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support
research
awareness
Wicked GENES

The MPS Magazine

Spring 2014

Society for Mucopolysaccharide Diseases

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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.

Our Aims:

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

To promote and support **research** into MPS and Related Diseases

To bring about more **public awareness** of MPS and Related Diseases

MPS and Related Diseases

Mucopolysaccharide (MPS) and Related Diseases affect 1:25,000 live births in the United Kingdom. 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, neurological deterioration can result in death in childhood.

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

Will you be part of our Wicked Walkabout?

We've seen a fantastic response since we first launched our Wicked Walkabout – people from Derry to Dorset, Bucks to Bridport, have donned their walking shoes in the name of fundraising and awareness for the MPS Society.

We would love to see more people wending their Wicked way across the UK, so if you would like to hold a Wicked Walkabout please drop us an email at **fundraising@mpssociety.org.uk** and we can send you out a Wicked Walkabout Guide. Get your friends, family and even your pet dog involved and get walking, running or cycling!

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.

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Welcome!

Welcome to the Spring 2014 edition of your MPS Magazine, the place to find all the latest news on MPS and related diseases, as well as stories from our members and fundraising ideas.

Christine's Chief Executive Report on page 4 mentions issues surrounding ERT for MPS IVA, as well as news of the MPS Symposium.

We have a moving and heartfelt account from the mother of a Sanfilippo sufferer in 'Jordane's Story' (P. 8-9).

Aisha's 16th birthday can be found on page 12, which details not only her glamorous party, but also her inspiring attitude.

Our advocacy support team have written a handy guide to benefits (p.15), as well as guidelines on how to choose the best school for parents of children with MPS or related diseases (p.16-17).

Read all about the wonderful family day at Lapland UK on page 19.

Recent clinical trial updates can be found on pages 20.

An update on the Genistein clinical trial is on page 22.

Christine's interview with Genzyme is on page 26.

All things to do with fundraising can be found from page 29, including your amazing stories, plus our fundraising ideas to mark MPS Awareness Day.

Contents

Chief Executive's Report


Christine Lavery, MPS Chief Executive

Whilst a majority of the LSD Clinical experts were in San Diego for the WORLD meeting on Lysosomal Storage Diseases the news came that the US Food and Drug Administration (FDA) has approved Vimizim ERT for MPS IVA. Focus now turns to the European Medicines Agency (EMA) where approval is expected imminently. However this is not the whole story and concern is rising here in the UK that it may take many months, possibly 1 to 2 years, for NHS England or ultimately NICE to approve Vimizim as a reimbursed enzyme replacement therapy in England. In Wales based on past and current experience the challenge may be greater. We are hopeful but not complacent that the situation for Scotland and Northern Ireland may actually be better than England and Wales. The MPS Society will be keeping our MPS IVA families and adults informed as the picture evolves and ask you to look out for emails and calls to action. Please do let us know if we do not have your up to date email.

In my last Chief Executive Report I wrote of the impending clinical trial activity for Sanfilippo disease. In Sanfilippo Type B during the WORLD meeting BioMarin announced their intention to join Shire and Synageva in developing ERT for MPS IIIB.

We feel that it is important all MPS IIIB families have an insight of all the pre-clinical work being carried out and clinical trials for the disease that may be open to UK MPS IIIB patients. To this end in consultation with Drs Simon Jones, Suresh Vijay and Maureen Cleary we are developing a MPS IIIB research and clinical trial parent information sheet.

As many of you are aware the MPS International Symposium on Mucopolysaccharide Diseases will take place in Sauípe, Bahia , Brazil, 13 – 17 August 2014. I am delighted to announce that funding has been secured to take five young adult MPS stakeholders to participate at this conference and Thomas Garthwaite (MPS II), Simran Bachu (MPS I H/S), Aidan Kearney (MPS IVA), Roswen McKnight (sister of Sarah, MPS I HSCT) and Jessica Reid, Trustee (sister of Daniel, MPS II) will be our team of young ambassadors. Our Chairman, Sue Peach will also be in Brazil with myself and two members of the MPS Advocacy Team. I do hope that we will be joined by other members for this very important meeting in the MPS calendar.

Christine Lavery MBE
Chief Executive
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What's on 2014

CONFERENCES and REGIONAL EVENTS

Ed Wraith Memorial Conference
5 April

MPS Awareness Day
15 May

Cadbury World Family Day
2 August

13th International Symposium on MPS Diseases, Brazil
13 -17 August

Chessington Family Day
15-17 August

All Ireland MPS Conference
12-14 September

MPS REGIONAL CLINICS

MPS I - GOSH: 22 July

MPS III - GOSH: 8 April, 8 July

MPS IV - GOSH: 24 June

N. Ireland MPS Clinic: 9 May

MPS I Post HSCT (over 6's) - RMCH:
4 July, 3 October

MPS I Post HSCT (Under 6's) - RMCH:
25 April, 11 July, 10 October

MPS Clinic - Birmingham:
9 May, 13 June, 14 November

Fabry Clinic - Birmingham:
23 May, 24 October

News from the Board of Trustees

The Society's Trustees meet regularly. Here is a summary of the main matters that were discussed and agreed at the Trustee Board Meeting held on 30th November 2013.

Governance

Trustees considered the MPS Strategic Plan presented by the CEO. Minor changes to the clinical trials section, conferences and income were suggested. It was agreed Trustees take the paper away and bring amendments to the next meeting where the Strategic Plan will be an agenda item.

Risk Management/Health & Safety

The Chief Executive confirmed that the regular health and safety checks and reporting were up to date and that Toni Ellerton has recently undertaken Health and Safety training. The Risk Management Register was considered and it was agreed that Senior Management succession planning will be discussed at the next meeting. Trustees, James Garthwaite and Tim Summerton agreed to work on improving the Business Continuity Plan and bring their findings to the next Trustee Meeting.

Personnel

The Chief Executive spoke of the changes to seating and the moving of the Fundraising and Communications and Finance personnel to the ground floor of MPS House. Trustees, recognising that all staff salaries had been frozen since 2009, agreed a 3% increase as of January 2014.

Treasurer's Report

Judith Evans read her report and it was agreed that the two mortgage figures on MPS House be shown separately. The Chairman invited comments and discussion on the draft budget for year ending 31 December 2014. Finance Officer, Gina Smith answered questions on income and expenditure. The draft budget was approved unanimously.

Advocacy

Trustees were informed that the MPS Chief Executive and the CEO from the Gaucher Association met with NHS England Commissioners, Sheila Upadhyaya and Barbara Howe to discuss the LSD Highly Specialised Service. Following on from this meeting on 17 October the MPS Society confirmed it has been feeding back on the Homecare Patient Charter and Service agreement in preparation for the ERT treatment efficiencies to be rolled out on 1 January 2014.

The Chief Executive updated Trustees on the situation with the two remaining national Homecare providers for ERT in England, BUPA and Healthcare at Home. The services by Healthcare at Home continue to be very patchy and this is being addressed by NHS England Commissioners and the LSD Collaborative led by Tanya Collin-Histed from the Gaucher Association. It was confirmed Sophie Thomas has drafted the Risk Charter documentation.

Trustees congratulated the Chief Executive on her invitation to be the patient representative on the Steering Advisory Group for Shire's Global Charitable Access Programme.

Clinical Management

It was confirmed that the MPS Society has been working closely with managers of the LSD service at Great Ormond Street Hospital (GOSH) in respect of MPS member concerns or complaints. The investigation into two formal complaints is drawing to a conclusion and the MPS CEO gave positive feedback that changes are emerging that address concerns raised. Trustees were advised of the resignation of GOSH's CEO, Jan Filochowski announced on 8 November 2013. Also that Locum Consultant Paediatrician, Dr Alex Broomfield who has been seeing LSD patients at GOSH two days a week has been appointed to one of the two vacant paediatric posts in metabolic medicine at Manchester Children's Hospital. The other went to Dr Berndt Schwan currently at Yorkhill Children's Hospital, Glasgow.

Research Update

Trustee, Professor Bryan Winchester spoke of the dramatic improvements to diagnostic approaches over the last three or four years, however he advised that this should only be done in large expert and accredited diagnostic laboratories attached to expert clinical centres.

Professor Winchester also spoke of pre-implantation Genetic Diagnosis and the need for this too to be done in larger centres, eliminating the potential for mistakes such as the one seen recently by the advocacy service.

The Chief Executive confirmed that three grants have now been made by the MPS Society to Dr Brian Bigger for the Genistein trial and that recruitment is expected to start in January 2014 at Manchester Children's Hospital.

MPS Commercial

Bob Stevens, a Director of MPS Commercial and Trustee of the MPS Society confirmed that the MPS Commercial Board of Directors met on Friday 29 November 2013 and that he was elected Chairman, Jessica Reid was elected Vice Chair and Gina Smith was elected Treasurer. Christine Lavery was appointed Company Secretary.

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New members

Mr Brown and Ms Chapman have recently been in contact with the Society. Their son Harvey has a diagnosis of MPS IV, Morquio. Harvey is 4 years old. The family live in the South East.

Simon and Elouise have recently been in contact with the Society. Their daughter Seren-Rose has a diagnosis of Hurler disease. Seren-Rose is 2 years old. The family live in East Anglia.

Amanda has recently been in contact with the Society. She has a diagnosis of Morquio disease. Amanda is 38 years old and lives in Scotland.

Mr and Mrs Paterson have recently been in contact with the Society. Their daughter Thea has a diagnosis of Hurler disease. Thea is 6 months old. The family live in the North in Ilkley, West Yorkshire.

Births

Congratulations to **Joanna (MPS IS) & Alex WIlson-Smale** on the birth of their daugther Elizabeth Violet Wilson-Smale born 30 December 2013 weighing 5lbs 7oz.

New faces at the MPS Office

CHRIS SCALES - Clinical Trial and Patient Access Officer

Hello, my name is Chris Scales. I started working for the MPS Society around the end of January as Clinical Trials and Patient Access Officer.

I am married with four wonderful children and three beautiful grandchildren. In a previous life I was a swimming teacher and I have worked in various offices, mainly clerical and admin., which I hope stands me in good stead for the work I will be doing with the Society. I was made redundant about 18 months ago, so decided to have a complete career change and went to work for the NHS in the maternity department, then decided I was happier working in an office.

In my spare time my main hobby is my animals: I have two dogs, a cat, three chickens, and also a horse, which I have had for many years. I enjoy reading, James Patterson being one of my favourite authors, although I will read most things.

I am hoping that whilst I am working at the MPS Society I will build up lots of good relationships with the families at the Society.

Chris Scales
c.scales@mpssociety.org.uk

ELKIE RICHES - Fundraising and Communications Assistant

Hi, my name's Elkie Riches and I joined the MPS Society as the Fundraising and Communications Assistant in March 2014.

I have been impressed by the MPS Society from the moment I walked in to their wonderful head office, and the more I learn about them and the work they do the more impressed I become! Their commitment to research and support for those affected is truly admirable and it is a pleasure to work for such an organisation.

My working background is based in Administration, but I am also a published writer of both fiction and non-fiction. Although these two roles seem very disparate, I think that both my administrative background and writing abilities will serve me well as I strive to communicate the importance of research into these diseases and the support that those affected need.

Outside of work I enjoy film, reading and theatre, especially Shakespeare, and to keep fit I practice a defensive martial art called Aikido and enjoy woodland walks.

I would like to thank my colleagues for their warm welcome and for helping me to settle in. I was instantly able to tell that each and every one cares about what they do and is passionate about the cause, which is a very refreshing and inspiring environment to work in. I am very much looking forward to exploring my role and making a difference.

Elkie Riches
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First MPS IVA Home Infusions for Olivia and Luke!

We had our first Home Infusion last Friday! It was a welcome change to travelling in and out of London. Olivia and Luke relaxed at home, and the Home care nurses, Lisa and Carly, set the pumps and drugs up to run inside a little backpack, which Olivia and Luke could walk around with. It was a very successful day and much shorter than our usual infusion day. The nurses arrived at 9.30am and were gone by 4.30pm. It is a day I thought we'd never see.

Ita Vickery, mum of Olivia and Luke

Please update your contact details

We like to keep you informed of news, events, information and opportunities. To minimise our costs we aim to contact you by email wherever possible rather than by letter so it is vital that you keep us informed of any changes to your contact details and let us have your current email address.

Please email **mps@mpssociety.org.uk** to advise us of your email address and we can amend our records.

Supporting re-housing at the Olympic Village

I have been supporting a family living in London who needed re-housing due to their existing home not being suitable with lots of steps inside. I wrote a letter of support to the local authority explaining about their MPS condition and how it limits their mobility and how the family find it very difficult to live in their home. They were looking for social housing with level access inside and out.

Suitable accessible housing in London is hard to come by so I was delighted to hear that the family had been offered a lovely flat which was part of the Olympic village. It is good to know that the developers of the Olympic site have thought about the future and housing needs and the athletes village is now to be a new London postcode E20 and known as East Village.

The family moved into their new Olympic home over Christmas and I was fortunate to visit the family in February. The village has a new school, GP surgery and is close to the Westfield shopping centre with excellent transport links to the rest of London. The family are thrilled with their new home and how accessible everything is.

However, I was also impressed by the fantastic views of the Olympic park from the balcony and knowing that in 2012 Olympians and Para-Olympians had stayed in that flat – what a legacy!

Debbie Cavell
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All Ireland Advocacy Officer nominated for a Woman of the Year award

Alison Wilson who was nominated for the Belfast Telegraph Woman of the Year Award in the voluntary sector by Dr Fiona Stewart and two patient representatives was shortlisted and requested to attend an award ceremony on 27 February where the final winners were to be announced. Although not successful in being awarded a prize she was absolutely delighted to make it to the final four.

'I am so humbled to have been nominated for such a prestigious award. There were dozens of truly inspirational women shortlisted for the award and it was an honour just to be in their company. All of the MPS Team work tirelessly to support our members and I consider myself to have been at the awards dinner representing the entire MPS Team.' **Alison Wilson**. Well done Alison for making it through to the finals.

Congratulations to Dr Fiona Stewart MBE

Dr Fiona Stewart and her family went to Buckingham Palace on January 24th so she could receive her MBE from Prince Charles. His Royal Highness asked her about Genetic Medicine and what was being done to improve treatment for rare genetic disorders. Afterwards she went for lunch at the Goring Hotel with other family members and friends. Everyone had a great day out!

Jordane's Story

I begin to write this as I start to buy presents for my daughter's birthday.

It's obvious I'm a little upset after struggling to find more than 2 suitable things. 'This isn't how it was meant to be,' I thought.

Even now, after all these years, it is the small, mundane things that make you stumble, which jolt dark thoughts buried deep in the recesses of your mind.

Soon our daughter celebrates her 18th birthday, that landmark moment when a child turns almost overnight into an adult. There will be a small party and presents.

But sadly she will not toast her big day in champagne, nor dance the night away with friends. She will not tear open her presents, blow out candles on her cake nor laugh as we recall happy childhood memories.

Sadly our daughter suffers from a rare life-limiting condition that means she no longer is able walk or talk. She is peg fed, endures epileptic seizures and needs round-the-clock care.

She relies on others for the most basic everyday tasks and must have someone with her all the time in case of emergency.

Our house is filled with carers and adapted with a hoist, there are bottles of oxygen in her bedroom, life-saving drugs and sleep medication in the kitchen. None of this was what we envisaged when she arrived on 10th March 1996

But for all the tough times, for all her pain and our heartache, soon we will celebrate her big day. Not just because she is brave, but because it was thought that she may not be expected to make this momentous occasion.

We will celebrate because while the rest of the world sees only a tragically disabled child we see an affectionate, adorable and amazing daughter.

Yes, through a quirk of fate she has turned our lives upside down, inside out, back to front and made daily life at times unbearably difficult.

But set against that have been so many unexpected highs — the joy, the laughs, the warmth of her uncomplicated love and rich glimpses into another world and different reality of life. Without her, our own lives would have been potentially so much easier but in truth, so much poorer.

So a very happy 18th birthday to our beautiful daughter Jordane.

There has been a moment in so many conversations over the past years when I know exactly what is coming next. My heart sinks: I am about to be asked about my children...

Not that I mind talking about them. On the contrary, I am deeply proud of them all...

It's just I know how the conversation will go. The embarrassed pauses, the faltering questions, the fumbling sympathy, then the search for a way out. Like I said, it was never meant to be like this.

Jordane's birth was normal, after spending 3 weeks in the Special Care baby unit at Sheffield Children's Hospital, she seemed strong and healthy — and it was hard to see how life could have been better with a nice house, marriage and soon to be baby number 2 on the way.

I remember every detail of the day everything changed. A Paediatric assessment was set up and we saw the Paediatrician who did numerous tests, including the impossible urine sample. Two weeks later on Tuesday 24th November 1998 a telephone call came from the Paediatrician requesting we came to see him the very next day! Immediately I knew it was bad news.

Wednesday 25th November 1998 - METABOLIC DISORDER?... SANFILIPPO SYNDROME?...

The Paediatrician examined Jordane before asking a book of questions about her and our own medical histories. Then he broke the news all parents dread: 'I am afraid it is terminal.'

Outside, pouring rain mingled with tears streaming down my face, the doctor's words replayed over and over in my head, pounding my brain and reeking of despair, as I walked to the car.

It seemed life was over. These were the darkest of days, putting on a brave face and pretending all was fine while struggling with deep depression. We felt like zombies, deprived of sleep and tormented by fear of the future.

Psychologists believe parents in such instances suffer grief for the baby they have lost, before gradually coming to terms with the child they have.

Meanwhile, there were endless hospital appointments, meetings etc, again and again, doctors, nurses, many professionals asked the same questions about her birth and our backgrounds. We were quickly starting to discover life becomes harder, not easier, as a profoundly disabled child gets older and bigger.

First and foremost was the fight to keep her alive and in best possible health.

But nothing prepared us for the wretched seizures that blight her life, which can leave her wiped out for hours/days and needing emergency medication.

Even now, having seen lots of them, they are torture to watch. They don't just endanger her life — each one erodes her brain, taking a little more away of our baby girl.

The other day, I was looking at pictures of her when younger, she truly was so beautiful, innocent...the way it should be.

Life is like being on a roller-coaster. Long-planned social events are thrown at the last minute, while even a small walk can end with a seizure.

There are months when her condition is comparatively stable, followed by weeks in steady decline. But it is too easy just to see the disability rather than the child.

She is a warm and affectionate girl, often smiling when she hears us, perhaps stretching out her hand or nestling her head against mine. She loves to watch tv, be around people, clapping, singing and simply just to hear music. There are many happy memories of that childhood that we will treasure forever...

We ensure she has as full a life as possible, doing things other children and teenagers do, holidays, swimming with the dolphins, Lapland, concerts!

She is, of course, too big to throw around and sit on our knee for 'Louie's' (our nick-name for hugs and kisses) while ourselves and carers need a hoist to help her into her wheelchair, bath and bed she still has the same lop-sided 'Elvis' smile, although she looks young for her age and we estimate she is 4 to 6 inches smaller than she would have been without disabilities, she still remains the most adorable, infectious and intriguing girl I know.

Somehow things seem more brightly coloured with her in the picture. A friend whose Sanfilippo child died told me it felt like life was less 'fizzy'; I know exactly what she meant.

Behind our front door, it seems normal to have this precious girl, albeit sometimes manic, sometimes passive, but with a strange ability to bring joy, love and happiness to people who see her for how she really is.

She has had a huge impact on my life — on my family, my friendships, my career and even my politics. For it was never the disabilities that were the biggest hurdles.

The real problems, as for so many of the other 100,000 families in our situation, lay in trying to clear a path through the undergrowth of a welfare system that fails too many of the most vulnerable people.

This is what makes daily life such a task, that forced me to abandon work — the soul-destroying struggle against dysfunctional public services, which costs so much time, saps so much energy and wastes so much money... A dysfunctional system that grinds you down, driving you to despair.

This is one reason why poverty, divorce, depression and unemployment are so much higher among families with disabled children.

We have even seen these attitudes in the National Health Service. Yes, there are some fabulous people working there, but we have seen too many who shame supposedly caring professions: the doctor who professed to be comparing two X-rays when only one had been taken, covering his mistake by saying there was another child with the same name, age, condition and on the ward at the same time, discharged with wrong medications and even after questioning the nurse, we were told to administer the medication and the rude receptionists too busy doing nothing to speak to you are amongst the list of many incompetences.

Hospitals are ill-equipped to cope with complex long-term conditions needing a multi-agency response, despite an ageing population and growing numbers of profoundly disabled and terminally Ill children.

This is why complaints are rising and the reason for scandal after scandal over shameful neglect of elderly patients. Yet still they carry on with bad and outdated practices.

The simplest things take forever. It takes months, numerous phone calls, meetings and home visits from professionals for much needed equipment and resources.

But so often professionals fail to read or answer reports, emails, phone calls and ask questions answered many times before.

But for all the trials and tribulations, all that really matters is we still have our wonderful daughter who has brought so much love, joy and happiness into our lives.

For the first nine/ten-years, we struggled through on our own, never really asking for help. A couple of years later, a social worker saw how close we were to breaking. She helped win first one night, then a second, of respite with our daughter staying overnight at a couple's home, the woman (Val) used to work at Jordane's school. However, at first it was hard to accept we needed help and to have our daughter stay elsewhere with someone else overnight. It felt unbelievable to have an evening out together, and an entire night's sleep.

Now after a long struggle we have a good package of care based at home. Our house is filled with a constant flow of carers — but at least Jordane is kept safe.

In the disjointed world of public services, everything is about to change now Jordane is soon to be 18... an adult! We lose all the things we spent so long fighting for - doctors, the integrated services for disabled children, the children's continuing health team. However, the life-saving palliative care team will hopefully be introduced along with the adult continuing health team and her end of life and Metabolic Consultants. We must, however, now learn how to navigate the bewildering new world of adult services, so a fresh battle opens up. More meetings, more questions and a flood of new forms to fill in and an abundance of the same old questions to answer... all over again!

Normally 18 marks the age your child sets out on their own path, making all the choices over their own future. Instead, our daughter must remain dependent on our decisions — the choices available here are so limited and poor.

But for all the struggles, hardships and heartache all that really matters is we are still blessed to have our wonderful daughter who has and still does bring such unexpected love, joy and happiness into our lives.

Happy 18th Birthday Sweetheart!

Paula Robjohns

The Olly and Willow Show

Olly and Willow were both diagnosed with MPS I Hurler disease some 9-10 years ago and underwent haemopioetic stem cell transplants (HSCT) soon after diagnosis. These transplants have served the children well.

However whilst educationally and socially both children are thriving over the past few years they have encountered increasing mobility problems. When the MPS Society secured three places for UK children on the Phase I/II Enzyme Replacement Therapy Study for MPS I children who have undergone HSCT, Olly, Willow and a third child were soon recruited and have been on weekly Enzyme Replacement Therapy (ERT) at home for 5 months now. The article below is Olly and Willow's own story of their 6 month assessment visit to see Dr Paul Orchard at the University of Minneapolis in February.

Christine Lavery, Chief Executive
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Our trip to Minneapolis for our 6 month ERT Clinical Trial Evaluation

We took off from London Heathrow on Saturday 15th February 2014 during the gale force winds bound for Minneapolis in America. The Pilot warned us that it would be a bumpy and turbulent ride ahead. We arrived in Minneapolis where it was minus 20 degrees centigrade and snowing.

We were staying Downtown about a 10 minute drive from the University of Minnesota Hospital which is located on both the East and the West Banks of the Mississippi River which was frozen over.

After settling in at the Marquette Hotel and having a few room changes, we went out to explore the local area and found the nearest Starbucks for much needed hot chocolate and coffee for our Mums. Christine arrived in from the WORLD conference in San Diego and we gave her a big hug. We went out to find somewhere for a good American burger and fries.

Sunday – Fun Day!

We all met for breakfast to fuel up before a day of action at the Mall of America.

We took the Minneapolis Tram to the Mall of America. It was freezing waiting for a train (and we only had to wait for a couple of minutes).

We arrived at the Mall of America.

Once inside we hit the rides for Olly and a bit of shopping for Willow.

Olly, *"feeling the need for speed"* went on a huge amount of rides - 8 in total! He got our Mums on the Spongebob Rollercoaster three times, we went on the Log Flume twice, the Pepsi Train once, the Flying Ninja Turtles once and the spinning Fairly Odd Parents Rollercoaster twice. Willow took a more cautious approach starting with Spongebob, the Log Flume and the Pepsi Train.

Olly wanted to go on the most daredevil ride "The Airbender" but none of the girls had the stomach for it so we said he could do it with his Dad next time. In between rides we did a little bit of shopping. There are so many things to do in the Mall of America - rides, shops, cinema, seaworld and they even had an iPad vending machine!

In the evening, we celebrated our 6 month anniversary of weekly Enzyme infusions which are making us feel a lot better. We can walk much further now and we are not in so much pain or as stiff as we were before. It has also helped us at school to concentrate more and we can write more easily and for longer. We can now keep up more with our friends at school. We all laughed when Willow said we had a bit of 'Jet Leg'.

By Olly and Willow

Monday – Hospital Day

We were so excited as we woke up to a blizzard of snow over Minneapolis. We were expecting 7 inches of snow overnight!

We arrived at Amaplatz Children's Hospital for a day of tests to see how we were doing. As you can see it was under a thick layer of snow.

First of all we had our bloods taken. They took 12 pots and neither of us had EMLA numbing cream.

Then we had our meeting with the 'main man', Dr Paul Orchard. He made us laugh. We then did a physiotherapy appointment and they really knew their stuff. Our Mummies were given lots more forms to fill in.

We then went out for a large snowball fight and lunch at Applebees across the road the from the Children's Hospital on the other side of the river.

After lunch we had another snowball fight with Christine launching the first snowball.

This was on the way to our next appointment at Biodex Testing which tests the muscle strength of our legs, arms and hands. Both of us have got stronger since we have been on enzyme.

Olly made Snow Angels and found a brick too (!) on the way to our next appointment.

Dr Orchard came to our next appointment and it was nice to see him again and he made us laugh.

We did the Balke Treadmill Test where they measure our heart rate and oxygen levels during exercise. We didn't like the heart rate monitor stickers so we stuck them on Joe the nurse too.

Hard day's work (hardly!!) finished in time to play some more. As you can see we did not start the next snow ball fight!

So Olly had to wade in there...

And then Willow threw the next snowball!

We had such a lovely time, spending time together made the appointments so much more fun. We look forward to our next trip in August for our yearly review and some sunshine too.

We have a good friend Leon who is running the London Marathon on Sunday 13th April 2014 and all sponsorship goes to the MPS Society to help fund their invaluable day to day support for families and children living with MPS and related diseases.

Please Sponsor Leon!

Visit http://uk.virginmoneygiving.com/LeonBatchelor

Aisha's Glitz and Glamour 16th Birthday!

Our Aisha was born 3rd November 1997. Aisha was diagnosed with Morquio Syndrome in June 1998. She recently celebrated her 16th Birthday and what was to be a small get together with family and friends turned out to be her best birthday, and she did it all for the MPS Society, raising a substantial €635.00!

A few months back Aisha commenced ERT at Manchester Children's Hospital. The risk considerations were discussed with us at length as Aisha has a narrowing airway. Aisha's aspiration in life is always to help others, and knowing the risks she opted to go on the trials with a determination to help other Morquio patients in future as well as herself. The team at Manchester were absolutely fantastic. The care we received whilst Aisha was on the trial was significant and we as a family were supported very well during the months of Aisha receiving the drug. I talk in the past tense because unfortunately for Aisha the side effects were too significant and in week five of her infusion she went into a severe anaphylactic shock. A dreadful and bloodcurdling experience. The CRF ward team and emergency medical personnel reacted instantly. Aisha was stabilised and discharged when she recovered.

This was very distressing for Aisha and us as family. We had started the ERT knowing it meant weekly infusions in Manchester. Travelling from Leicester to Manchester every Monday night to be at the hospital every Tuesday morning. We had decided this was a small sacrifice to maybe give Aisha a better quality of life and help the drug get licensed in the UK quicker with a range of patients with different challenges on the drug.

We had waited for the enzyme ever since Aisha was diagnosed and now that it was available to her, her body reacted. We were heartbroken to see that the enzyme Aisha's body lacked was available to her but now caused her this life threatening episode. Aisha had to stop and be withdrawn from the infusion. Our Almighty creator has reasons for everything. Aisha has been exceptionally supported by the team in Manchester, consultants in Leicester and her college since she has had to stop the drug infusions. One thing she said to us was that at least the pharmaceutical company and the health professionals are now aware of risks for someone with a narrow airway. This is heartening and we know Aisha will have in some way helped so many.

So where did the Glitz and Glamour 16th come into all this... Aisha had always wished her 16th birthday celebrated around fundraising for the MPS Society and research into Morquio syndrome. Earlier this year she had contacted various organisations who hire venues and planning her 16th with a night of celebration and fundraising. She had over 150 people on her list ranging from old school friends, to support workers to people in the community that have always been there for her and not forgetting her dear family and friends.

Unfortunately, all these plans came to halt as she became quite unwell after her anaphylaxis incident. As parents we decided she could still possibly have her wish but on a smaller scale. So we called the 'party planners'. These so called 'planners' were in fact my niece, aunt and her children. Within six days they organised a venue, food and invites, whilst I organised a few gifts for the attendees. It was incredible. Sumeya, my niece, bought the trimmings. The venue was kindly donated by my aunt Salma. Day by day things were falling into place and as far as Aisha knew it was just a get together with family and friends. She had no idea what was going on behind the scenes. All she worried about in the meantime is what are we going to give as presents to everyone who comes.

The theme was still centred on the MPS Fundraising so the invite asked everyone to be glamorous and "wear it BLUE!" No presents but only donations. On the day of the party, we had yet another surprise. A beautician turned up at our door to get us all glamorous for the party. It was all thanks to Salma for arranging that.

When we arrived at the venue it was surely a 'red carpet' event! Aisha even had her own bouncers surrounded by papparazzi! It was tearjerking and everything was so special. Special because everyone had just come together to make Aisha's night so special and an event we shall never ever forget. There were speeches, a powerpoint presentation and prize giving. Lots of food and desserts and cake! Everyone was part of it and contributed to the event.

A few days later we opened the donation box and in there was everyone's kindness in support of Aisha's charity and cause that is very close to her heart and ours. To date Aisha has raised more than £2000 in total for the MPS Society and her goal in life is to continue to raise awareness for the Society. (www.justgiving.com/aisha-seedat). **Asma and Sharif Seedat**

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** 0845 389 9901– 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 claim forms for Personal Independent Payment, 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:

Email: advocacy@mpssociety.org.uk

Telephone: 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.org.uk

- **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**
- **Benefits including Personal Independent Payment, Benefit Cap, Council Tax Benefit and Universal Credit**

Each of our England based Advocacy Officers works with specific disease groups as listed. However, every member of the Advocacy Team has knowledge of all the diseases and may at times provide support in other areas dependant on need and individual assessment

Team members

SOPHIE
Manages the MPS Advocacy Team

REBECCA
Fabry
MPS II Hunter
ML III / ML IV
Mannosidosis
Fucosidosis

STEVE
MPS III Sanfilippo
MLD, AGU
Winchester,
Geleo Physic Dysplasia
Sly, Gangliosidosis
Sialic Acid Disease

DEBBIE
MPS IV Morquio
MPS I Hurler BMT,
Hurler Scheie, Scheie
MPS VI Maroteaux-Lamy
MSD, ML II

ALISON
Supports members living in Ireland

All Ireland Advocacy Support Update

A lot has happened in Ireland in the first quarter of 2014. It's been wet and very windy but the work of the All Ireland Advocacy Support Service has continued. As usual I have had the pleasure of meeting with many of our families face-to-face to provide individual advocacy support in relation to all sorts of issues. We can only hope that the MPS Society has been successful in supporting families and making life a little easier when they needed it most. If you live in Ireland (North or South) and have an unmet support need please do not hesitate to get in touch!

Alison Wilson
Telephone: 0044 77862 58336
or 0044 28950 47779
Email: a.wilson@mpssociety.org.uk

Education

You will have read my articles in the past and be aware that education is one of the key priorities for the All Ireland Advocacy Support Service (and for the rest of the Advocacy Support Team based in Amersham). We are always looking for opportunities to spread the word; and this year those opportunities are coming in 'think and fast' (as we say in Northern Ireland).

On 7th February, Dr Fiona Stewart M.B.E (Northern Ireland Regional Genetics Service) and I were delighted to host the first 'Recognising a Rare Disease' Study day (made possible by educational grants from three pharmaceutical companies).

This was a day targeted at Doctors working in orthopaedics, radiology, and rheumatology. Our aim was to increase awareness of metabolic conditions (including the MPS and related conditions) so that medical teams are more likely to recognise one of these rare conditions when they see one.

We are all too aware that many of our members go through months of confusion prior to their diagnosis and one of our main aims as a Society is to increase awareness and shorten the time to diagnosis.

On the day we heard from Dr Fiona Stewart, Dr Simon Jones, Ms. Andrea Jester, Prof. Richard Baker and Dr Richard Cowie who gave an excellent overview of some of the key orthopaedic features of some of these very rare conditions. I was also pleased to be able to educate the professionals who attended about the work of the MPS Society.

Thank you to all involved! We hope to run more of these study days for other specialties in the future.

Our next big event in the world of education will be a training day for nurses - supported by the Royal College of Nurses - in April 2014.

Rare Disease Day

This year the theme for International Rare Disease Day was 'Join Together For Better Care'. The MPS Society always strive to work closely with other patient lobbying groups to promote the interests of our members; and on Rare Disease Day we did just that.

On 28th February professionals, families, parents, carers, patient organisations, policy makers and politicians joined together at an All Ireland Event (organised by The Northern Ireland Rare Disease Partnership, the Rare Disease Taskforce Ireland and Rare Disease UK) to celebrate Rare Disease Day.

The event was a roaring success and we were delighted to welcome Minister Poots (Minister of the Department of Health, Social Services and Public Safety NI) and Minister White (Minister of State for Primary Care ROI) to the event to update the delegates on the progress of the Rare Disease Plans for both the North and South of Ireland.

Minister Poots spoke about how 'rare disease is a key commissioning priority' and Minister White gave his full backing to 'increased cross-border cooperation between Northern and Southern Ireland'. Those living in Ireland will not underestimate just how important a step it was to have both Ministers standing side-by-side speaking so positively about joint working and the future for rare disease care.

I was delighted to be given the opportunity to address delegates while chairing a session entitled 'Joining together for better care – examples of good practice'. It is always fantastic to be able to promote the MPS Society and raise awareness at these types of events.

Celebrations

In December we celebrated the 1st birthday of the Northern Ireland Clinical Trial satellite site. We never let an occasion go by without a little celebration and this was no exception!

Aidan Kearney (MPS IV - Morquio Disease), Alison Wilson (All Ireland Advocacy Support Officer) and Dr Fiona Stewart (Consultant in Genetic Medicine)

Advocacy - Focus on ... BENEFIT INFORMATION

Personal Independent Payment (PIP)

Due to some last minute changes and difficulties with the new forms, only a small number of people, in restricted areas, were transferred from DLA to PIP in October 2013. It is envisaged that this will be rolled out to more areas during 2014. A map highlighting areas being assessed can be found at https://www.gov.uk/government/publications/pip-postcode-map-uk. However, it is anticipated that the majority of claimants will not be reassessed until 2015 or even later dependant on the outcome of the independent review which is to take place sometime in 2014. This is particularly so for those individuals who have an indefinite award of DLA of which there are no changes to their circumstances.

Incapacity Benefit

It is envisaged that all Incapacity Benefits which includes Incapacity, Severe Disablement Allowance and Income Support on disability grounds, will transfer to ESA by the end of March 2014.

Child Tax Credits

As of April 2014, parents/guardians will be responsible for notifying HMRC by the 31 August, each year if any child aged between 16 – 19 years, included in the claim is remaining in full time non advanced education. If this is not done on a yearly basis and by the 31 August, the child will automatically be removed from the claim and your award will be reduced or stopped accordingly.

Job Seekers Allowance

There are a few changes being implemented to job seekers allowance these are:

- The waiting period for first claims is being extended from 3-7 days
- New claimants will have to attend longer interviews and provide a Curriculum Vitae (CV)
- Those who have poor spoken English will have to attend English classes or face sanctions
- For those deemed to be not doing enough in finding a job will have to attend weekly meetings instead of fortnightly.

Universal Credits

The full roll out of Universal Credits in April 2014, will not happen as planned. Instead from April 2014, couples and families will be able to claim universal credits only if they live in one of the 10 pilot areas who were assessing single people. Areas included in the pilot scheme can be found at https://www.gov.uk/universal-credit/eligibility. By the end of 2014 it is hoped that Universal Credits will be rolled out across more of North West England but it will not be available to all areas until 2016.

In April 2014, Universal Credits may be introduced in Northern Ireland. However, this is dependent on the outcome of the welfare reform bill, which is yet to be passed.

Using genetic information in pre-employment checks

The MPS Society Advocacy Team were contacted by a member of the Society in respect of being refused a specialised job because she had a genetic condition 'Fabry' which may affect her later on in life. Despite the fact that she was non symptomatic and was very fit and well.

The family decided that this was a matter which they needed to pursue as it was felt that this was an important issue which could surface for anyone dealing with potential discrimination by employers, insurance companies, government etc over decisions made on the basis of an individual's genetic inheritance.

The matter was referred to solicitors who dealt with this area of expertise and an internal appeal was made to the potential employers. This did not result in a favourable outcome and the case was pursued in the High Court, following the family being able to obtain legal aid for the judicial review.

The arguments were based around various legislation such as Article 8 and 14 of the European Convention on Human Rights, Data Protection Act, and the Equality Act.

It was agreed that the applicant should not have been refused the job solely based on her genetic information and happily she is now pursuing her chosen career.

It is always hard to fight the system but it can be worth it in the end....

Bereavements

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

Faye Barnett who suffered from Sanfilippo disease and who passed away on 30 December 2013 aged 19 years.

Kayen Ramani who suffered from I-Cell and who passed away on 30 December 2013 aged 2 years.

Edward Morley who suffered from Sanfilippo disease and who died on 25 September 2013 aged 26.

Advocacy - Focus on ...
A GUIDE TO VISITING SCHOOLS

A few tips and advice when choosing a school for your child

Below are some tips and points you may like to think about when visiting a school. You may want to consider your child's individual needs as there might be important points or questions not included here.

Allow plenty of time for the visit and think about whether you would like your child to attend with you. Choose a time when lessons are happening so that you get a true picture of the school and the atmosphere and teaching styles. Also ask if you can meet the staff who would be involved in you rchild's school day such as class teacher, assistant or SENCO (Special Educational Needs Co-ordinator).

Visiting the School

You may like to go with a friend or relative so that you can discuss the school after the visit. Think about how your child would travel to the school. What arrangements would you need to make to get your child to school?

Ask to meet the staff who could be involved with your child's education and care. If you would like to meet with a specific member of staff, such as the Headteacher, ask for an appointment in advance. Ask how the individual needs of each child is met within the classroom. Are there any children in the school with similar needs? Has the school any past experience or knowledge of MPS conditions? How is progress measured and how is this shared with parents/carers? How does the school communicate with parents/carers?

Therapy

Ask which external professionals visit the school relevant to your child such as:

- Speech and language therapist
- Occupational therapist
- Do specialist teachers visit?
- Are learning assistants trained to deliver specialised programmes?
- Is training available?
- Would the school be happy for the child to receive ERT whilst at school?
- What provision would the school put in place to ensure learning is not missed through accessing therapy and medical appointments?

The School in General

• Ask if the school would be able to provide any equipment/adaptations your child may need eg hoist, specialist seating. If your child has received funding via a statement how would the school use these funds?

• Ask how your child would be supported outside lessons, such as at lunchtime and playtime.

• Is the whole school accessible to your child, both inside and out?

• Will your child be encouraged to attend clubs and school activities and trips?

• Is there the opportunity for dual registration or outreach support at a special school?

• Does the school have a school nurse?

• Ask for copies of any policies that interest you such as behaviour, SEN or disability/inclusion (these may be available on the school website).

• Are there opportunities for parents to become involved in the school?

• How does the school manage challenging behaviour?

• Do the children look happy and well-behaved?

• Does the school seem welcoming?

Additional Support and Information

These organisations can be a useful source of further information:

Parent Partnership Service
http://www.parentpartnership.org.uk/find-your-pps

This service is offered by local authorities to help provide information and support to parents/ carers at all stages of school life. It offers confidential advice on special educational needs and the statement process.

GOV.UK
www.gov.uk/browse/education

For general education information such as admissions, attendance, special educational needs and transport.

To find local schools in your area: www.gov.uk/find-school-in-england (this page also gives links to schools in Scotland, Wales and Northern Ireland)

Ofsted
School reports can be downloaded from www.ofsted.gov.uk/ofsted-home/inspection-reports

MPS Society
www.mpssociety.org.uk

We also have available on the MPSSociety website a downloadableresource about the educational statement process entitled Statutory Process for Statutory Assessment for Special Educational Needs. Additionally, if you would like support and advice regarding visiting a school or the education process the MPS Society advocacy support team are happy to help with any queries you may have.The team can be contacted on 0845 389 9901.

Positive Changes for the Lysosomal Storage Disease (LSD) service at Great Ormond Street (GOSH)

It is always of considerable sadness when patients and parents raise concerns with the MPS Society in relation to the quality of the LSD service at any of the expert LSD centres. The MPS Society and the LSD Collaborative have had a very respectful relationship with Great Ormond Street Hospital in working to shape the LSD Service and up until Dr Jane Collins left her post as Chief Executive in 2012 we had had regular face to face meetings.

Unfortunately over the past two years concerns relating to the administration of appointments, clinical management and access to a clinical trial have risen. It is never easy for families to make a complaint even when supported by the MPS Society but I must pay tribute to senior members of the medical faculty, the complaints team and Anna Jebb service manager at GOSH for their diligence in investigating these concerns and consulting with the MPS Society on improvements going forward. The outcome of this LSD Service review highlighted below, we believe, will result in a greatly improved LSD service for all GOSH patients and their families.

"The Metabolic team at GOSH have recently completed an internal review of their staffing structures in Metabolic medicine/LSD and are reconfiguring the staffing structure to ensure that all complex metabolic patients have a single point of administrative contact into the Metabolic service, that there is one single Metabolic consultant who takes an overall lead for their care and to ensure there is good CNS cover for the service. As a consequence, the team have appointed 2 additional CNS posts and are about to appoint to 2 additional medical secretary/coordinator posts to support the service. With the additional posts there will be a greater level of cross cover with the administrative and CNS roles which will give the service greater long term sustainability. The consultant model particularly for the LSD service is also changing with all the Metabolic consultant team, over the next 6 months, taking on an equal LSD patient workload. This is to ensure that all consultants keep up their expertise seeing and treating patients with LSDs, and that the LSD service is sustainable longer term.

All new LSD referrals are discussed weekly at an internal Multi-Disciplinary Team (MDT) meeting with the whole metabolic/CNS team to ensure there is good peer discussion about each patient and that the expertise at GOSH is shared across the team for the optimum benefit of patients, and also staff. The team have also appointed 1 additional substantive Metabolic Consultant post at recent interviews, and a fixed term 12 month locum consultant post." Anna Jebb, LSD Service Manager

The MPS Society warmly welcomes the outcomes of this internal review and will continue to support GOSH in the best way we can. As Anna Jebb has indicated the review is in its implementation stage and it will take a few months to appoint staff and bed in the changes in clinical management. Another development is an increase going forward in disease specific Multi-disciplinary Team clinics with the introduction of three or four a year for children diagnosed with MPS I who have had a Haemopioetic Stem Transplant. MDT clinics for MPS III and MPS IVA will continue to develop.

Through the MPS Society we would be pleased to pass on your positive experiences of outcomes of the internal review of the LSD Service. Should you have any concerns related to any aspects of your child's care at GOSH or any other LSD specialist centre including the out of hours service, appointment management, transition or clinical management please let us know so that they can be addressed without delay and empathetically.
Christine Lavery, Chief Executive
c.lavery@mpssociety.org.uk

www.mpssociety.org.uk

For information on the MPS Society please visit our website. We do update the site constantly with all our current news on research and treatments as well as fundraising events and ideas. The format is very user friendly, so you will have no trouble finding what it is you are looking for, whether that be resources and guides from our Advocacy Team, to news regarding clinical trials. We even have a whole host of fundraising ideas as well as posters, sponsorship forms and guides to download. While you're visiting us, please don't forget to take a look at our online shop for T-shirts, badges and bands.

If you have any suggestions on ways to improve our site, please don't hesitate to get in touch by emailing **info@mpssociety.org.uk**.

'Like' and 'Follow' to Raise Awareness

With Twitter's users reaching the 175 million mark, and Facebook climbing to a staggering one billion, it is undeniable that social media plays a huge role in modern communication. These platforms offer us a fantastic opportunity to raise awareness of Mucopolysaccharide and related diseases, as well as allowing us to keep all our supporters up to date with the work of the Society. With your help we can spread the word, so please support us with your 'likes', 'shares' and 're-tweets'!

Find us on Facebook
www.facebook.com/mpssociety

twitter
twitter.com/MPSSocietyUK

Scottish Family Day

Fun, excitement and a little bit of magic, was the order of the day on Sunday 2nd March when the MPS Society held a special event for Scottish families at the Hilton Edinburgh Airport, all of which was made possible by the kind support of a grant from the Souter Charitable Fund.

The weather on the day was clear and fine (with no snow, unlike last year!), which helped our aim of banishing those post-Winter blues and welcoming in the Spring.

There was something on offer for everyone as all the families sat together for a fabulous two-course buffet, with plenty of food and drinks, giving all a chance to get to know each other, or else to catch up with families they had met before. There is always so much to talk about! A few of the families who attended had never been to any of our events before, so it was nice to see some new faces.

After dinner the entertainment came out in the form of Tricky Ricky, who delighted and amazed with his magic tricks and balloon models. Both children and adults alike were spellbound!

Thanks to the Souter Charitable Trust, and all those who attended, we were able to provide a fantastic afternoon with a truly great atmosphere, allowing the Scottish families affected by MPS and related diseases to meet one another and to know that they are not alone... and, of course, to have lots of fun in the process!

MPS Regional Specialist Clinics

Spring 2014

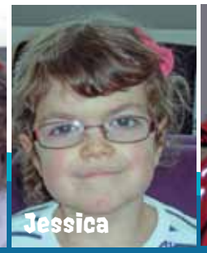
The advocacy team have been busy attending clinics across the country.

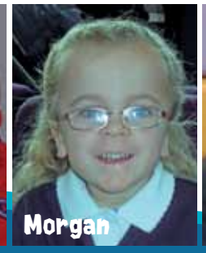
Since the last magazine we have travelled to Manchester for the under and over post HSCT MPS I clinics and to the Birmingham MPS clinic.

The purpose of these clinics is to not only allow individuals to regularly see one of the specialist consultants but to also be able to see other medical professionals and hopefully limit the number of separate appointments individuals and families have to attend.

The clinics also allow individuals and families to meet together and to access support from the MPS advocacy support team, who attend these clinics. Thank you to all the medical teams who attended the clinics and helped in their organisation.

Sophie Thomas
s.thomas@mpssociety.org.uk





Photos left to right - April Losztyn, Jessica Stringer, Mikko Astle and Morgan Wright - HSCT Clinic; Sultan Ali (Morquio) - Birmingham Clinic

Lapland UK Family Day

Following an award from BBC Children in Need, it was wonderful to be able to arrange A special Superstar Day visit for our members to Lapland UK, recently set up in the Windsor Forest and providing all the excitement and attractions of a Christmas environment without the extreme cold or difficulties of travel.

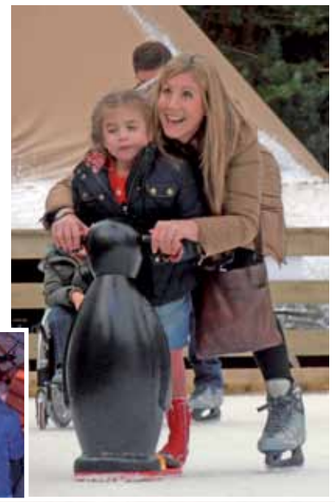
The day was magical from start to finish. The elves helped to keep the magic alive, with their energy and stories and there were lots of exciting activities on offer, from helping Santa in the toy workshop building teddies and puppets, to listening to Mother Christmas's stories, munching on gingerbread men that the children decorated beforehand and helping one of the elves earn his Elf Bell.

Families were then given some free time to experience the mystical landscapes, meeting real husky dogs and having their photos taken with one of their reindeers. Ice skating was also on offer and those in wheelchairs were able to go on the ice too. A warming restaurant was offering hot food and beverages and there was a gift shop, traditional sweetshop and a post office for children to write their letters to Father Christmas or post letters they had brought with them. All the shops and restaurants accepted Jingle currency which could be purchased at the elf Jingle Bank.

To end the day perfectly children got to see the big man himself, Father Christmas, who amazingly knew lots about the children, the families and the children's likes and wishes for what they wanted to see in their stockings! Thankfully I think that all the children were on Santa's 'very good' list.

The experience at the venue was extremely well organised as we attended their Superstar Day for children with special support needs. The schedule for this day allowed extra time for the children to join in the activities, smaller groups to accommodate those in wheelchairs and all children were able to go on the ice, even in their wheelchairs and buggies which on an ordinary day would not be allowed. The venue although based in the middle of a forest, was accessible and was truly magical.

Liz Hardy e.hardy@mpssociety.org.uk



Clinical Trial Update

MPS I

Pilot Study of Administration of Intravenous Laronidase (Aldurazyme®) Following Allogeneic Transplantation for Hurler Disease. This study is fully recruited with the principal investigator being Dr Paul Orchard at the University of Minnesota in the USA. Three UK children are enrolled in this study.

MPS II

HGT-HIT-094, - Hunter phase II/III clinical trial. This study will involve the administration of Idursulfase Intrathecally in MPSII patients. This study is recruiting at Manchester Children's Hospital and Great Ormond Street, London and is sponsored by Shire Pharmaceuticals.

MPS III

The Genistein clinical trial will take place at Manchester Children's Hospital and is open for recruitment for children with MPS III type A, B and C. The principal investigator is Dr Simon Jones and the study is funded by the MPS Society's members' ongoing fundraising efforts and grants and donations from international MPS support groups.

MPS IIIA

HGT-SAN-093, a randomised, controlled, open-label multi-centre phase IIb safety and Efficacy study of HGT-1410 (recombinant human Heparan N Sulfatase) administered intrathecally in paediatric patients with early stage MPS IIIA, Sanfilippo disease is recruiting at Manchester Children's Hospital. This study is sponsored by Shire Pharmaceuticals.

MPS IIIB

There are two Natural History Studies open for recruitment for patients with Mucopolysaccharidosis, Sanfilippo Disease Type IIIB. One study sponsored by Shire is underway at Great Ormond Street Hospital, London and the second sponsored by Synageva is recruiting at Birmingham Children's Hospital.

A study to evaluate the integrity of the Blood Brain Barrier in relation to structural brain abnormalities in MPS IIIB patients using cerebrospinal fluid and magnetic resonance imaging sponsored by Synageva and recruiting at Birmingham Children's Hospital.

Synageva plan the first human clinical study using Enzyme Replacement therapy to evaluate the safety and activity SBC-103 in patients with MPS IIIB. This study is planned for initiation in mid 2014.

MPS IVA

Discovering New Biomarkers for Monitoring Disease Progression in Patients with Mucopolysaccharidosis IVA

Efficacy and Safety Study of BMN 110 for Morquio A Syndrome Patients Who have Limited Ambulation

MPS VII

Phase I / II Intravenous Enzyme Replacement Therapy for MPS VII, Sly Disease sponsored by Ultragenyx is taking place at Manchester Children's hospital and is still recruiting.

Gene Therapy
Sanfilippo A Syndrome

REGENX Biosciences, an AAV (Adeno-Associated Virus) gene therapy company, and Lysogene, a clinical stage biotechnology company developing therapies for rare diseases, announced on 5 December 2013 that both companies are entering into an agreement enabling the development and commercialisation of products to treat Mucopolysaccharidosis type III A (MPS III A or Sanfilippo syndrome Type A) using NAV rAAVrh10 vectors.

Sanfilippo B Syndrome

uniQure, a Netherlands gene therapy company, announced on 28 November 2013 that a phase I/II gene therapy clinical trial for children suffering from Sanfilippo Type B Syndrome enrolled its first patient in October 2013. The clinical trial is being conducted in Paris and involves:

- Institute Pasteur (trial sponsor)
- Inserm (French National Institute of Health and Medical Research)
- AFM-Telethon
- Vaincre le Maladies Lysosomales (VML).

The gene therapy Phase I/II clinical trial for SF Type B, or MPS III B, is based on the development of a viral vector that delivers the mutated gene for the appropriate Lysosomal enzyme deficiency, to the patient's brain cells.

"Cells incorporate the missing gene, provided by the viral vector, into their DNA thus enabling them to produce the missing enzyme. The treatment consists of several intracerebral vector deposits in several areas of the brain. It was administered to the first patient in October 2013".

Also, uniQure in November 2013 announced the filing of a confidential submission of a draft registration statement on Form F-1, to the US Securities and Exchange Commission (SEC), relating to a possible initial Public Offering of its ordinary shares.

In Europe, the EMA (European Medicines Agency), has given regulatory approval to uniQure, to sell its gene therapy, Glybera, for the treatment of the rare disease Lipoprotien Lipase Deficiency (LPLD). No gene therapy has ever been approved for the sale in the United States.

Christine Lavery
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Regenx Biosciences and Lysogene enter into exclusive license agreement for development of treatments for serious, rare lysosomal storage disorder using NAV rAAVrh10 vectors

December 5, 2013 – Regenx Biosciences, LLC (Regenx) and Lysogene SAS (Lysogene) announce that they have entered into an agreement enabling the development and commercialisation of products to treat mucopolysaccharidosis type IIIA (MPS IIIA or Sanfilippo syndrome Type A) using NAV rAAVrh10.

Under the terms of the Agreement, Regenx granted Lysogene an exclusive worldwide licence, with rights to sublicense, to Regenx's NAV rAAVrh10 vectors for treatment of MPS IIIA in humans. In return for these rights, Regenx receives payments in the form of an up-front payment, certain milestone fees and royalties on net sales of products incorporating NAV rAAVrh10.

"We believe this exclusive license agreement will enable Lysogene to advance the development of its NAV based treatment for patients with MPS IIIA," said Ken Mills, President and CEO of Regenx. "As a leader in gene therapy, we are pleased to be formally collaborating with the Lysogene team that, by the successful completion of a recent Phase I/II trial, demonstrates outstanding expertise, resources and commitment to patients. Providing partners with access to our NAV technology further advances Regenx's mission to enable the development of successful new AAV therapeutics."

"Lysogene is a leading clinical stage gene therapy company committed to the development of breakthrough therapies in rare diseases. The company successfully completed a phase I/II study (NCT01474343/ EudraCT2010-019962-10) using the NAV rAAVrh10 technology in Sanfilippo syndrome. We are very pleased to enter into this agreement with Regenx, which we believe offers us the best path to expeditiously advance the clinical development and commercialisation of our lead product for patients with Sanfilippo sydrome", said Karen Aiach, Founder, President and CEO of Lysogene.

For more information regarding Regenx, please visit www.regenxbio.com.

For more information about Lysogene, please visit www.lysogene.com

Clinical trial of gene therapy for MPS VI - a severe lysosomal storage disorder being developed

Mucopolysaccharidosis VI (MPS VI, or Maroteaux-Lamy syndrome; is a rare lysosomal storage disease caused by deficient activity of arylsulfatase B (ARSB). MPS VI is characterised by growth retardation, corneal clouding, cardiac valve disease, organomegaly, skeletal dysplasia, without central nervous system involvement. Thus, systemic therapies targeting peripheral organs have the potential to fully correct the MPS VI phenotype. Enzyme replacement therapy, the current treatment for MPS VI, requires weekly infusions of a costly enzyme and has limited efficacy on bone and corneal disease.

Based on the encouraging preclinical results generated by our group, gene therapy based on a single intravascular administration of adeno-associated viral (AAV) vectors targeting liver has the potential to provide a lifelong source of ARSB.

The MeuSIX consortium plans to conduct a multicentre phase 1/2 clinical trial to investigate the safety and efficacy of AAV-mediated gene therapy in patients with MPS VI. An orphan drug designation (ODD) has been obtained from both the European Medicinal Agency and the US Food and Drug Administration for the MPS VI therapeutic AAV vector. The results from this clinical trial proposed by the MeuSIX consortium has the potential to have a tremendous impact on the natural history of MPS VI and to significantly improve the quality of life of the affected patients. Moreover, the approach developed may facilitate the development of similar approaches for other inborn errors of metabolism.

I am very pleased to have been invited to join the Ethics Advisory Group, one of the work packages for this study led by Bioethicist Prof. Jan Helge Solbakk from the Centre for Medical Ethics, Oslo. Norway.

Christine Lavery
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Third rock Ventures Launches Therapeutics to Develop Life-Changing Gene Therapeutics for CNS Disorders

On February 12, 2014 Third Rock announced the formation of Voyager therapeutics, a gene therapy company developing life-changing treatments for fatal and debilitating diseases of the central nervous system. Voyager's adeno-associated virus (AAV) approach to gene therapy has the potential to transform treatment for a wide range of CNS diseases with one-time therapies that may dramatically improve patient's lives.

In their press release Voyager states that they are a company committed to advancing the field of AAV gene therapy by innovating and investing in areas such as vector optimisation and engineering, dosing techniques, as well as process development and production. Mark Levin, interim Chief Executive officer of Voyager and Partner of Third Rock said "Our world-class founders have driven significant advances in their respective field, and the combination of their deep clinical and scientific knowledge, our experienced management team and Third Rock's investment positions Voyager to deliver breakthrough therapies to patients suffering from devastating CNS disorders".

Update on the ground-breaking Genistein clinical trial for MPS III, Sanfilippo disease

January 2014 - With thanks to the support from the international MPS community and the amazing fundraising efforts of MPS families affected by MPS III, Sanfilippo, we are now only €120,000 short of our €650,000 target.

What is MPS III?

MPS III Sanfilippo disease is an inherited disease caused by the lack of an enzyme that breaks down complex sugars. Sanfilippo disease results in progressive neurological deterioration, severe behavioural difficulties and a greatly shortened lifespan due to storage of these complex sugars in the brain. No treatment currently exists for Sanfilippo children and young adults, aside from palliative care, therefore there is an urgent unmet clinical need.

What is Genistein Aglycone?

The University of Manchester have trialled a drug called Genistein aglycone at a high dose and it reduced the amount of complex sugars stored in the brain of mice with Sanfilippo disease, much improving brain function. A compound of Genistein is found in soy foods, it is non-toxic, can be taken by mouth and is relatively cheap. Before purified Genistein aglycone can be prescribed for children with Sanfilippo disease at a higher dose it must be assessed in a double blind placebo controlled clinical trial of children with Sanfilippo disease to determine the correct and safe dose, the effect of the drug on delaying the progression of the disease and how it improves symptoms.

Who will be affected by this treatment?

At present there are approximately 130 individuals in the UK living with Sanfilippo disease A, B and C. With no current treatments available the outlook is bleak. If successful, this treatment will be available to both UK and worldwide sufferers today and into the future.

Where and when will the Clinical Trial be carried out?

The Clinical Trial for Genistein aglycone will be carried out at the Manchester Children's Hospital. The trial opens for recruitment from January through to Spring 2014.

What does the clinical trial involve?

24 children with MPS III Type A, B and C will be recruited. The clinical trial is for 1 year duration with 1 year extension and will not involve any crossover. The total duration of the clinical trial is 42 months.

How will funding this clinical trial help other Sanfilippo children?

At the end of the trial the results will be analysed and published. If the results demonstrate safety and significant benefit of Genistein aglycone, the evidence would be presented to regulators for marketing approval, providing an essential treatment whilst Gene Therapy is developed.

Our fundraising target

In 2012 in collaboration with Dr Brian Bigger of the University of Manchester, the MPS Society began fundraising €650,000 to fund a clinical trial of high dose oral Genistein aglycone in Sanfilippo disease. Although originally the cost of the trial was €800,000 the MPS Society working with Dr Bigger was able to reduce the cost of the trial to €650,000.

It is truly amazing the support received in our quest to raise €650,000 to fund the Genistein clinical trial in Manchester.

We are very grateful for the fantastic support of a small number of affected families in the UK, the National MPS Society, grants pledged from trusts and contributions being raised by the MPS Societies around the world. However, we still need to raise €120,000.

The Manchester Children's Hospital have agreed to start the clinical trial to help the children but also subject to the MPS Society raising the rest of the money. If we don't, the trial will run out of money.

On behalf of the Board of Trustees, the UK MPS Society would like to thank its members, Share A Gift, the National MPS Society in the USA, Austrian MPS Society, Hong Kong MPS Society, German MPS Society, Japanese MPS Society, Swiss LSD Patient Organisation, Irish MPS Society, Australian MPS Society, VML France, Lysosomal Diseases New Zealand and the Spanish MPS Society for their support in enabling us to start this important clinical trial.

Please help us to raise a further €120,000.

To donate directly to the MPS Society's Genistein Appeal please visit **http://www.mpssociety.org.uk/research/latest-news/mps-iii-genistein-clinical-trial/**

For more information

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The MPS Society
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Shire Global Charitable Access Programme

The Shire Charitable Access Programme strives to improve access to Shire's medicines including Enzyme Replacement Therapy where Shire has no plans for commercial representation. The programme is managed centrally and the initial scope of the programme will only include Elaprase for MPS II; NPRiV for Gaucher disease; and Replgal for Fabry disease.

The aim of the Shire Charitable Access Programme is to provide fair and equitable access to their donated medicines. The programme is administered by Direct Relief and will bring the existing 71 global charitable access patients and enrol an additional 20 in 2014. This careful start will enable Shire and Direct Relief to build a foundation in order to test and learn their plan for the mid-term.

As a member of Shire's Global Charitable Access Programme Steering Committee I look forward to working closely with Direct Relief and the Gaucher Association in seeing vulnerable MPS II patients, as well as Fabry and Gaucher patients benefit from donated medicine.

Christine Lavery MBE
Chief Executive

Parents of boy with rare disease pleads for help on World Rare Disease Day

Eight years after Arian Chowdhury was first diagnosed with Hunter Syndrome access to Enzyme Replacement Therapy still eludes the 12-year-old from West Bengal. With the clock ticking, his distraught parents can only hope help arrives in time.

His parents desperately want the Union and West Bengal governments to take steps to ensure something is done for their only child as with each day that passes and Arian goes without ERT, his chance of living a healthier and longer life also fades.

To date ERT is not reimbursed in India. Whilst Arian's physical disease is progressive his mental development is normal. Without ERT Arian's condition will continue to decline and the damage will become irreversible.

Arian's parents are part of the Lysosomal Storage Disorders Support Society (LSDSS) headquartered in New Delhi and in a few weeks Mr and Mrs Chowdhury will be submitting a petition to their Health Minister, Ghulam Nabi Azad.

Arian's father is also seeking a meeting with West Bengal Chief Minister Mamata Banerjee to seek reimbursement of the treatment for his son.

For the other children battling rare diseases in India, Mr Chowdhury envisages the inclusion of rare diseases in the country's health policy.

"Since one of the results of these diseases is disability, these patients should also be mentioned in the Disabilities Act," said the father, adding Arian can't bend his fingers.

Arian's parents say "He can read and is in Class 6. He should be given a chance to live for a few more years. We can't look beyond Arian... if he's gone...we will be lifeless."

Welsh Review of Orphan and Ultra-Orphan Drugs

The Welsh Government announced a review of their appraisal process for orphan and ultra-orphan medicines early in 2013. Mark Drakeford, Minister for Health and Social Services, requested a group to be established to review the appraisal of orphan and ultra-orphan medicines in Wales.

On August 27, 2013 three patient organisations were invited to present oral evidence to the review panel regarding access to funding for treatments, the experience of patients who had experienced delays from the local health boards for processing funding applications and the opportunity to share experiences both good and not so good. The panel also wanted to hear about systems that work well elsewhere and make this happen.

On behalf of the UK LSD Patient collaborative (of the which the MPS Society is a partner), Tanya Collin-Histed, Chief Executive of the Gaucher Association attended the meeting and gave an oral presentation on the challenging experiences LSD patients have had over the past few years accessing Enzyme Replacement Therapy compared to England where patients are able to access these treatments through the LSD highly specialised service if they meet the eligibility criteria. Tanya has now been invited to represent the Collaborative by providing advice and feedback on draft policies, and advising on implantation of the Rare Disease Strategy for Wales.

If you live in Wales and experience problems accessing Enzyme Replacement Therapy for your MPS or Fabry disease, please do speak up and tell the MPS advocacy team.

On Friday 28th February, the Deputy Chief Medical Officer for Wales, Dr Chris Jones launched the Welsh Implementation Plan for Rare Diseases at the Institute for Medical Genetics at the Heath Hospital, Cardiff.

If you are interested in taking part in the consultation, the document is now available on the Welsh Government website at http://wales.gov.uk/consultation/healthsocialcare/disease/?status=open&lang=en

The consultation period of 12 weeks is attached to the plan and should be submitted to the Welsh Government.

Amicus Therapeutics Highlights Data Featured at Lysosomal Disease Network WORLD Symposium 2014

Encouraging Additional Post-Hoc Analyses of Interim Data from Phase 3 Fabry Monotherapy Study 011

John F. Crowley, Chairman and Chief Executive Officer of Amicus Therapeutics, Inc., stated, "At this year's WORLD Symposium we are highlighting data from several of our ongoing development programs. Using our Chaperone-Advanced Replacement Therapy, or CHART, platform these technologies may provide a unique tool set to address some of the major challenges with currently marketed ERT products – enzyme activity and stability; targeting and uptake; and tolerability and immunogenicity. We are also pleased to present further, post-hoc analyses of 6-month data from our first of two ongoing Phase 3 Fabry monotherapy studies, which we believe demonstrate that migalastat HCI is having a positive impact in Fabry patients with amenable mutations."

Updated 6-Month Data from Fabry Monotherapy Phase 3 Study 011

Migalastat HCI monotherapy is being investigated in two ongoing Phase 3 studies (Study 011 and Study 012) in Fabry patients with amenable mutations. Study 011 enrolled a total of 67 patients to compare megalastat HCI to placebo in reducing kidney interstitial capillary globotriaosylceramide (GL-3). Top-line data from the 6 month double-blind, placebo-controlled treatment period (Stage 1) in Study 011 was previously reported. Updated Stage 1 data, including a post-hoc analysis of mean change from baseline in inclusions per capillary as a controlled variable ("mean change in GL-3"), are being presented at LDN WORLD.

Raphael Schiffmann, M.D., M.H.Sc., Medical Director of the Institute of Metabolic Disease, Baylor Research Institute, stated, "As an investigator working on the development of migalastat HCI and other new treatments of patients living with Fabry disease, I was very encouraged to see the post-hoc analysis of Stage 1 data from Study 011. These data clearly show that migalastat HCI has a biological effect in Fabry patients with amenable mutations. We look forward to seeing the 12- and 24-month data from this study later this year."

The primary endpoint in Study 011 analyzed the percent change in kidney interstitial capillary GL-3 inclusions from baseline to month 6 (responder analysis with a 50% reduction threshold). However, the variability and low levels of GL-3 at baseline contributed to a higher-than-anticipated placebo response. Following the unblinding of the Stage 1 data, and while still blinded to the Stage 2 data, Amicus identified a more appropriate way to control for the variability in GL-3 levels in Study 011. A post-hoc analysis of the mean change in GL-3 was deemed appropriate to measure the biological effect of migalastat HCI.

Amicus plans to analyse the mean change in GL-3 at 12 months (Stage 2) in the modified-intent-to treat population as well as subgroup of patients with amenable mutations in a GLP-validated human embryonic kidney (HEK) call-based in vitro assay ("GLP HEK assat"). The Stage 2 results and complete data from the 24-month study, including clinical outcomes measures such as eGFR and proteinuria, are expected during the second quarter of 2014. Amicus remains blinded to the 12- and 24-month data at this time. Top-line data is anticipated in the second half of 2014 from the second Phase 2 study, or Study 012, which is comparing migalastat HCI to current standard of care ERTs.

About Study 011

Study 011 is a 24-month study consisting of a 6-month double-blind, placebo-controlled treatment period (Stage 1); a 6-month open-label follow-up period (Stage 2); and a 12-month open-label extension phase. All patients received migalastat HCI during Stage 2 and the open-label extension phase. Change from baseline in kidney interstitial capillary GL-3 is being assessed by histology in kidney biopsies at the end of Stage 1 and Stage 2.

About Chaperone-Advanced Replacement Therapy (CHART)

The Chaperone-Advanced Replacement Therapy (CHART) platform combines unique pharmacological chaperones with enzyme replacement therapies (ERTs) for lysosomal storage diseases (LSDs). In a chaperone-advanced replacement therapy, a unique pharmacological chaperone is designed to bind to and stabilize a specific therapeutic enzyme in its properly folded and active form. This proposed CHART mechanism may allow for enhanced tissue uptake of active enzyme, greater lysosomal activity, more reduction of substrate, and lower immunogenicity compared to ERT alone. Improvements in enzyme stability may also enable more convenient delivery of next-generation therapies. Amicus is leveraging the CHART platform to develop proprietary next-generation therapies that consist of lysosomal enzymes co-formulated with pharmacological chaperones.

Medical Trial Targets cure for Fabry Disease

Canadian researchers are taking part in research that may result in the first Gene Therapy Clinical Trial in the world for Fabry Disease.

Promising gene therapy results in mice performed in the laboratory of Dr Jeffrey Medin at the University Health Network in Toronto promoted this clinical trial initiative that scientists in Calgary will play a major role.

The team hopes to treat the first human Fabry disease patient in about two years' time after several phases of pre-clinical results satisfy the regulatory requirements of Health Canada.

Research Study: Living with Fabry Disease

insight RESEARCH GROUP
A Cello Health Company

In the latter half of 2013, the MPS Society and Shire Pharmaceuticals collaborated with a market research agency, Insight Research Group, to run a study to gain a better understanding of people's experience of living with Fabry Disease. Interviews were conducted by the MPS Society and analysed by Insight. We explored the impact Fabry has on people's lives, their views on treatment and what support they feel they need. We spoke to sixty people across the UK who had either been diagnosed with Fabry Disease, or whose children had Fabry. We included a mix of males and females of various ages and around 3/4 were on treatment.

What struck us is that there is a broad spectrum of experiences, but we have identified a number of common themes which help to illustrate what it is like to live with Fabry Disease today.

Life with Fabry Disease

The type and severity of physical symptoms of Fabry Disease varies widely. Some people experience virtually no symptoms, but around half of the people we spoke to report severe pain in hands and feet, gastrointestinal problems and fatigue. These symptoms can be difficult to cope with as they can place limitations on an individual's life which in turn impacts their social and emotional wellbeing.

The emotional impact of Fabry should also not be underestimated. Although many of those we spoke to feel they are coping reasonably well, it's not uncommon for others to feel isolated and alone (especially if they have limited family support). Maintaining relationships with friends or partners can be difficult, particularly if fatigue prevents socialising.

The Journey to Diagnosis

The journey to diagnosis typically takes one of two forms. For those who do not have family members who have been diagnosed with Fabry, the journey can be a very long one. The range of symptoms presented to GPs and lack of knowledge about the condition often means it takes years before a referral to a specialist (usually a kidney specialist but opticians also common) who can diagnose them.

Contrastingly, for those with family members who have been diagnosed, the journey can be much faster. Family testing is becoming more common and ensures treatment is initiated more quickly if necessary.

Feelings of shock and despair at diagnosis are common, and it can be hard to deal with learning about what the disease means for everyday life. Conversely, many are relieved to have a diagnosis – finally there's an explanation for the symptoms and they can't be accused of being a hypochondriac.

Treatment experiences

ERT therapy is usually started within 3 - 6 months of diagnosis. After an initial series of infusions at hospital, the majority of the people we spoke to who are on treatment receive home infusions administered by a nurse. Nurses are felt to be flexible and friendly which makes the process easier to cope with. However, 1/3 of the people we spoke to self—administer treatment, which they feel offers them additional flexibility and independence.

Regardless of the method of administration, the vast majority who are on treatment believe it has had a positive impact on their lives and has eased their symptoms. For this reason, people very rarely miss doses of their treatment. Sometimes infusions may be delayed by a few days (e.g. if an individual is very ill), but treatments are rarely missed altogether.

However, not everyone receives treatment. Some doctors do not recommend treatment if symptoms are very mild. However, most are open to the idea of ERT in the future, and will contact their doctor immediately if and when their symptoms start to impact their lives more significantly.

Additional support needed

Although the people we spoke to generally feel well supported, this research has highlighted a few areas where people feel more can be done. For example, some are looking for guidance on how to get travel insurance, while others would like more information on what benefits they are entitled to. This kind of information can be found through the MPS Society.

For those who struggle with the emotional impact of the disease, having someone to talk to would help to ease feelings of isolation and loneliness, as would being in contact with others who have Fabry Disease. Advice on how to explain Fabry to children would also be welcomed.

We would like to say a big thank you to all those who took part in this research for sharing their stories with us, and also to the MPS Society for conducting the interviews on our behalf.

Further information about Insight Research Group can be found at: www.insightrg.com

The Power of Partnerships – The MPS Society in the UK

As one of the first Biotechnology companies in the world with a dedicated patient advocacy function, Genzyme's collaboration with patient organisations has always been a unique and foundational component of our company culture to business strategy. It is our way to ensure that the patient perspective is always top of the mind, and that our work remains focused on the issues most important to patient communities. Employees in the Rare Diseases business recently heard first-hand from Christine Lavery, the head of one of the most respected and renowned rare diseases patient groups around the world, the Society for Mucopolysaccharide Diseases, UK.


Christine Lavery,
MPS Chief Executive

Christine founded the UK MPS Society and has serviced as its Chief Executive since 1993. In 2002, she was awarded the Member of the British Empire (MBE) for services to metabolic diseases. Under her leadership, the organisation has grown dramatically, playing a key role in patient support, policy advocacy, research and clinical trial support. During a recent trip to the U.S., Christine took time out of her busy schedule of discuss the wise range of topics with Corporate Communications, from the key role her organisation plays in providing advocacy and support to patients and their families to her expectations of industry partnerships.

IG: What should the Genzyme employees know about the MPS Society?

CL: The Society has been around for 31 years and it is the only UK charity supporting individuals and families affected by MPS and related diseases. We are the leading provider of information and support for those patients and their families.

With 14 employees, we manage a large research budget and a unique advocacy service that provides support to more than 1,200 children and adults, their families and professionals throughout the UK. The support we provide is quite broad and includes home adaptations, special educational needs, access to new therapies, respite care, palliative care and pre- and post-bereavement support. Our work is centred on partnerships and collaboratively with companies like Genzyme, as well as other specialist centres, government agencies, other patient organisations and medical professionals.

IG: You founded the Society. What were your reasons for doing so?

CL: My son Simon was diagnosed in 1976 at 18 months of age with Hunter disease (MPS II). We were living in Japan at the time and we were trying to learn all that we could about the disease. At the time, there weren't any rare disease groups – I began to do some outreach, and it was really through letters from various families that I was able to connect with people with similar experiences. Simon passed away in 1982, and when I moved back to the UK, I felt obliged to do something. I've always been a very active person, with a particular interest in philanthropy, and it was important to me to try to make a difference for patients living with the disease.

IG: The MPS House is the focal point of your organisation. What's that all about?

CL: We're very proud of our MPS House, which is about a 25-minute drive from Heathrow airport. All of our advocacy and support services are provided from this place, and we also have a conference area and a research library that houses a range of publications and resources on all aspects of rare genetic diseases. We welcome guests to MPS House at any time who need our support or access to our resources.

IG: Can you give us an example of a project the Society is currently working on?

CL: We are always dealing with very complex, personal situations for our families, including reimbursement for products, clinical trial enrolment and even immigration issues. Nothing daunts us at this point. One current project that comes to mind is what we call the 'Transition Passbook' which considers the needs of all patients living with lysosomal storage disorder (LSD) as they transition from adolescence into adulthood. The book, which addresses issues such as health insurance as you move from your parents' coverage to your own, and the importance of adherence, is being piloted at various health clinics across the UK. After speaking with Genzyme employees earlier in my visit, our plan is to make this book more broadly available, as there seems to be similar needs in other disease communities.

IG: What can Genzyme employees do to help the Society in its mission?

CL: What we need is information. Consider us when you have new information to share with the LSD community. We would like for our partners such as Genzyme to liaise with us and be proactive in their outreach. We cross paths with many of your colleagues and competitors, and it's critical for us to stay up-to-date on what's happening so that we can provide support to our families. We all have the same goal of helping patients living with rare diseases and equal access to treatment, and it's an honour to visit Genzyme today to share our work with you.

This interview with Christine Lavery was published on Genzyme's internal website and is reprinted with permission.

Jurgen Zumbro 10.11.1940 - 15.12.2013

Jurgen Zumbro and his wife Brigitte first came into my life in 1983 following the diagnosis of Sanfilippo disease in their only child, Natalie. This was just months after the UK MPS Society was founded.

A first and lasting memory of Jurgen is that of a family man who wanted to make a difference in the lives of all children with MPS and related diseases.

And make a difference he did. In 1985 Jurgen, Brigitte and Natalie travelled to England to take part in the UK MPS Society's third annual family weekend conference at the Post House Hotel, London Heathrow and at a post conference gathering at my home, Jurgen took the decision to formalise an MPS Society in Germany.

Not only was Jurgen supported by the UK MPS Society and Austrian MPS Society but also Professor Michael Beck and the late Professor Ed Wraith. Over the next few years UK MPS families, Professor Beck and Professor Wraith joined the growing army of German MPS families for their annual family conferences.

In 1993 Jurgen led the German MPS Society in organising a very successful Second International Symposium on Mucopolysaccharide and Related Diseases in Essen.

Jurgen and Brigitte ran the MPS Society in Germany for close to twenty years and sadly lost their beloved Natalie in 2009 aged 31 years.

My fondest memories are of a man with an enormously generous heart who welcomed my husband Robin and our children into his home as he did many of the MPS families at that time.

Jurgen, Brigitte and Natalie made many visits to Australia and it was their life's dream to retire to the sunny East Coast. Despite every effort by the family supported by the UK and Australian MPS Societies it wasn't to be. I am sure I speak for many in mourning the loss of a man with a very generous heart, Jurgen Zumbro, just 10 days before Christmas last.

On behalf of all those who knew Jurgen our thoughts are with his wife Brigitte at this very sad time. **Christine Lavery**

10th annual WORLD Symposium 2014

10 - 13 Feb 2014, San Diego, California

Again we should congratulate Dr Chet Whitley and his team at the University of Minnesota for organising an outstanding WORLD Meeting on Lysosomal Storage Diseases. Close to a thousand scientists, clinicians, geneticists, pharmaceutical industry representatives and patient organisations gathered at the Manchester Hyatt Hotel for three days of lectures and networking solely dedicated to the Lysosomal Storage Diseases. This is a unique meeting that enables people from across the world to present their work and their findings through short oral presentations and poster sessions. It is a meeting where people can explore new ideas and network to their hearts content.

From the MPS Society's point of view being at WORLD facilitated the opportunity to meet with all the different pharmaceutical companies and biotech companies with an interest in our diseases. It was at WORLD that we learnt that BioMarin have a pre-clinical pipeline drug for MPS IIIB. More information on this and other new developments can be found in the research and therapies section of this Magazine.

The MPS patient support community plays an important role in the world of LSDs. During this meeting I was able to catch up with my counterparts from the USA, Canada, Spain, France and Brazil.


Christine Lavery (UK MPS Society), Barbara Wedehase (National MPS Society) and Kirsten Harkins (Canadian MPS Society)

Excellence in Paediatrics

2013 Doha, Qatar, 4-7 Dec 2013

The MPS Society's Chief Executive, Christine Lavery was invited by Excellence in Paediatrics (IEP) to attend a forum on the diagnosis and treatment of patients with LSDs. Christine also gave a plenary talk in EIP's Middle East and North Africa LSD Session offering an insight into 'Living with an MPS or Related Disease' from a patient organisation perspective on . The overall objective of this meeting was to bring together interested paediatricians and physicians from all corners of the Middle east and North Africa to foster networking and develop an understanding of LSD experiences outside the region.

One of the difficult areas of discussion was around treatment as access to Enzyme replacement in this region is very varied with some countries providing ERT through Government reimbursement schemes and other countries not funding ERT at all. Another challenge for the region is diagnostics, genetic counselling, raising awareness of these diseases and critically lack of and need for patient support and advocacy. Christine's presence highlighted the need for clinicians and the patient community to work together to improve clinical and social outcomes for the LSD community.

Carrier testing for Siblings of those affected by MPS

Over the last ten years we have seen the number of adult brothers and sisters whose sibling(s) died from a Mucopolysaccharide disease carried by both the parents seeking carrier testing rise considerably. In the case of MPS II, Hunter disease where the condition is usually carried by the female and passed to the male other female relatives are also seeking support.

To put things into perspective when carrier testing for the main MPS diseases became available over twenty years ago ethical considerations were less stringent than they are today and many brothers and sisters were tested as children in the lifetime of their affected sibling. My own daughter was tested at the age of eight years and some 12 years after her brother died from Hunter disease.

More recently carrier testing in children has not been encouraged and we now have a generation of young people growing up, settling into a relationship or considering a pregnancy or indeed are pregnant and then the carrier question arises.

Our members' experiences in obtaining carrier testing years after their siblings have died have on the whole been less than optimal and in some cases almost cruel. The majority of siblings asking for support from the MPS Society have been seeking carrier testing for MPS II Hunter and MPS III. It should be said that if the two disease causing mutations (one in MPS II Hunter) are not known and there is no DNA material from the affected person carrier testing may not be as straight forward but is usually not insurmountable.

Based on recent experiences we have some suggestions:

Adult Siblings

• If you want to be tested for carrier status for a MPS disease plan to do this some time before you plan a pregnancy

• Talk to your GP

• Consider whether you (and your partner) would benefit from seeing a genetic counsellor. This appointment should be timely and the information non directive

• If you are advised testing is not available you will have to pay; you have to go through genetic counselling and do not want to, or the time scale is not realistic please call the MPS Society. Some siblings have found it helpful to speak to the MPS Society first.

Parents of MPS children today

The diagnosis of a much loved child with an MPS disease is understandably a very difficult time but as soon as you feel able make sure you receive a letter from your child's consultant detailing the disease causing mutation(s) if known in your affected child(ren). Keep this information in a very safe place so that years from diagnosis any unaffected children in the family may have the code (mutation analysis) to unlock their carrier status if they wish.

If you or your son or daughter have recently gone through carrier testing and feel able to share your experiences in the MPS magazine please do send in your article.

Christine Lavery
c.lavery@mpssociety.org.uk

New resource for children!

'Dr DNA's guide to genetic science'

The MPS Society is excited to develop a children's guide to understanding genetics called ***'Dr DNA's Guide to Genetic Science'***.

It explains simply and clearly to siblings, friends and affected children themselves, how and why some children may not be able to run as fast, or hear as well because of genetic conditions but how deep down we are all the same.

The book takes the reader on a genetic journey with Dr DNA explaining how the human body works and the role DNA plays. It is bright and colourful with simple to understand cartoons and diagrams.

As well as being used by our members and supporters, the book will also be available to schools (with a special section for teachers) that ask for further information as a guide to help promote understanding and awareness of genetic conditions.

The MPS Society believes it is very important that families, friends, relatives and the wider community are able to understand the nature of rare diseases and over the years the MPS Society has created a comprehensive and widely used library of publications that provide detailed information on the 24 rare genetic diseases that we support.

Thank you to the Hospital Saturday Fund for kindly supporting this project.

Fundraising

Fast approaching is one of the most important days in the MPS calendar – MPS Awareness Day on May 15th – so get ready to dust off that blue feather boa, break out those walking boots and get ready!

We would love to make this year's MPS Awareness Day the most successful yet, and that of course, depends on your support. As a charity for rare genetic diseases we do not get the exposure of bigger charities, and as a result we are still not well enough known, so please help us gain some attention by raising awareness for the MPS Society.

Although it is a cliché, it is true to say that every penny counts, so no matter how small your event or donation, you are still helping to further our aims of support and research. We have helped to fund labs which have developed treatments for these diseases which simply did not exist before, but of course we would like to do more, and that is why fundraising is so important.

We would like to thank everyone who has fundraised for us. On the following pages you will find some of your stories and further information on fundraising. In addition to this please visit our website for a whole host of exciting ideas for your own events.

However you decide to fundraise, let's make it big and bold and let everyone know all about the MPS Society!

Wear It Blue fundraising packs

Phone 0845 389 9901

Email fundraising@mpssociety.org.uk

Visit www.mpssociety.org.uk

Wear It Blue for MPS Awareness Day

Thursday 15th May 2014

Coming up is the most significant day in the MPS calendar, a day for all the individuals and families who have been affected by MPS diseases, but also a day to celebrate the progress made by the dedicated scientists who have been engaged in researching treatments for these diseases. It's a day we can look to the future and the continuation of this progress.

We want MPS Awareness Day to be as big and memorable as possible, so please help us to mark this day. Whether you want to Wear It Blue or go for a Wicked Walkabout, or even just make a donation via our website, there is a way everyone can help raise awareness for this special cause.

Donate €1 and Wear it Blue for MPS Awareness Day on 15th May!

Think peacock! Think Avatar! You can even think Smurf! Just as long as you think blue! Whether it's cobalt or azure, hunt down that blue item of clothing from your wardrobe and wear it to work, school or university for the day, and please encourage your classmates, colleagues or students to participate and spread the word. Anything from blue face paint to blue socks will do, just make sure you send us your photographs!

The premise is simple – encourage people to pay a €1, wear blue, help save lives. Easy.

Order your posters and leaflets by emailing us at **fundraising@mpssociety.org.uk**

And if someone asks why you're dressed as a smurf, please take the opportunity to tell them all about the MPS Society!

Visit **www.mpssociety.org.uk** for more information or give us a call on 0845 389 9901 to explore the different ways you can get involved.

Your fundraising stories

Cambridge Mill Hill Road Winter Fair

It was early November and I asked my friend Graham if he was doing anything for the Cambridge Mill Road Winter Fair. He had nothing planned. I asked him if I could use the space outside his shop Cavendish Classic Footwear to do a fundraiser for the MPS Society. He asked for some information on the charity before he could say yes, so I gave him the MPS Magazine and leaflets to read.

After the weekend Graham and his wife Julie said it was a good cause and would be to happy to help raise funds. In previous years Graham and his staff had dressed up to raise money for different charities, so we thought we could also get some costumes. Santa and Dancer the reindeer accompanied by a snowman were coming to town!

On Saturday 7th December it was time to pick up Jamie (the snowman) and Amanda. With decorated buckets, MPS balloons, leaflets, and tinsel, we headed for the shop. Zack my grandson who has Hunter's was coming to see us later with his nanna. We blew up some balloons decorated the gazebo and got into our costumes.

Amanda and other helpers joined us through the day. All the children loved Santa. We met Puffles the dragon, had our photo taken with him and put this on Twitter. We managed to get a live interveiw on Radio Cambridge 105 and had a fantastic day,giving all the balloons and leaflets away. We raised £186.31 on the day. My partner Angela made it £200 and my relative Pam gave me £5.Thank you Graham, Julie, Jamie, Amanda, Angela and Eddie helping to raise £205.00.

Mark Hughes

Thank you to Marina and Friends

We would like to extend a special thank you to Marina Foster and friends. Marina runs a charity shop in Bristol, Marina and Friends Fundraisers, donating the proceeds from the sale of second hand items to the MPS Society. If you would like to support the MPS Society by providing items for Marina to sell, please find below the address for the shop: Marina & Friends Fundraisers, 44 Sandy Park Road, Brislington, Bristol, BS4 3PF. You can also follow Marina and Friends Fundraisers on facebook.

MPS National Draw 2013 Results

Thank you so much to all those who purchased MPS Raffle Tickets. The prize winners are listed below:

The support that we receive is invaluable in enabling us to continue our work supporting individuals and families affected by MPS, Fabry and related diseases and funding research into treatments.

1st: £350 Marks & Spencers Vouchers - Dearden No. 07469

2nd: 2 night stay at Birmingham Copthorne Hotel for 2 – Browning No. 06241

3rd: £100 Marks & Spencers Vouchers - Thompson No. 13905

One night stay and breakfast at Brent Cross Holiday Inn - Stump No. 21973

One night B & B at Copthorne Hotel, Manchester - Cooney No. 18997

£50 M & S Vouchers - Marquis No. 21592

£50 Robert Dyas Vouchers - Peach No. 13750

£50 Next Vouchers - Kershaw No. 03792

£25 Argos Vouchers - Lane No. 15920

£25 Tesco Vouchers - Martin No 13780

£15 Tesco Vouchers - Brown No.17616

100ml Christalle Eau Verte by Chanel - Wray No. 00207

Selection of Quality Street, Roses and Thornton's chocolates - Wheeler No. 00006

Selection of Quality Street & Roses - Todd No. 14521

Knight Frank' Day of Giving

Knight Frank

For the first time the Knight Frank partnership created an annual Knight Frank Day of Giving which saw all Knight Frank departments and offices around the UK raising money for their 5 chosen charities on Thursday 10 October 2013.

They hoped to raise as much money as possible, in as many different ways as possible challenging each department and office with new and innovative fundraising ideas.

The MPS Society was one of the five recipient charities and we were delighted to receive £7488.48 from Knight Frank. We would like thank everyone at Knight Frank and their customers for supporting this event.

Haddenham Mummers raise £680 for MPS

Over the Christmas period, the Haddenham Mummers performed their short seasonal play which tells of good overcoming evil in various places such as local pubs, the Chiltern Open Air museum and Aylesbury Town Centre. After each performance they took a collection in total raising about £2000. They shared the takings between three charities including the MPS Society. MPS was delighted to receive £680.

Run, cycle, jump for MPS - Please email us at **fundraising@mpssociety.org.uk** to register your interest in future running and cycling events or check out the fundraising section of our website **www.mpssociety.org.uk** for the latest news. We also advertise our places on **Facebook** and **Twitter** so keep checking these sites out too!

Good luck to all our runners in the Virgin London Marathon 2014. Thank you for all your fantastic support in fundraising for the MPS Society - have a wonderful day!

Events and Challenges

Adam Painter's Great North Run 2013

Whilst I am certainly one for a challenge, the task that I was about to embark upon became the hardest yet most rewarding of my life.

Stepping out cautiously in June I had no idea how far I would go, or indeed what I was doing. Although I'd call myself healthy and fit I'd never really ran before and usually used the treadmill to catch up on the news! There I was, excited and nervous in equal measure, I planned a route and just ran. Everything seemed to be going so well until I suddenly had to stop, I was gasping for breath and every muscle was hurting. When I looked and saw that I'd barely ran a mile I knew how far I had to go.

When such a challenge is placed before you it can be daunting. However, the challenge facing my Mum (who has Fabry disease) and for all those that the MPS stands up for is much greater. I had this in my thoughts every time I ran; they dragged me out of the door, pushed me further and focused my mind on the enormity of my task. From wearing a scabby old vest and poor shoes, I bought running shoes and socks. Moreover, I trawled the sports stores looking for equipment and researched all the techniques and diets that I needed to apply. As each week went by I ran further, I'm not entirely sure how - practise I guess but this filled me with exuberance.

Through all of this - and a chest infection - I had only the race day in mind. When it came the pressure was immense. However, the atmosphere upon approaching the venue was astounding. I'd purchased my flapjacks, had my delicious 6am pasta and remembered all of my race information. Then, only one hour before the race I'd lost my headphones! How would I race without them? It seemed like this and the weather meant that the day would work against me. That being said, the feeling at the start line was incredible; everybody encouraged each other as we warmed up and I discovered muscles I never knew I had.

The whole race came down to focus, I couldn't even think about it until 4 miles in. As the rain pelted against my frozen face it almost became insignificant. Everybody was together, together in representing charities, together in making a difference and together in remembering those who they loved and lost.

At 10 miles I began to think that there was no way I'd finish. Nevertheless, despite the worst pain I'd ever felt I pushed and focused on the next step, the next water point. The cheers coming from thousands of people along the route gave you that extra mile. I'd never felt anything like it before. As I approached the finish and saw the South Shields coast reveal itself on the horizon the enormity of what I'd done began to sink in. Of course, I wanted to finish but the fact that I knew who I was representing and why I was running the race made it all the more special.

Holding my medal in my hand and limping back home I simply hoped that my small contribution could go some way to making someone's life just that little bit better.

Time: 2:28:29 Raised: €180

Running vests now available for all MPS runners.
Please contact us, small, medium and large unisex sizes
fundraising@mpssociety.org.uk

MPS Society Challenge Event Opportunities for 2014

BUPA Great Manchester Run – 18th May 2014
Help us mark MPS Awareness Day and make the most of this fantastic fundraising opportunity!

The nation's favourite 10k running event, the Bupa Great Manchester Run, has seen over a quarter of a million enthusiastic runners pound the streets of Manchester since its first staging in 2003.

First held in May 2003, the run was created as a legacy event following the 2002 Commonwealth Games. The Bupa Great Manchester Run has since grown into a weekend festival of sport, including mini and junior events and the Powerade Great CityGames (link to city games site) – a unique elite event that consistently brings together some of the world's top sprinters on a purpose built track in the heart of the city centre.

BUPA Great North Run – 7th September 2014
The Bupa Great North Run is the premier event in the Great Run series and firmly established as Britain's biggest participation event.

From just 12,000 runners at the very first staging in 1981, the event has now grown to a record 55,000 accepted entrants from over 100,000 applicants in 2012.

Starting in Newcastle, the course takes in the iconic Tyne Bridge, goes through Gateshead passing the famous international athletics stadium and finishes in the coastal town of South Shields. Live music, on course refreshment and thousands of cheering supporters will keep you motivated every step of the way.

BUPA Great South Run – 26 October 2014
The fast and flat 10 mile route takes in the iconic sites of the Portsmouth Historic Dockyard including Portsmouth Cathedral, Spinnaker Tower and the HMS Victory- which has played host to the Band of HM Royal Marines, who often perform for passing runners at the site. The final flat stretch along the sea front has given thousands of people the opportunity to get a personal best time for more than twenty years.

British London 10k Run – 13 July 2014
The British 10k London Run is staged on the world's greatest road race route through the heart of central London passing many of the capital's truly world class historic landmarks.

Do It for Charity London to Brighton Cycle – 7 September 2014
The London to Brighton cycle will start in South London, travel through Mitcham, Carshalton, Chipstead, Banstead, Haywards Heath and finally to Brighton having completed 54 miles!

Virgin London Marathon 2015
Our guaranteed MPS charity places for the Virgin London Marathon 2014 have been fulfilled. If you would like to register your interest for our 2015 places, please let us know.

Other opportunities – subject to interest

Three Peaks Challenge 2014
In 2014, the Open Bus Three Peaks Challenge will run on the 7th June, 21st June, 12th July, 2nd August, 23rd August and 13th September.

For more information and to register your interest for any of the above events please email **fundraising@mpssociety.org.uk**. Available places will be allocated shortly.

Even if you are unsuccessful in obtaining a charity place, for some of these events you can still enter yourself as an individual and fundraise in aid of the MPS Society.

Our support to you
Fundraising packs and MPS T-shirts and/or running vests will available to all our challenge events participants for 2014. We will keep in regular touch with you to provide advice on fundraising and to help you make the most of the sponsorship opportunity including publicity support and advice and email updates.

If you have an event you would like to suggest
If you know of an event that you would like us to consider obtaining charity places for, please drop us an email at **fundraising@mpssociety.org.uk**.

Give As You Live

Shop online with Give As You Live. Thousands of retailers have signed up to donate to the MPS Society a percentage of every online purchase you make, without adding to the cost of your shopping. For more information and to start shopping head to **http://www.giveasyoulive.com/join/mps-society/63954/150009**

Thank you to all our donors including . . .

Garth & South Berks Club donated £500 as committee member, Kirsty Adams knows someone with MPS.

Peter Archard donated £10 in lieu of sending Christmas cards.

Pat Rowan donated £20 in memory of her son Denis.

The Solihull & District Orchid Society raised £108 with a charity raffle held at the British Orchid Growers Association Orchid Fair.

Dukinfield Methodist Church Social & Entertainment Group raised £225 being the proceeds of their charity raffle.

Ian Woodman donated £25 in support of Jacob Carter.

Mark Hughes and friends Graham and Julie Gardiner raised £205 with a charity collection at the Mill Road Christmas Fair.

Barbara Arrowsmith donated £50 in memory of her son Colin.

Edna Wallace donated £50 in memory of her grandson Colin Arrowsmith.

Mathew Stevens raised £745 with a Do It For Charity London to Brighton cycle in support of his nephew Sam who has Sanfilippo MPS III.

Dixons City Academy, Bradford raised £300.

Sharon and Darron Allen donated £50 in memory of their son Daniel.

Joan Crispin donated £40 to MPS III research.

Staff at Towergate Insurance, Leeds raised £100 for the Society.

The team at BioMarin Wore it Wicked and raised £48.46.

Elizabeth Butler donated £25 in support of her nephew.

Maria Yanev donated £10 in support of Lizzie and Jacob.

Val Lucas donated £50 in thanks for the support the MPS Society has given to the Hiller family following Joe's diagnosis with Hunter's.

Vanessa Stottor sent in an additional donation of £24 from her Evening of Inspiration.

Shire HGT held a Wear It Blue event for MPS Awareness Day raising £151.97.

Karen and Andrew Weedall donated £51 being commission from a Webb Ivory order.

Ann Parsons has sold MPS trolley token keyrings at Asda Eastbourne raising £106 for the MPS Society.

Paul Whitworth raised £225 by organising a fundraising Raffle at a Charity Production held in November 2013, this amount will be matched by Lloyds Banking Group.

Strictly Social Dance Club held a Raffle at their monthly dance and raised £210.

Family and Friends of Jean Armsby donated £500 in the memory of her daughter Julie Bennett at her recent birthday and retirement lunch.

Jude Butler raised £135 from a tombola and Christmas card collection at her work for the continued support of Jacob Carter.

Farringdon Community Academy raised £171.50 from a 'Wear a Christmas Jumper' non-uniform day.

Marina Foster and Friends have raised £186.74 for research into Sanfilippo.

Michala Mawdsley and Affinity Water raised £1438 for Genistein trial.

Doug Cridland along with Chorley Council raised £127.92 with a dress down day.

Mr Robson Brown donated £75 for his participation in market research and donated the money towards Hadrian's Wall Cycling Challenge.

Stuart Garwood along with members of Bowen Lodge in Beaconsfield raised £300 over the past year.

Ben Hope-Gill donated £50 in support of work the MPS Society does and towards James Hope-Gill running the London 2014 Marathon.

The Purely Recruitment Company donated £300 to MPS as a close friend of one of the directors is personally affected by MPS.

Marilyn Eggleton has sent in used postage stamps, coins and jewellery for recycling along with a donation of £6.28 from all the coins found on the street.

Kath Hiller made a donation of £100.

The Hampden Arms held their usual Boxing Day quiz, once again to a packed house. Ian Evans gave a wonderful, moving speech about the support the MPS Society has given to his family and others and they raised £253 for the MPS Society.

Mr Tom Walker and Mr Hugo Machin of AMP Capital Investors undertook some market research and donated £150 from Makinson Cowell to the MPS Society.

St Hilda's C of E Primary School chose to give donations to the MPS Society rather than buy Christmas cards.

Dinah and Damien Adair hosted a meal cooked by 2008 Masterchef finalist Jonny Stevenson and raised £1020 for research into Morquio.

St Bernadette Catholic Secondary School held an annual Charity Day during which they raised £171.45 for the MPS Society.

Ravjit Dadi ran the marathon back in 2009 and raised £150 in the name of his brother who suffered from Fucosidosis.

The Edgecombe Group raised £70 from a dress down Friday, inspired by Marina's hard work.

Kath Hiller donated £100 comprising £26 from her sister-in-law (Diane Painter)'s sale of teddy bears at a Christmas Craft Fair and the rest is from the collecting tin at the MPS Christmas Tree in the Bridport Christmas Tree Festival.

Elizabeth Heath donated £647.30 to the MPS Society and £5 cash from the closure of her son, Jack's, account.

Graham and Margaret Moore raised £150 for the MPS Society being the proceeds from their annual Phoenix Card Party.

Sigurd Dreyer kindly donated £2850 to the MPS Society in memory of the grandfather of Ola Dreyer (MPS II).

The Mayflower Pub in Hazlemere held a pub quiz raising £220 for the MPS Society.

Marina Foster and volunteers at Marina and Friends charity shop in Bristol donated £7402.61 raised from the sale of second hand items. The cumulative total is £114,694.27.

Mr R Ehrmann kindly donated £1000 to the MPS Society in honour of Oliver Gosling.

Andrew and Vivienne Culley donated £270 being the proceeds from the sale of personal items at auction.

MPS received £200 from an Asda 'Chosen by You' nomination. A colleague has nominated the MPS Society after seeing a fundraising campaign for MPS run by Luke Blignaut's family and friends.

Evelyn Jarvie sent in £550 being the proceeds of monies collected at the funeral of her son Andrew. Our thoughts are with the family at this time.

The Terry School of Dance held a ballet school show at Gryffe High School, Houston, Scotland in November 2013 raising £500 for MPS. One of their first pupils was Joanne Evans who has Morquio and her mother Judith is still involved with the dance school, processing all the tickets for the performances!

Jenny and Andy Hardy held a Christmas card sale which raised £211 for MPS.

Jennifer Warren ran in the Oxford half marathon in October raising £300 for the MPS Society. This is being matched by her employers, Thomson Reuters.

Matthew Stevens raised a total of £745 for the MPS Society for completing the DIFC London to Brighton Cycle 2013. His nephew Sam has Sanfilippo disease and his other uncle, Alexander Watts, also completed the ride for MPS as did a family friend Stuart Knipe who also cycled all the way from Dunstable to the official start.

We would like to thank all our fundraisers and supporters who kindly set up online pages to collect donations via **Justgiving.com** and **Virginmoneygiving**. Once your event is over and you have finalised the amount you have raised, we would love to feature articles and photos about your wonderful fundraising. Please email them to **fundraising@mpssociety.org.uk**.

A Special Thank you

Fundraising often mean getting as many people involved as possible. One of the easiest ways you can do this is by promoting the MPS Society at your work or place of education. This can prove to not only be an effective way to raise cash and awareness, but as can prove to be lots of fun!

Fundraising at Work

We are always striving to raise awareness for our cause, and one of the ways that you can help us do this is by talking to your employer or employees about fundraising for the MPS Society. Not only do companies want to help charities like us, but it is also a great way to raise their profile, and for those of you who need an excuse to dress up in the office, it is a perfect opportunity!

If you would like to fundraise and raise awareness for the MPS Society, please do send an email and we will be happy to support your fundraising ideas, or to provide inspiration if needed.

Fundraising at University

Calling all students, lecturers and professors! We would love to be nominated as your university's charity partner, so please do speak to your RAG team about the MPS Society. We would be absolutely delighted to support any student fundraising so please drop us an email (see below) for more information or to let us know of a nomination. This kind of support is vital for the continuation of our work.

Most UK universities have amazing RAG committees (Raising and Giving) who manage to raise millions of pounds annually for UK charities through their fundraising efforts, which includes everything from sponsored waxes to blind date evenings! Keep up the good work!

Fundraising at School

Fundraising in aid of charities is a regular activity in most schools. It is a great way to teach children about the different causes and concerns that affect different people and to help foster a feeling of pleasure when helping others. Fundraising activities also develop a child's organisational and creative skills as they work together to discover a fun and effective way of raising money. The awareness of the cause does not stop at the classroom door, but parents will also learn of the charity in question, and perhaps even get roped in to helping fundraise!

If you think that your local school might be interested in supporting the MPS Society and spreading awareness for a small rare disease charity – especially with the upcoming MPS Awareness Day on May 15th – please do drop us an email.

For any questions regarding fundraising, or to request a fundraising pack, please send us an email at **fundraising@mpssociety.org.uk.**

Grants, CharitableTrusts, Grant-giving Foundations and Community Groups
Lloyds Community Fund; JTH Charitable Trust; City Bridge Trust; The Hobson Charity; The Adamson Trust;

Donations
Mr R Ehrmann; Niall & Dermot Devlin; Mandy & Alan Playle; Sue and Vic Lowry; Michael J Newell; David Tonge; Jaspreet Chandi; David Boothe; Mc Grattan Family; Mr and Mrs Kelly; Moira and Keith Darke; Janet Hillier; Mrs Baker; Miss E Jenkins; Marlene Murty; Diane Peirson; Mr and Mrs Watts; Mrs Gardner; S E Skipper; Pam and Ken Ballard; Sacha Knight; Leighann Claridge; Anne Franklin; Mrs K Berry;Marilyn Eggleton;The Purely Recruitment Company; Alan Byrne; Price Forbes, Rhodri Bowen; The ACT Foundation; Silvia Marsella; Emma Ashton; Fred Kimblin

In Memory
Paul Franklin; Ian Sanderson

Collection Boxes
Christine Hancox; Jackie Cooper; N C & B Lunt Pharmacy; Vivienne Culley; J Fair; Mrs D M Robinson

Stamps, foreign coins, mobile phones, ink cartridges; jewellery
Ellen Graham; Derwent and Solway Housing Association;

The Society would like to thank the following donors for their regular contributions by either Standing Order or Give As You Earn.
N Cadman; J Wilson; J York; M Rigby; K Robinson; J Wood; K Osborne; A Tresidder; Mr Thompson; E Cox; B Weston; M Peach; R Arnold; J Ellis; I Pearson; C Gibbs; A Cock; A Dickerson; M Kalsi; P Summerton; J Dalligan; M Malcolm; E Mee; A Sabin; K Brown; E Brock; M Fullalove; M Reeves; G Ferrier; R & E Parkinson; A Wells; R Taylor; R Gregory; C M Pierce; A Ephraim; Mrs Wallis; K Bown; E Moody; S & J Home; V Little; Z Gul; S & D Greening; J Casey; D Palmer; J & V Hastings; E Lee; A Byrne; D M Robinson; B M Giles; M Peach; R Arnold; J Ellis; I & V Pearson; D & S Peach; C & M Gibbs; Mr & Mrs Cock; A Dickerson; M Kalsi; A Weston; E White; C Hume; R Dunn; S Littledyke; N Saville; M Tosland; S Bhachu; C Cullen; I Hedgecock; D Forbes; P Shrimpton; G Simpson; W Cavanagh; A Sabin; B Harriss; L Brodie; A Ephraim; C Garthwaite; R & K Henshell

Wear It Blue!

Thursday 15th May 2014

Wear It Blue for genetic, life-limiting diseases

On Thursday 15th May 2014 we're Wearing It Blue to mark this year's MPS Awareness Day.

MPS Awareness Day is an international campaign raising the profile of Mucopolysaccharide (MPS) and related diseases. These are rare, genetic conditions which are devastating to the families they affect.

Taking part is easy, just wear something blue and donate £1 or more to the MPS Society to help fund vital research and support affected families.

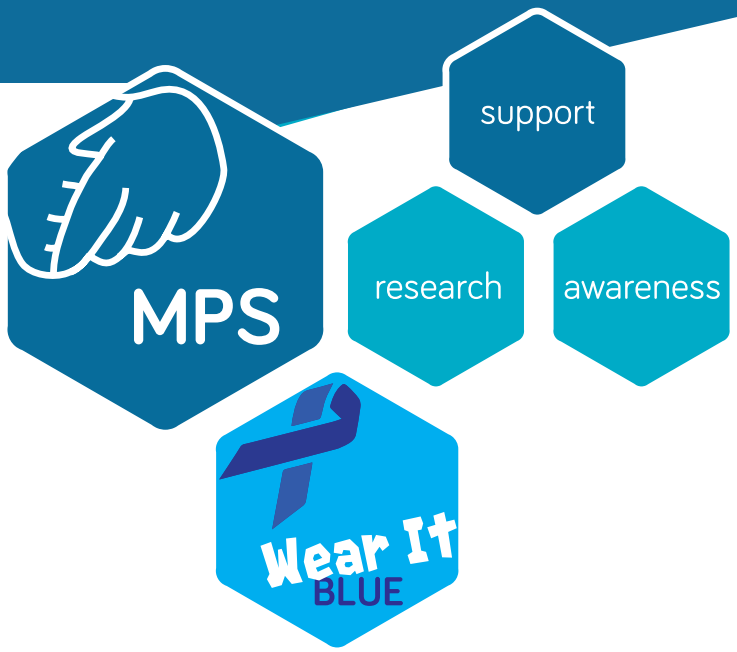
We know we can make a difference but we need your help.

To take part, give your £1 or more to:

_____

_____

_____



The Society for Mucopolysaccharide Diseases is the only UK charity providing professional support to those affected by 24 MPS and related diseases, funding research and raising awareness of these rare genetic diseases.

www.mpssociety.org.uk

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