telecom, based in Alderley Edge, (www.chesstelecom.com) support 2 regular charities – Prince's Trust and East Cheshire Hospice. Last December staff were asked to make welcome calls to businesses of a new acquisition by Chess. At the end of each call the option of 3 charities was given to each business where they were to choose 1 and then Chess would make a £1 donation to that charity. This is where the 3rd Charity to support was the MPS Society after Nev had made the recommendation. My husband Nev was given the cheque this week for £443 which is the money raised from last December. We were really impressed as not only is it a good sum of money for yourselves but it also means that 443 additional businesses have now heard of the MPS Society which is another great start in promoting awareness. **Gayle and Nev Bradshaw, parents of Emily (MPS I)**

Joanne Haines and her daughter Jodie chopped off their ponytails for MPS in memory of Lewis Cato and raised £130 on their justgiving.com page

Denise Main has donated £25 to Morquio research.

70 Mile Sponsored Cycle from Bramley to Shoreham (and back!)

Early this year, I persuaded six school friends to join me in a 100 km cycle from Bramley in Surrey to the coast at Shoreham – and back – to raise money for the MPS Society. After weeks of fundraising and a training run, the big day arrived.

We set off at 9am on a clear morning on Thursday 12th April. The wind was cold on the face at this time but by 9:30 we had the sun shining on us, the temperature was rising, and the kilometres were flying past.
We reached Shoreham in 4 hours 25mins, no problem.

After a quick lunch, we set off again on the return straight. Another 52km path stretched out in front of us… but we had no idea how much longer it would seem. We were crossing the main road 3 miles out of Shoreham when it happened, the unmissable sound of a tyre bursting. The second burst tyre for Ben came 4 miles later and although we fixed it, the rear wheel had to be pumped every half hour, slowing us down further.

Then during one of our pumping stops, we managed to break the valve of Ben's rear tyre so it no longer filled with air. Ben and Damon carried it along the path, while I headed off to meet Will (Ben's brother), who had agreed to drop off another bike.

We were on our way again after a long delay, until my pedal sheared off as we entered Cranleigh, and we needed another replacement bike to get us all home. As soon as it turned up we knew it would be a non-stop dash home in a hail and torrential rain storm. And so we finished at 20:10 that evening (11 hours after setting off), chilled to the bone and with mud coating every part of our body.

It was a day that I will never forget. We achieved what we set out to do, which makes me feel really good inside because, honestly, I didn't think that I would make it. However, had this challenge have been too easy for me then I don't think we would've had the sponsorship response that we did get. In total we have managed to raise in excess of £3700!
Louis Garthwaite (Hunter)

From top left, left to right: Rob Platt, Will Harrison, Andy Day, Louis Garthwaite, Tom Campbell, Ben Moore, Damon Elson

Coventry half marathon in memory of Daniel

Gurchetan Shergill ran the Coventry half marathon on 14 October 2012 and raised £535 for the MPS Society on his justgiving page. Here he writes: Daniel Singh has been a dear friend to me for over 15 years and has been the GGNP FC mascot, performing the role with distinction, during this time. Daniel's parents, Rashpal Singh and Sandra as well as being family friends, have proven to be a true inspiration to us all with their dedication and support for Daniel. Rash has also been our football coach, ever present with his trusty right hand man Daniel come rain or shine.

Daniel suffered from a rare illness called Hunters disease (MPS II), it affects 1 in 150,000 live births. Over a ten year period 39 babies with Hunters disease were born in the UK. MPS II is even rarer in girls, as boys are the main suffers. Children with MPS II are missing an enzyme called Iduronate Sulphate Sulphatase, which helps the breakdown of the sugar molecule cells stored in the body. Due to the missing enzyme the body fails to breakdown sugar molecules, which therefore remain stored in the body causing progressive cell damage. There is currently no cure for MPS II and it is a terminal illness.

Daniels football ability was beyond that of similar MPS II sufferers, not to mention beyond that of most of the first team. He was mostly seen at the senior team games but did make special apperances at youth team games looking out for the new Messi. All players, members and supporters across GGNP FC aknowledge Daniel's unreserved support for the club and he is an inspiration to us all. Daniel is an honorary member of GGNP FC.

Daniel unfortunately passed away in December 2011.

Jump out of a plane for MPS!

Parachute for Free!

If you have ever wanted to do a parachute jump here is your chance. The MPS Society is looking for adventurous volunteers to make a fundraising parachute jump and if you raise enough sponsorship you will get to jump for free!

There are three types of jump available - an accelerated freefall where you can experience the thrill of skydiving solo from up to 12,000 feet, a tandem skydive from 10,000 feet attached to a professional instructor and a static line jump which is performed solo from up to 3,000 feet - and you can jump from any one of over 20 British Parachute Association approved airfields across the UK.

No experience is necessary as all training is given and if you raise from £360 (depending on the type of jump you choose) you will receive your jump for free.

For a full information pack and everything you need to take part in the experience of a life time, please email **fundraising@mpssociety.co.uk** or phone the MPS office on **0845 389 9901.**

Gillian McCann and her sister Gemma did a sponsored skydive for MPS. They raised about £700 in memory of their nephew Jack Malcolm, born 24 April 2007. Jack was diagnosed with MPS I Hurler and after many months in hospital and a bone marrow transplant he lost his fight on 25 November 2008 aged 19 months.

Kim Morgan did a tandem skydive and raised £280.30 in sponsorship money and £120 on her Justgiving page.

Steven Scott raised £422 on his justgiving page from a sponsored skydive.

Leigh Jones has sent in £100 in sponsorship money raised from a sponsored skydive on 27 October 2012.

Chris Brown did a sponosored skydive for MPS. He writes on his Justgiving page: 'The MPS Society helps look after children with diseases like my sister Claire had and my family has always supported this charity so I thought it was only right to try and raise as much money as I possibly can by doing this skydive to try to repay the kindness they have shown to my family and all the rest of the children they look out for!' Chris raised £640 on his justgiving.com page.

Jodie's skydive for MPS

Jodie McNally did a tandem skydive on behalf of Daniel Muers who has MPS II Hunter. Jodie's aunt is Elaine, Dan's mum. She writes: "We went to watch the dive, we all arrived at 8am, luckily it was a nice day. The girls were nervous but ready to do it. Unfortunately there were alot of people diving and it was 4.30pm before they had their turn! So, I think the nerves were well going after such a long wait but Jodie said that what Dan goes through, he is so brave, so they would be too! All went well and we are very proud of the money raised which was about £1500 with half going to the MPS Society and half going to St Oswald's hospice. Big thanks to Jodie and her friend Emma Champion and of course the MPS Society and the hospice for all the help we receive with Dan'. **Elaine Quin**

Jodie works for Greggs and the Greggs Foundation kindly agreed to match Jodie's efforts to the amount of £500.

Find us on Facebook

Sponsored walk for MPS

We decided we wanted to do a good deed and help out the MPS Society. On Saturday 9th November we did a sponsored walk from Bexleyheath to Blackheath.

We set ourselves a time limit of a hour and a half. We managed to complete the walk in 1 hour 28 minutes and 4 seconds. We managed to raise £174.70. We had lots of fun doing this event and we will certainly do more in the future. **Danielle Wyatt and Linda Woods**

We have places on the following Great Run events: Great Manchester Run - 26 May 2013, Great North Run - 15 September 2013, Great Birmingham Run - 20 October 2013, Great South Run - 26/27 October 2013. For more information about the run events please visit www.greatrun.org. **To register your interest for one of our MPS charity places please email fundraising@mpssociety.co.uk.**

Liz Stillo in the Great North Run raising £185 in sponsorship money on her justgiving.com page, www.justgiving.com/lilz-stillo

Rebecca Brogden did a sponsored skydive for the MPS Society raising £120 on her justgiving page, www.justgiving.com/rebecca-brogden

Iain Gibson raised £164 on his justgiving.com page, www.justgiving.com/iain-gibson0 by doing a sponsored cycle Lands End to John O'Groats

Luke Holmes took part in the Great North Run 2012 and raised £330 on his justgiving.com page, www.justgiving.com/leholmes

Sarah Cohen took part in the Great North Run 2012 and raised £335 on her justgiving.com page, www.justgiving.com/sarah-m-cohen

John Hills took part in the Perkins Great Eastern Run 2012 and raised £226 on his justgiving.com page, www.justgiving.com/john-hills

David George ran the Glasgow Half Marathon on the 2nd of September 2012. "I want to try and raise as much money as possible to help out my friend's daughter Chiara Peterson Fox who was born with Hurlers". He raised £145 on his Justgiving.com page.

Sian Thorpe-Gibbard ran in the Cardiff Half Marathon on 14 October 2012 raising £292.50 on her justgiving.com page, www.justgiving.com/sian-thorpe-gibbard

Sarah Davies ran in the Cardiff Half Marathon on 14 October 2012 raising £325 on her justgiving.com page, www.justgiving.com/sarah-davies81

Will Newbury was planning to run in the New York Marathon and raised £1495 on his Justgiving page towards the Genistein trial. On his page he writes: 'Unfortunately the New York marathon isn't on this year due to Hurricane Sandy. As I am here, I'm going to run 26.2 miles round Central Park.'

MPS Trustee takes on Triathlon!

On the glorious sunny day of Saturday 22nd September one of our Trustees Jessica Reid and two friends took part in the London Virgin Olympic Distance Team Triathlon.

An Olympic distance Triathlon involves a 1,500 m swim, 40 km bike and 10km run. This event took part at the London Excel Centre which involved swimming in the Docklands and cycling around Canary Wharf.

There were over 14,000 participants in this event, which is the largest Triathlon in the world. Jessica did the (second) cycling leg and beat her personal best by 25 minutes, by completing the cycling in 1 hr 29 minutes. This had something to do with cycling and keeping up with David Hasselhoff (aka "The Hoff!").

The team completed the Triathlon in a respectable 2 hours 51 minutes. Jessica raised £685 (plus gift aid) for the MPS Society.

She is now considering either entering the Sprint Triathlon herself next year... or re-entering as a team and beating this year's time!

£1695 raised in memory of Jake

Debbie and Mark Burniston have raised a fantastic £1695.64 from a series of fundraising events including MPS Awareness celebrations, the Great Manchester Run, the Sherwood Pines 10K and the Great North Run in memory of Jake Marshall Corcoran.

Debbie wrote to tell us what they had done: 'On 6th May 2012, the tenth anniverary of losing Jake from Hurlers, we had an MPS Awareness Day. We had a fun run for the children and a 5k run for adults followed by a BBQ and lots of fun and games. The day was so successful we hope to make it an annual event. Throughout the summer we then held BBQs, raffles and quizzes.

Then in September Mark and Debbie completed the Great North Run for the fourth time. They also completed the Manchester 10K in May and the Sherwood Pines 10K in July which was particularly emotional as we then visited the Childhood Wood.

Thanks to our great friends, family and residents of Woodland Glade we raised a total of £1695.64 which we are very pleased about and hope this makes a difference to the families you support. Special thanks as always to our very special friends Helen and Anthony Corcoran (Jake's parents), Hamish Ogilvie and many others without their support the events would be impossible to hold.

The MPS Society also received a donation of £380 from Derek Borland who completed the Three Peaks in memory of Jake. Derek raised £700 on his justgiving page http://www.justgiving.com/derek-borland.
The photos here are of Debbie and Mark at the 10k in Sherwood Pines and Derek Borland during the Three Peaks.

JustTextGiving by vodafone

Donate by Text

The Society for Mucopolysaccharide Diseases supports individuals and their families affected by MPS and related diseases throughout the UK.

We rely on your generous support to enable us to provide a unique advocacy support and information service including access to clinics, conferences and events.

Donations can be made online at www.mpssociety.co.uk.

Or make a donation by text...

Text MPSS01 £2 / £5 / £10 TO 70070 to donate now!

Thank you!

Jessica Roberts raised money for MPS Society by doing the Great North Run on 16th September raising £375 on her justgiving page, www.justgiving.com/Jessica-Roberts-86

Jessica works for Barclays Bank and so she arranged for the funds to be matched which has been agreed. Total funds raised is over £700.

Weaver School PTA does the Welsh 3000's

The Weaver School PTA raised money for the MPS Society by undertaking The Welsh 3000's (15 peaks over 3,000 feet with 27 miles of walking including 13,000ft of ascent all in 24 hours! They were raising money for the school and the MPS Society. Hannah Brock, who has Maroteaux Lamy goes to Weaver School.

Two dads from Hannah's class - Ed Brown, Jon Griffiths and Rob Price (the head teacher's husband who happens to be a mountain leader!) and myself, tried back in June to undertake the challenge but the Great British Summer called that attempt off! On 1st September we tried again (Rob had prior commitments and it was the only date the rest of us could get together and with sponsorship money already in we thought we had better give it a go!). We completed the first section 'The Carnedds' in line with our route plan (7 peaks done) - all was good. We had an excellent lunch served by Rachel, Jo, Megan, Jacob and Taylor and we all set out on the second section over 'The Glyders' - unfortunately the weather turned against us and we had to retire after peak number 10. In all we have raised £635 for the MPS Society and £595 for school funds. **Lucy Brock**, mother of Hannah

10km Santa run for MPS Society at Greenwich Park on Sunday 9th December 2012

My lovable son Maurice was diagnosed on 9th march 2011 with MPS I, since then our family life has been a rollercoaster with great challenges that required lots of strength to be able to cope with everyday life.

We have been blessed from the support of Dr Vellodi , Nurse specialist Victoria and Niamh at GOSH and the precious help of MPS Society. Without them my son and my family would not have been able to celebrate a happy Christmas!

On the other hand, my colleague at work in response to help Maurice and other children and their families had the brilliant idea to support the MPS society and to do a charity run, to be more precise a SANTA RUN!

When my colleague Alex, propose me the initiative, I had embraced the idea with enthusiasm and when I introduced it to my family and friends the bravest ones decided to join us too! We were going to be only 2 people to participate but with my great surprise we became 10 at the end... to run for a good cause!

I would like to thank you the brave Santa's: Silvia Marsella (mum), Nikki Francis (Maurice's sister), Skye Francis (Maurice's sister), Lyndon Joseph (Dad), Marco Sambiagio (Our best Friend), Alex Eaves M&S colleague, Lorraine Nicolson M&S colleague, Corinne Howarth M&S colleague, Ian Moore M&S colleague, Henry Courtier M&S colleague

On the day all of us had to wear extremely big Santa costumes, trying to keep the beard and the hat in place whilst warming up/dancing to Gangman Style and during the 10km run.

It was hard to run in a particular hot day, but it turned out to be a great day out for my family after 5 months of isolation and for my colleagues was a good day out too!

Out team raised £800 for the MPS society for the Santa run! Well done and thanks you for your kind donations!

Furthermore, the M&S Cost and Capital Finance team has raised and donated 500 pounds to the MPS society during the RSG Charity Quiz Night on 28th November at the May Fair Hotel in London. Well done! **Silvia Marsella**

Once you have read this MPS Magazine, please pass it on to your family, friends and colleagues. Help us spread the word about MPS and related diseases and the work we do. www.mpssociety.co.uk

Great South Run for Anabelle

Here is a photo of Anbelle and friends and family who kindly took part in the Great South Run raising funds for the MPS Society.

Anabelle sadly passed away at Christmas and our thoughts are with the family. Thank you to everyone who took part in the Run.

Continued thanks to the NATS IS team where Anabelle's Aunt Sally works for their fundraising efforts. Kathleen Butler raised £100 in cake sales and Matie Thomas and Louise Kelly raised £270 from a pub quiz.

Thank you to everyone who took part in the Great South Run 2012...

Lee Shepherd ran in the Great South Run 2012 and raised £1480 in sponsorship money on his justgiving.com page, http://www.justgiving.com/LRShepherd

Andrew Heazelton ran in the Great South Run 2012 and raised £410 in sponsorship money on his Justgiving.com page, www.justgiving.com/andyheazelton

Paul Brown ran in the Great South Run 2012 and raised £208 in sponsorship money on his Justgiving.com page, www.justgiving.com/Paul-Brown14

Terrie Brown raised £180 on her justgiving page www.justgiving.com/terrie-brown in sponsorship for her run in the Great South Run 2012.

Kelly Mitchell ran in the Great South Run 2012 and raised £370 in sponsorship money on his Justgiving.com page, www.justgiving.com/kelly-mitchell3

Kate Evans and Paul Traill ran in the Great South Run 2012 and raised £190 in sponsorship monies on their justgiving page, www.justgiving.com/kateandpault

Marie Shepherd ran in the Great South Run 2012 and raised £267 in sponsorship money on his Justgiving.com page, www.justgiving.com/marie-shepherd

Beth Nichols took part in the Great South Run 2012 and raised £160 for the MPS Society.

Sarah Williams took part in the Great South Run in October and raised £260 on her justgiving.com page

CONGRATULATIONS TO THE GAELFORCE TEAM!

Mick Kirwan, Neil Mitchell, Philly O'Carroll, Barry Hicks, Conor Swendell and Stephen & Eamonn Tighe completed Gaelforce on August 18th 2012.

Gaelforce West is the largest one day adventure race of its kind in the world. It is a multi-sport adventure race involving cycling, running, hiking and kayaking. The course of approximately 67km stretches from the stunning Glassilaun beach to Westport. In total they ran for 21K, cycled 45.5K, Kayak for 1K and climb Croagh Patrick while we are at it! The Gaelforce team raised £2161.40 on their Justgiving page, www.justgiving.com/gaelforce

Corporate Giving

In these challenging times, the MPS Society is looking to increase the number of companies that we talk with to gain their financial support. We would really appreciate your help with this as a personal contact and introduction is the most effective to introducing our charity, the work we do and how we would benefit from a company's support.

Many companies have a really positive attitude to supporting charities that their employees are involved with. If you, a family member or a friend work for a company work for a company please see what you can do to help…

Corporate Responsibility

Fundraising at work is a great way to boost your employer's reputation and image with shareholders, suppliers and the community. Many companies now have Corporate and Social Responsibility Programmes and welcome the chance to work with charities as part of this.

Charity of the Year

Many companies have a charity of the year. Not only can it generative positive local and national PR opportunities attracting valuable media attention, it can boost staff morale, encourage team building, and gives you something to talk about with your customers, clients and partners!

The selection process, although different for each company, usually requires an employee to nominate their charity. Sometimes the nomination process is simply emailing the name of the charity (the Society for Mucopolysaccharide Diseases) to the Committee. However, if they require supporting information, please direct them to our website www.mpssociety.co.uk or contact fundraising@mpssociety.co.uk.

We can provide press releases, case studies and photos which companies can use to put up on their websites, publish in internal magazines etc to give ongoing exposure and advertising. We always write to say thank you and try to offer a local MPS representative to collect cheques where asked.

Events

Here are some fun ideas for getting your workplace involved...

- Dress up, or down, or choose a fancy dress theme - those who take part donate £1, those who don't pay £2 (as a penalty for not getting involved)
- Guess the baby competition - get everyone to bring in a photo of themselves from a particular decade, or under the age of 3 for example, and pay to guess who each photo is of. The person with the most matches wins
- Pack your lunch - save money by bringing in your lunch instead of buying it, and ask people to donate what they would have spent into the collection pot. Designate a week to do this and watch how the money builds up!
- Swear box - any container will do. Every time you or a colleague swears on a designated day means that a £1 penalty must be paid
- Use the stairs - have a forfeit box near the lift and ask people to make a donation every time they give in and the use the lift
- Abandon your car - get sponsored to walk or cycle to work
- Guess the number/weight - either fill a jar with an item and ask people to guess the quantity or display something and ask people to guess the weight
- No email day - get everyone walking and talking and impose a penalty fine for those who cheat
- Ask us for an MPS Collection Box and have this on display so you can collect up all that loose change
- Recycle old mobile phones, ink cartridges - ask us for a freepost envelope
- Send us used old stamps from incoming post - we can exchange these for money to MPS
- Plan a sponsored challenge such as a skydive, cycle ride, trek or run with a group of colleagues. Ask us for a fundraising pack or check our website for the latest opportunities www.mpssociety.co.uk

For more information about how you can get your workplace involved, why we value corporate support and how we use the monies raised please visit **www.wickedgenes.co.uk**. You can also download a fundraising pack and other useful materials.

Matched Giving

https://www.cafonline.org/giving-as-a-company/engaging-employees/matched-giving.aspx

Many larger companies now operate a matched funding scheme, whereby they will match all the funds raised by one of their employees up to a specified limit. So, if you raise £200, you may find that your company will match that money with another £200.

Payroll Giving

http://www.hmrc.gov.uk/businesses/giving/payroll_agencies.htm

Payroll giving is one of the easiest ways for employees to give regularly to the MPS Society – plus it's both flexible and tax efficient.

Employees choose how much they want to give each month, and the charities they want to give to. The donation then comes off their gross pay, before the tax man touches it.

Charities benefit from payroll giving because receiving regular donations means that they can make financial plans for their long-term future – knowing they can rely on the regular gifts of their payroll donors. Employees can give as much or as little as they like, and because the donation is taken from their pay before tax, it actually costs them less.

Payroll giving is completely flexible - you can change the amount you give or which charities you donate to at any time. If you want to stop making donations then simply contact your payroll department.

Ask your Payroll Department whether or not your company offers the scheme. If they don't, they can set up a Payroll Giving Scheme by signing up with an approved Payroll Giving agency as listed on HM Revenue and Customs.

Gifts in Kind

If appropriate, your company might be able to support us by providing gifts as donations in kind for our annual prize draw!

Further information

For further information or to discuss any of these opportunities further please contact **fundraising@mpssociety.co.uk** or phone the MPS office on **0845 389 9901**.

Project Sanfilippo

The concept of Project Sanfilippo was developed by Karen Robinson, mother of Oliver who has Sanfilippo. Project Sanfilippo raises vital funds for the MPS Society to put towards research into Sanfilippo Disease. Here is an update of what's been happening so far.

Skydive for Project Sanfilippo

In the Spring Lindsay Webbs' partner Marcus Cooper (a keen skydiver) won a long weekend in Seville sky diving. With encouragment from her friend Ali Evans(who used to work with Oliver at Primary School and still cuts his hair-a difficult task!) she decided to do her first ever skydive and to raise money and awareness for Project Sanfilippo at the same time. Lindsay raised £290. "It was an experience I would never want to repeat " she said afterwards. Lindsay also found out a few weeks later she was pregnant (the baby is due on Christmas Eve) so perhaps this was the youngest ever skydiving fundraiser?

Auction of Promises

On 15th September 2012 the 4th Annual Fundraising Auction of Promises was held, this year raising £3400 for the MPS Genistein Trial.

It was a warm September evening and the garden was full to bursting with 70 people. Many friends who could not be with us for the evning donated either promises or money. Two of which donated on-line, Sally Martin £230 plus £20 sent for raffle tickets and Tim Hall £300.

The Cadbury Sisters sang again for us and Andy Rowley once again did an amazing job of auctioneering. All made possible by help from lots of different friends who always support us.

Stu, Sam, Ollie and I would like to extend a HUGE and heartfelt thank you to everybody who attended, helped, contributed and donated prior to, during and after the event! **Karen Robinson**

Fundraising at St Bartholomew's School

On 27 September 2012 Karen Robinson received a cheque for £4,880.89 from Davis House at St Bartholomew's School on behalf of the MPS Society for the Sanfilippo Project.

Davis students have raised the money over the past year by organising cake sales, sponsored walks, bike rides, penalty shoot mornings, a disco, village market stalls, raffles, carol singing and donations. The Davis House Evening where students performed various different talents on the stage also raised money towards this total. Davis House will continue to raise money for the MPS Society (Sanfilippo Project) over the next academic year.

The Cirencester Scope Committee kindly donated £500 towards the Project Sanfilippo.

Nicola Stearn and Diana Dee ran in the Cirencester Park 10k race raising £143.10 for Project Sanfilippo.

Jump for Genistein

Karen Robinson and others are doing a sponsored skydive, Jump for Genistein in February raising funds for the Genistein Appeal. If you would like to do something similar and for more information please contact **fundraising@mpssociety.co.uk**

Further information

There will be further updates about Project Sanfilippo in future editions of the MPS Magazine.

To sign up to receive our MPS Magazine by email, please visit our website www.mpssociety.co.uk

Gift Aid Q&A

www.wickedgenes.co.uk

What is Gift Aid?
It's a scheme introduced by the government which allows charities like the MPS Society to reclaim the tax that supporters have already paid on their donations. For every £1 donated, the taxman gives us an extra 25p, so our income from your donations increases by almost a third. We can only claim on donations made within the last six years', tax unclaimed before this will be lost forever, so please act now.

Why 25p per £1?
It's linked to the current basic rate of income tax (which is currently 20 per cent). As a basic rate taxpayer, for every £1.25 you earn, you will pay 25p to the taxman and receive £1 in your pocket. It is this 25p we claim.

Am I eligible?
If you are a UK taxpayer and have paid enough income or capital gains tax in the financial year to cover the amount we'll reclaim, then yes: please complete a Gift Aid declaration.

Where can I get a Gift Aid declaration form?
You can download a Gift Aid declaration form from our website or we can post one to you. Please complete one of these forms and return it to the address specified on the form. If you require assistance please call 0845 389 9901.

What if I'm a pensioner?
We can still claim the full 25 per cent on your donations, providing you pay at least as much tax as we will be reclaiming in the year in which you made your gifts. As a pensioner, for example, you may still pay tax on a private pension scheme or a savings account, or pay Captial Gains Tax if you sell property or shares.

What if I'm a higher-rate taxpayer?
We can only claim the basic rate back, but this is still extremely valuable. You will also be able to claim additional personal tax relief on your self-assessment form.

I'm not eligible; should I still return the form?
Yes please. It would be a huge help if you could return a declaration to us letting us know not to claim. This way, we won't contact you about Gift Aid again in the future, saving us time and money.

I've already completed a declaration for another charity. Do I need to complete one for The MPS Society?
Yes. You will need to complete a separate form for each charity you want to benefit from Gift Aid.

What else does ticking the Gift Aid box commit me to?
Nothing at all. It just ensures that, if you donate to us, we can claim money back from HM Revenue and Customs.

How long does my declaration last for?
Until you tell us to stop. At the moment, and as long as you remain eligible, your declaration lets us reclaim past (up to six years), present and future donations.

What do I do if I need more information on Gift Aid?
If you would like to find out more, or if you would prefer to make your Gift Aid declaration over the phone, please contact us on 0845 389 9901 and we will do our very best to help.

What if I change my mind, or am no longer paying tax?
Please contact us either by letter to The MPS Society, or by e-mail to fundraising@mpssociety.co.uk, and tell us that you no longer wish us to claim the tax on your gift.

What if I'm an events participant?
If you have received a benefit by participating, although anybody can sponsor you, MPS Society may not be able to claim Gift Aid from all your sponsors, including family members ('Connected Persons'). Please check with us for further details.

Will it really make a difference?
Yes. You will be increasing the value of your donation which will go the extra mile for those who need our support.

giftaid it

Making a donation

By post: Send us a cheque or postal order. Don't forget to enclose your address so we can reply and send you a thank you. Please don't send any cash by post.

At a bank: Pay in your cash or cheque at any branch of Barclays. Our account number is 33986306, sort code 20-02-06. Just let us know who you are and what you've done!

By credit card: Phone us or visit our website to donate online. Please ensure you let us know your details and how you have raised the money. Most major credit cards accepted.

Online: You can donate online through our secure server by visiting www.mpssociety.co.uk

For other ways of making a donation, please ask us for our Making a Donation factsheet.

Please let us know when you are making a donation or paying in money directly into our bank account and please include a reference for the payment. This way we can confirm safe receipt of the funds and say a big thank you. If we can't identify where the funds have come from then we can't thank you! **fundraising@mpssociety.co.uk**

Stamps, foreign coins, mobile phones, ink cartridges, jewellery: Andy and Jenny Hardy; Karen and Andrew Weedall; Tony Moran; Arriva; Judith Evans; Hannah Brock; Mr and Mrs P Harriss; Lockhart Catering Design Services; Langlea Care Home

In memory: Mrs Janice Oakes; Mrs Sylvia Dilys Morris; Jordan Lacey; James Nicholson; Denis Rowan; Ben Illingworth; Colin Arrowsmith; Daniel Allen; James Edwards; Colin Arrowsmith

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