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Get ready for some Fangtastic fundraising with **Wear It Wicked** for Halloween

Plus, features on the MPS Conference, research, treatment and advocacy updates

MPS

support

research

awareness

Wicked GENES

The MPS Magazine

Autumn 2013

Society for Mucopolysaccharide Diseases

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The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Management Committee.

The MPS Society reserves the right to edit content as necessary. Products advertised in this magazine are not necessarily endorsed by the Society.

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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:

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

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

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

MPS 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 resulting in death in childhood.

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

MPS Christmas Cards available to order online at **www.mpssociety.org.uk** or by post, phone **0845 389 9901** or email **info@mpssociety.org.uk**.

Contact us for our Christmas Collection 2013 order form.

Once you have read this MPS magazine, please pass it on to your family, friends and colleagues.

Help us spread the word about MPS and related diseases and the work we do.

www.mpssociety.org.uk

Welcome!

Putting the finishing touches to the magazine is always a bit of a mission and we always seem to be adding bits right up to the deadline. But we love working on it as reading the inspirational and moving stories always make us feel so proud to be part of such an organisation.

Christine's report on p4 highlights latest MPS achievements both in research and in the outstanding service our Advocacy team deliver.

This edition there is a special report on the 2013 MPS Conference (p15), which was a huge success, thanks in no small part to the incredible childcare volunteers.

P18 has an article on the 10th Anniversary of Aldurazyme, the Enzyme Replacement Therapy for MPS I Hurler, including first-hand accounts of the difference this treatment has made to our members.

A big thank you to our fundraising superstars and some ideas for events on p23 (Wear it Wicked anyone?) followed by your stories.

Don't miss the Kembrey's special report on Marina and Friends shop fundraising milestone (p24).

Our Wicked Walkabouts initiative has reached a new high with a 3 legged pub crawl (Thanks Steve Brooks!) p27.

Best wishes, The MPS Team

Contents

Front Cover
Volunteers and children at the MPS Conference 2013.

This magazine was printed with the kind support of a grant from The Albert Hunt Trust.

Chief Executive's Report

Asked what is the MPS Society's greatest achievement these past months unequivocally I would answer, three UK children starting the MPS I Hurler post HSCT ERT Clinical trial.


Christine Lavery, MPS Chief Executive

To put this simply, the MPS Society has been able, with the support of MPS I families, to secure three places on this clinical trial hosted and sponsored by the University of Minnesota. The aim of the trial is to establish a safety and efficacy profile for treating children with Hurler disease aged 7-14 years with Aldurazyme (Enzyme Replacement Therapy) who previously underwent a bone marrow or cord blood transplant earlier in their life. The 7th August was a momentous day for Willow and Olly when late in the afternoon they each received their first infusion of Aldurazyme at Great Ormond Street Hospital. As I write both children have had three infusions on this two year trial with the Aldurazyme funded by NHS England. We are also most grateful to the charitable Trusts and Genzyme who are providing funding for the logistics of this important clinical trial.

It is timely that 10 years after Aldurazyme was given marketing approval in the USA and Europe as Enzyme Replacement Therapy it is now at the forefront of a clinical trial to see if it can benefit another group of MPS I patients.

Turning to another disease and another clinical trial I am delighted to report that the MPS Society has been able to confirm further funding of £50,000 bringing the total to date from MPS to £210,000 to Dr Brian Bigger for the Genistein Clinical Trial at the University of Manchester. We also want to thank the efforts of our members and the international community for their fundraising and donations. We are also most grateful to the Gem Appeal who are donating £50,000 for this important study. We offer special thanks to the National MPS Society in the USA who granted S250,000. This means to date we have raised £440,000 and we only need your help to find another £210,000 over the coming year. The wonderful news is that the study is now ready to recruit MPS III patients over the coming months.

The MPS Society fully appreciates that research and clinical trials are not the priority for many of our members with the day to day living of those affected and their carers uppermost in their minds. A strengthened MPS Advocacy Team are working very hard to meet your needs and expectations. If you have an advocacy problem, however big or small, please do contact the MPS Advocacy Team who will be pleased to help. Phone 0845 389 9901 or email advocacy@mpssociety.org.uk.

Support activities are high on the Society's agenda too and I was pleased to see over 100 members and children enjoyed their weekend at Legoland. There is a lot on the horizon starting with a day at the Harry Potter Experience and a visit to Lapland UK coming up in the near future.

Christine Lavery MBE
Chief Executive
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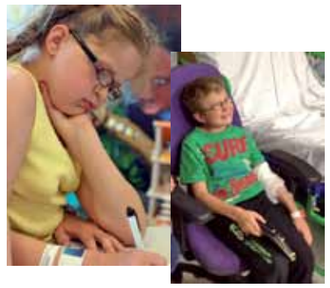

Willow and Olly on the MPS I post HSCT ERT trial

Find us on Facebook
www.facebook.com/mpssociety

twitter
twitter.com/MPSSocietyUK

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 in April 2013.

The meeting was opened by the Chairman, Susan Peach and started with a one minute silence in memory of Professor Ed Wraith who was such a valued advisor to the MPS Society and who has given 25 years to the clinical management and treatment of MPS children and young adults.

Personnel Report

Trustees welcomed Sue Cotterell back to work following her maternity leave. Trustees stated that they had considered the unrestricted income of the MPS Society and agreed they were unable to approve the creation of a further Advocacy post at this time.

The Chairman introduced the need for regular performance management of Trustees and referred to the appraisal form and thanked the majority of Trustees who completed it.

Clinical Management & Treatment

The Chief Executive appraised Trustees of the developments of both the MPS I Hurler post HSCT ERT clinical trial and the Genistein clinical trial due to start in September 2013. She also reported on the activities of the specialist LSD centres and relationships with MPS.

Advocacy Support

The Chairman presented a report by the Senior Advocacy Officer, Sophie Thomas, and gave examples of work being undertaken due to the change in the benefit system and the huge number of enquiries. Trustees acknowledged this influx is currently stretching the advocacy team to the limits.

Treasurer's Report

The Treasurer presented the accounts to the Board of Trustees and sought approval of the year end accounts which were for a period of 14 months to include incorporation to a Charity Limited by Guarantee. The accounts had been sent to Trustees well in advance and after further discussion were agreed. A revised reserves policy was tabled and agreed.

Risk Management

The Finance Officer gave an update on the position of extending the mortgage to purchase the adjoining building to MPS House to provide more adequate desk space. She confirmed that the Society had received restricted grants from two funders to allow the alterations to be made. It was also confirmed that the sitting tenants upstairs would stay and the rent cover the additional mortgage.

The Risk Management Register was tabled for discussion. Trustees agreed that a full cost recovery charge of 5% for research grants up to £100,000 and £10% for research grants over £100,000 be made as stated in the MPS accounts for year ending December 2012.

Fundraising

A discussion was had on the various highlights for the 'Wear it Blue' Campaign and 'Wicked Walkabouts' to promote MPS Awareness Day in May. Trustees were advised that there is a lot of promotional activity and the Fundraising and Communications team are busy promoting legacies, direct debits and Give As Your Earn.

Retirement of Bob Devine

At the July Trustee Meeting Bob Devine retired as Trustee of the MPS Society after many years service. Bob and his wife Lyndsey joined the MPS Society after their youngest daughter, Katie, was diagnosed with MPS I Hurler disease in 1996. Sadly Katie died in December 2000 aged 5 years but that didn't stop Bob continuing his outstanding support to MPS.

As a token of their appreciation the Trustees and Chief Executive presented Bob with an engraved beer tankard and a selection of old ales. Thank you Bob for your unstinting service.

www.mpssociety.org.uk

The MPS website is constantly updated with latest news on research and treatments and MPS member events. There are lots of useful downloads and resources from our Advocacy Team from knowing your rights to understanding the benefits systems and guides on how to manage difficult behaviour. The site is full of advice on how to access help when and if you need it.

The MPS Society website is also packed with imaginative ideas for fundraising for the Society, whether you are running a marathon, holding a school cake sale, or planning a Wear it Wicked event for Halloween, there is something for everyone. We also have posters, sponsorship forms and guides to download.

If you are planning an event don't forget to visit our on-line shop for T-shirts, badges and bands and with Christmas not that long away, you can pick out some MPS Society Christmas cards to send to friends and family. We are always looking for ways to improve our site, so if you have any suggestions or ideas please don't hesitate to contact us at **info@mpssociety.org.uk**

New members

Jordan has recently been in contact with the Society. Jordan was diagnosed with Fabry disease in February 2009 after suffering a stroke. He has two young children aged 1 and 4. The family live in North Yorkshire.

John has recently been in contact with the Society. He has Fabry disease. The family live in the Newcastle area.

Ms Mohammed has recently been in contact with the Society. Her son Ayaan has a diagnosis of Sanfilippo. Ayaan is 3 years old. The family live in the North West.

Vicki has recently been in contact with the Society. Her daughter Skye has a diagnosis of ML II. Skye is 1 year old. The family live in Essex.

Cath and Chris have recently been in contact with the MPS Society. Their son Clark has a diagnosis of Hunter disease. He is 6 years old. The family live in Surrey.

Mr and Mrs Ramani have recently been in contact with the Society. Their son Kayen has a diagnosis of ML II. Kayen is 2 years old. The family live in the South East.

Sharon has recently been in contact with the Society. She, along with her two sons, Jake and Taylor, have a diagnosis of Fabry disease. The family live in the North East.

Jane has recently been in contact with the Society. She has a diagnosis of Fabry disease. The family live in the Bristol area.

Births

Congratulations to the **Hulse Family** on the birth of baby Scout Eiko Hulse on 16th April 2013. Scout is little sister to Miya (MPS IH).

Miya (MPS IH) and little sister Scout

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.

What's on

CONFERENCES and REGIONAL EVENTS

Childhood Wood Planting - 13 October

Newcastle Family Day - date tbc

Harry Potter Family Day - 20 October

Lapland UK Family Day - 4 December

MPS REGIONAL CLINICS 2013

Manchester BMT Clinic (under 6 years) - 11 October

Manchester BMT Clinic (over 6 years) - 18 October

Birmingham MPS and Fabry Clinic - 8 November

MPS III Clinic GOSH - 12 November

Bristol Clinic - 12 November

Northern Ireland Clinic - 22 November

Congratulations

Hi, my name is Billy and I have Fabry disease. I am 18 and was diagnosed when I was 15 along with my brother Lewis and my mum.

The MPS Society have been very helpful to me and my family giving us a great deal of support and advice. When I reached 17 I realised that learning to drive would be very beneficial to me and not make me so dependent on my family.

Advocacy Officer Rebecca Brandon kindly put me in contact with charities that could help with driving lessons and after writing to a few charities The Gibbons Trust offered me a grant to pay for some driving lessons.

I passed my driving test this year and have not looked back since. My life and independence have improved so much and I really appreciate the help I have had both from MPS and the Gibbons Trust. Thankyou!

Billy (Fabry) having passed his driving test

Congratulations to Naomi Carter
who sat her GCSE stats exam early and received an A!

Congratulations to Robert Wynn, Consultant Paediatric Haematologist and Director of the Paediatric BMT Programme at Manchester Children's Hospital who was recently made a Professor.

Professor Wynn was born in Sudan but grew up in Liverpool. He trained in medicine in Cambridge and London and qualified in 1989. He undertook post graduate medicine training in Newcastle and Edinburgh, Haematology training in Cardiff and Manchester and Paediatric Transplant training in Toronto.

He was appointed Consultant in Manchester in 1998 and has been Programme Director since 2004. He is a full time NHS Consultant and the author of over 80 published papers, mostly in Paediatric Haematology and Transplant. Professor Wynn is best known in the MPS community for his pioneering work in cord blood transplantation in MPS IH.

Ben Cooper (MPS II) got two As and an A star in his A-levels and will be going to Hull University to study British Politics and Legislative Studies for four years, starting in September.

Ben's father, Lee, writes: 'The University have been very good in identifying support for him during his course to help with his hearing problems and slightly reduced mobility. As part of the degree course Ben will spend a year as an intern at Parliament and will live in London during 2015/16 which will present some challenges, but no insurmountable ones.'

Dr Fiona Stewart is awarded an MBE

Congratulations to Dr Fional Stewart MBE

Dr Fiona Stewart has been awarded an MBE for services to genetics in Northern Ireland. Based at Belfast City Hospital, Dr Stewart has been a consultant in genetic medicine since 1995 and sees people with inherited genetic disorders, or who are at risk of inherited genetic disorders.

She has a special interest in inherited metabolic disorders and is very involved in trying to improve services for people with rare diseases in both Northern Ireland and the rest of the UK.

Dr Stewart said being awarded the MBE was "fantastic", although the news came as a "huge shock." She added: "It has been the biggest secret I have had to keep for five weeks. I wasn't even able to tell my children, although my husband knew because he was there when I opened the letter."

Dr Stewart said she initially thought the letter was from the Tax Office.

"I thought it was a tax return but when I opened it thankfully I was reassured it wasn't. "I was eventually able to tell my children and they are delighted for me. To receive the MBE is absolutely fantastic." she said.

Dr Stewart is a former pupil of Cambridge House Grammar School and qualified from the Royal Free School of Medicine in London. She is married to Sam and has two children, Emily and David.

Article taken from the Ballymena Guardian, 20 June 2013. www.ballymenaguardian.co.uk

In Memory

Jack David Heath

12th August 1999 – 19th June 2013

Jack passed away peacefully just after 1am on 19th June surrounded by his family.

Jack suffered from MPS II Hunter disease and fought bravely since his diagnosis in May 2002. Over the years Jack and his family overcame many obstacles with the fantastic support of the MPS Society, Social Services, Holy Trinity Primary School and numerous health professionals from Royal Manchester Children's Hospital, Rochdale Infirmary and Burnley Reedley Hall Team and in his final weeks, the Palliative Care Team from Rochdale.

Despite his decline in health throughout his life, Jack was well travelled, having visited the USA twice to Disney World, France to Disney Paris, Woolacombe in North Devon most years and trips out with his mum, dad and younger sister to various special events – especially air shows which he loved, and was even invited to open the Disney Store in Manchester. He also attended several of the MPS Society events over the years including conferences, Christmas parties, days out in London and weekend events at Camelot where Jack would enjoy time with other MPS sufferers and their families that he had met along the way.

The last few weeks, though very difficult for Jack and his family and despite having several tubes and a syringe driver attached, Jack managed to get outside to sunbathe and plane spot in the beautiful weather – what a way to go...

Although Jack's passing was not unexpected he has left a massive hole in our hearts and lives but his parting gift to us was a look of peace and tranquillity and unbelievably a smile on his face as he fell to sleep with the angels.

Jack was well loved by many in the Rossendale Valley after appearing in the local newspapers and was somewhat of a local celebrity – once met, never forgotten and this was apparent by the hundreds of people that turned out for his funeral.

Jack's final journey was in a white horse drawn hearse – which he would have loved, from his home to the church where he had been dedicated and later anointed. The meal after the funeral was also Jack's favourite – potato pie with peas and red cabbage!

Jack would have been 14 on 12th August and to remember this Jack was honoured by Ashworth and Baker Funeral Directors who have named their memorial garden in his memory.

Jason, Elizabeth and Katie would like to thank everybody who has played a part in Jack's short life.
Jason and Elizabeth Heath

Jason, Elizabeth and Katie Heath at the Memorial Garden dedicated to Jack

Bereavements

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

Richard Horner who suffered from Fabry and passed away on 24 November 2012.

Jack Heath who suffered from Hunter disease and passed away on 19 June 2013 aged 13 years.

Daniel Muers who suffered from Hunter disease and passed away on 6 September 2013 aged 17 years.

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

MPS Regional Specialist Clinics

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

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

The MPS Society supports the regional clinic programme, which can include arranging and co-ordinating dates of the annual programme and funding the tertiary centre doctors' travel and subsistence costs to the regional centres. Specific arrangements for each clinic and for organising appointments differ for each of the centres. At least one member of the Society's advocacy support team meets individuals and families face to face to offer individual advocacy support. **Sophie Thomas**

BMT Clinic under 6 years

12.07.13 - I was greeted at Royal Manchester Children's Hospital by Jean Mercer when I arrived at the clinic. I heard the good news that Rob Wynn is now a Professor and I offered my congratulations and let the MPS staff know on my return. Morgan was wearing a pretty dress and was having McDonalds after her appointment. Jessica is looking forward to starting school in September. April was happy with a lovely smile and enjoyed eating her yogurt. Jake was planning to go to the seaside at the weekend as the weather was going to be hot.

MPS IV Clinic

09.07.2013 - It was a lovely sunny day when I arrived at Great Ormond Street hospital for the MPS IV clinic. Kamal very proudly showed me photos of himself at all ages on his Ipad and showed Michelle the physiotherapist all his moves, including how he can score at basketball. Suhila and Sara came to clinic with their Dad and were excited about the summer holidays arriving soon and spending time with their family.

BMT Clinic over 6 years

05.07.13 - Ethan was bright and early to clinic at Royal Manchester Children's Hospital and brought Mum and Dad along. Rubina also brought her family along and was happy with a lovely smile. Lyla was looking forward to being a bridesmaid soon. Isaac had very proudly won a medal at a party the night before. Thomas had some cool new trainers and was excited to be starting secondary school in September. Steven also brought all his family and his brothers and sisters enjoyed playing with the toys. Demi-Leigh looked lovely in a very pretty skirt. **Debbie Cavell**


Ethan


Rubina


Isaac


Lyla

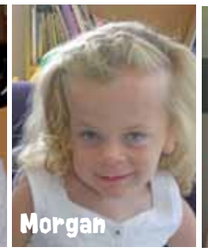

Thomas


Steven


Demi-Leigh


Suhila & Sara


Kamal


Morgan


Jessica


April


Jake

Bristol Clinic

08.07.13

Well it was a very sunny morning for mine and Steve's early morning drive to Bristol, not quite as early as Dr Jones and Jane Roberts who had a 4.30am wake up!

We all arrived a little bleary eyed but ready to greet the families. We had several new families to meet which is always a pleasure and I couldn't resist in finishing the colouring left behind by one of the children...

We had lots of children visiting but for one thing and another they did not want their photos taken, I blame it on the hot weather and that is a first I think for this year!

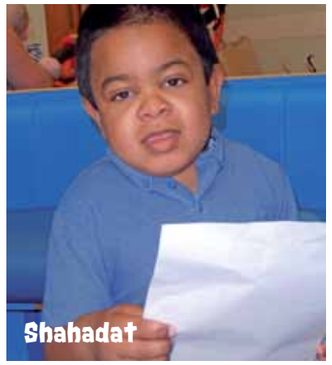
Lauren was very bright and cheerful, chatting away and something told me that she liked pink and sparkles, she had sparkles on her shoes and t-shirt.

Shahadat and Shafaat were quite happy to just stay and play, much more fun than having to listen to the grown-ups talking!

Soon it was time to leave and continue our journey to catch up with some families in Wiltshire and Somerset.

Our thanks to the team for making us welcome and see you all another time. **Rebecca Brandon**


Lauren


Shahadat


Shafat

MPS III Clinic

13.08.13

The MPS clinic at GOSH was held on the 13th August, the waiting room was full of hustle and bustle, it was good to see families taking the opportunity to catch up and compare notes.

Bobby enjoyed a tour of the outpatient department with Lily and Tillie not too far behind, Ollie surveyed the scene and grudgingly allowed me to get a photo. Despite reluctance to sit still and the many blurred attempts I managed to take photos of everyone. Jamie was on particularly good form singing "if you're happy and you know it" and shouting "WE ARE!" at the top of his voice.

Thanks as usual to Dr Vellodi, Niamh and the team at GOSH for hosting the clinic and we will see you next time. **Steve Cotterell**


Tillie Mae & mum


Bobby


Jamie


Ollie

Advocacy - Focus on... The Mental Capacity Act 2005

The Mental Capacity Act 2005

- Is effective in England and Wales, in Scotland there is The Adults with Incapacity Act 2000.
- There is no equivalent law in Northern Ireland.
- Applies to everyone over 18 years old.
- Is intended to protect the rights of individuals who lack capacity to make decisions for themselves.

What is capacity?

Being able to make your own decisions through the receiving, understanding and application of appropriate information to reach an informed decision or choice."

When a person does not have capacity

Carers have legal or statutory protection if they can demonstrate that they:

- Have taken reasonable steps to assess capacity to make a decision.
- Reasonably believes that the person lacks capacity to make the decision.
- Reasonably believe that the decision is in their best interests.

When a decision is needed

Once an individual has been assessed as lacking capacity there may be situations where informal decision making is not appropriate and an application to the Court of Protection is needed for example:

- Particularly difficult decisions
- Disagreements that cannot be resolved
- Situations where on-going personal welfare decisions must be made about someone who lacks capacity

Do you need to apply to the court?

- There are certain informal tasks that are carried out daily that will not require an application to the court. The court deals with more complex cases or situations that cannot be resolved or if "best interests" are in question.
- If the individual is in receipt of state benefits only you may not need to apply for property and affairs if you are already an appointee with the DWP.
- You may need to apply to manage assets, pay bills deal with income if there is no valid lasting/enduring power of attorney.
- If the person lacks mental capacity to make decisions about personal welfare an application to the Court is not usually necessary.
  Most care and treatment decisions can be made by those providing care, so long as they are acting in the individual's best interests. A best interests-meeting may be called to include family and relevant professionals.

Things to know

- Costs: There is a fee payable when making an application to the Court.
- If applying for personal and financial affairs you will be asked to take a security bond with an insurer. This covers mismanagement of affairs.
- The office of the Public Guardian offers support to appointed deputies.

Contacts

Office of the Public Guardian
customerservices@publicguardian.gsi.gov.uk
Telephone: 0300 456 0300

Court of Protection
courtofprotectionenquiries@hmcts.gsi.gov.uk
Telephone: 0300 456 4600

Further information

www.gov.uk

Challenging behaviour factsheets

The Challenging Behaviour Foundation has developed a series of three information sheets which look at the causes of challenging behaviour and explain how to make effective plans to reduce challenging behaviour.

The information is designed for families or professionals who support a child or adult with severe learning disabilities whose behaviour challenges. The sheets can be downloaded free of charge from **www.challengingbehaviour.org.uk** or phone 01634 838739 to request a hard copy (cost £1).

All Ireland Advocacy and Support Update

It's been a busy summer over in Ireland! Heat waves, torrential rain, the occasional thunder and lightning storm, a fantastic 'Wear it Blue' family day and lots of advocacy and support work.

The summer months are traditionally a quieter time for clinic appointments so we've had a few months without any of our 'big' MPS or Fabry clinics. The next MPS clinic in Antrim Area Hospital is booked for 22 November and we are anticipating a jam-packed day! I look forward to catching up with everyone on the day and as usual I would encourage those who are attending to get in touch before the clinic if you have any specific queries or concerns – this really helps to make the most out of your appointment.

The next Fabry clinic date is yet to be decided but I'll be in touch as soon as we have a date.

If you feel that you, or a member of your family, need to be included in one of our upcoming clinics please get in touch with Alison as soon as possible on either 028 950 47779 or 077 826 58336.

During this quieter time of the year I always enjoy getting stuck into form filling, letters, care plans, reports and lots of research to keep myself up to date with the ever changing world of advocacy support. There's nothing quite like a productive day in the office! Lots of families in Ireland have requested advice and support in relation to housing, adaptations and social support over the last few months and we are delighted to have made a difference to families in these areas.

N Ireland Rare Disease Partnership

As you are aware I represent the MPS Society in the Northern Ireland Rare Disease Partnership (NIRDP).

Over the summer there have been lots of NIRDP meetings and lots of progress is being made in raising awareness of, and developing services for, rare disease in Northern Ireland. I was delighted to represent the NIRDP and the MPS Society at a meeting with the Northern Ireland Health Minister (Edwin Poots) to discuss the future of rare disease services in Northern Ireland in August this year. With the publication of the UK Rare Disease Plan moving ever closer it's so important that those working in the rare disease field (particularly in the devolved Nations) work closely with politicians and policy makers to reach local solutions to some of the difficulties that arise when considering the provision of services for rare and ultra rare diseases. In the coming months we have lots of dates in the diary for meetings with MLAs, Health and Social Care Professionals, Commissioners and Policy Makers.

The MPS Society will be represented at a rare disease event in the Stormont Long Gallery in January 2014 and also at the Northern Ireland Rare Disease Day in Riddel Hall (Stranmillis) in February 2014. The first of these events will be a showcase day for national and local charities who work with those affected by rare disease in Northern Ireland. At the second event we hope that the Health Minister will make comments about how Northern Ireland will apply the UK Rare Disease plan locally. These are two very important events and I look forward to updating you with some photographs in a future edition of the MPS magazine.
Alison Wilson

Northern Ireland 'Wear It Blue' Family Day

On 8th June 2013 we held a Northern Ireland 'Wear it Blue' family day at the Stormont Sports Pavilion in Belfast to bring families together for a fun social event. Dr Fiona Stewart attended along with many MPS member families, for some of whom this was their first MPS event.

The room was set up with blue decorations in keeping with the Wear It Blue theme. There were tables where families could sit and chat over the buffet lunch. With the kind support of a grant from the Starlight Foundation, we were delighted to have party bags for all the children who attended. We were also able to offer a children's play corner with craft activities including clay models for painting, colouring in, finger/sponge painting and painting by numbers.

On the day we were delighted to have been offered the services of Ten21 photography without charge. This is a local photography company who are seeking to expand their current portfolio to include charity events and 'wishes' for sick children. The company kindly agreed to provide photographs of the day and are also working on an MPS Society awareness video clip.

Wearing It Blue! in Northern Ireland

Days like these are invaluable in enabling families to come together and share stories and experiences. On this day we were able to set up two befriending links which have proved very successful. These links have continued and families have been in touch regularly since the event. One befriending link in particular has been critical in supporting a teenager to come to terms with his condition and understand that he will be able to achieve independence.

Thank you to everyone who attended and supported this event making it so special. **Alison Wilson**

Scottish Get Together

The MPS Society held a Get Together for Scottish families on Sunday 3rd March 2013 at the Hilton Edinburgh Airport, made possible by the kind support of a grant from the Souter Charitable Trust.

There was a special two course buffet lunch with an entertainer, balloons and presents for the children. Everyone greatly enjoyed the meal and once lunch was over the entertainment began. The magician, who had been so popular with the children at last year's party, made some wonderfully imaginative and colourful balloon models for all the children and then delighted the party with his magic show, participation coming from the children and adults alike!

The date also coincided with the birthday of Jennifer (MPS IHS) so this year's party was a very special get-together. Jennifer's Mum had baked and decorated a birthday cake and her niece and nephew helped to blow out the candles and cut the cake at the end of the afternoon, so that everyone could take a slice home with them.

Everyone who attended agreed that they had enjoyed a fabulous afternoon together, with a chance to meet up with friends from past events and to get to know new families too.

Thank you to the Souter Charitable Trust for their kind support and to everyone who attended for making the day so fun!






Here are a selection of photos from the Scottish Family Day with entertainment, balloon modelling and a beautiful birthday cake for Jennifer!

2013 MPS National Conference

This year the MPS Society organised our 2013 MPS National Conference 28-30 June 2013 at the Hilton Hotel Coventry.

This year's programme was very diverse and included presentations from medical and health professionals and family speakers. Topics covered included Living with Fabry and MPS and related diseases, palliative care and the latest updates on research and treatment.

As always, a key feature of the MPS Weekend conference was the fantastic childcare programme assisted by our enthusiastic and experienced childcare volunteers.

We were lucky in having wonderful weather for the whole of the weekend and the outing to Drayton Manor Park on Saturday was a great success, with the volunteers helping all the children, both MPS affected children and their unaffected siblings, to get the very best from their visit. All the children had a wonderful time at the Park and returned to the hotel in the late afternoon with broad smiles on their faces and many exciting memories to take back home. Some of the older MPS affected children and their siblings and young adults, accompanied by volunteers, were taken to a local bowling alley on Saturday evening; this was another welcome opportunity for relaxed activity and for socialising with others living in similar circumstances.

The outing on the Sunday morning was to the National Sea Life Centre in Birmingham and was also a great success. The children could take 'a marine voyage beneath the waves' and find out more about sharks, turtles and other marine creatures. There was also a children's soft play area. Those children and young people who were unable to travel on either Saturday and Sunday were still able to enjoy a programme of supervised activities in specially allocated areas within the hotel, to help stimulate them physically and mentally in a secure environment.

The volunteer carers also looked after the children on Saturday evening, either individually in their rooms or with various organised group activities again in allocated areas within the hotel. As a result, Saturday evening's Gala Dinner for parents, MPS staff, professionals and for those MPS affected adults able to participate proved to be a memorable occasion, with opportunities both during and after the meal to cement friendships, to share opinions, insights and experience from the day's professional presentations and discussions and - for those with sufficient energy - to dance to music late into the evening.

Thank you to everyone who took part.

Thank you so much to everyone who supported our bric-a-brac and clothes stall at the MPS conference by donating and purchasing items. Also a big thank you to everyone who had a go at guessing the weight of our giant Winne the Pooh and Tigger teddy bears which were won by Paul Moody.

We look forward to seeing you at future events.




MPS Conference 2013 - A Volunteer's Experience

I truly didn't know what to expect when I arrived at the Coventry Hilton, for the MPS Conference.

I had volunteered to look after a child, and having only baby-sat friends' children, nieces and nephews, I was having some feelings of trepidation.

I was in time for the pre-meeting brief and safety presentation by the team leaders and then it was off to meet our charges.

I was introduced to my 'companion' for the weekend, a 4 year old chap who has MPS I. He had a bone marrow transplant some years ago and when I met the family I knew I was in for a busy time, so much energy! My earlier feelings of trepidation quickly vanished, I had nothing to worry about, he is one of the most fun-loving, delightful boys I had ever met.

After the introductions with Mum and Dad and a chat about any special requirements, the evening was soon over and the next day loomed... a day to Drayton Park Manor.

After an early start getting everyone onto the coaches, we were soon there, thankfully my companion and I both enjoyed train rides and boat trips... such a relief as I don't really like roller coasters ! We saw every train in the park, and more than once! I think I can speak for the whole group, when I say it was a highly successful day, with something for everyone.

The evening was a relaxed time recovering from the exploits of the day. We played with cars and watched 'You've Been Framed' whilst the older children and parents partook of their organised events.

The following day was another early start, this time to 'Sea Life World' in Birmingham. The fish and undersea life weren't such a hit as the trains and boats the day before, but 3D Sponge Bob certainly was.

The entire weekend was extremely well organised, the organisation team must be highly commended. The trips were well suited for the entire group and you could participate in as little or as much as you liked.

I sincerely hope we gave the parents some much-deserved respite and an opportunity to network with other families.

A big thank you to the MPS Society, and I would definitely consider volunteering again, if invited.

John Pringle
Clinical Field Specialist, Genzyme

The training of volunteers was kindly supported by funding from Chiltern District Council.

Chiltern District Council

"Overall it was a really good weekend and I really enjoyed it. I thought the trips out were really suited to everyone. Can't wait for the next one!"

"Extremely interesting presentations. Felt that I had gained a lot of new knowledge that I could apply to my service. As usual, the parent presentations were inspiring."

"This was the first visit for our family after recent diagnosis and we found the whole experience welcoming, informative and very well presented to ease any concerns."

"Very well organised conference. Thank you to all the staff involved!"

Clinical Trial Update

For full information about the trials listed here, please visit **www.mpssociety.org.uk** and click on the relevant diseases tab.

Clinical Trials currently recruiting

These clinical trials are active and recruiting patients. If you have any questions about clinical trials, please contact your clinician, the clinical principal investigator or the MPS Society.

MPS I

Intrathecal Enzyme Replacement for Hurler Disease
Laronidase (Aldurazyme TM) Enzyme Replacement Therapy With Haematopoietic Stem Cell Transplant for Hurler Disease

Pilot Study of Administration of Intravenous Laronidase Following Allogeneic Transplantation for Hurler Disease

A Study of Intrathecal Enzyme Therapy for Cognitive Decline in MPS I

A Study of the Effect of Aldurazyme® (Laronidase) Treatment on Lactation in Female Patients With Mucopolysaccharidosis I (MPS I) and Their Breastfed Infants

MPS II

Collection and Study of Cerebrospinal Fluid in Patients With Hunter Disease

Observational Study to Evaluate Neurodevelopmental Status in Paediatric Patients With Hunter Disease (MPSII) - HGT-HIT-090

Biomarker for Hunter Disease (BioHunt)

MPS IIIB

Natural History Study of Patients With Mucopolysaccharidosis Type IIIB (MPS IIIB, Sanfilippo Disease Type IIIB)

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 VI

Mucopolysaccharidosis (MPS) VI Clinical Surveillance Program (CSP)

Fabry

Open-Label Phase 3 Long-Term Safety Study of Migalastat (MGM116041)

Canadian Fabry Disease Initiative (CFDI) Enzyme Replacement Therapy (ERT) Study

Natural History and Treatment Outcomes(Changes) in Fabry Renal Disease Study (LDN6702)

Stroke in Young Fabry Patients (sifap2): Characterization of the Stroke Rehabilitation

The Efficacy and Safety of Switch Between Agalsidase Beta to Agalsidase Alfa for Enzyme Replacement in Patients With Anderson-Fabry Disease (SWITCH)

Establishment of Biomarkers for Fabry Disease

Immune Response in Subjects With Fabry Disease Who Are Switching From Agalsidase Alfa to Agalsidase Beta

Sophisticated Assessment of Disease Burden in Patients With Fabry Disease (SOPHIA)

Fabry and Cardiomyopathy (FaCard) Epidemiological Study for the Analysis of Biomarkers and the Clinical Course of Patients With Fabry Disease and the N215S-mutation

A Study of Renal Function in Treatment-naïve, Young Male Patients With Fabry Disease

Clinical Trials that are active but not currently recruiting

MPS II

Extension of HGT-HIT-045 Evaluating Long-Term Safety and Clinical Outcomes of Idursulfase (IT)in Conjunction With Elaprase in Paediatric Patients With Hunter Disease and Cognitive Impairment

MPS III

Extension of Study HGT-SAN-055 Evaluating Administration of rhHNS in Patients With Sanfilippo DiseaseType A (MPS IIIA)

MPS IVA

Long-Term Efficacy and Safety Extension Study of BMN 110 in Patients With Mucopolysaccharidosis IVA (Morquio A Syndrome)

Study of BMN 110 in Pediatric Patients Under 5 Years of Age With Mucopolysaccharidosis IVA (Morquio A Syndrome)

Safety and Exercise Study of BMN 110 for Morquio A Syndrome

MPS VII

Intravenous Enzyme Replacement Therapy for MPS VII, Sly Disease

Fabry disease

Study of the Effects of Oral AT1001 (Migalastat Hydrochloride) in Patients With Fabry Disease

Study to Compare the Efficacy and Safety of Oral AT1001 and Enzyme Replacement Therapy in Patients With Fabry Disease

Open Label Long-term Safety Study of AT1001 in Patients With Fabry Disease Who Have Completed a Previous AT1001 Study

10th Anniversary of Aldurazyme

On 11th June 2003, 10 years ago, Genzyme and BioMarin received marketing approval for the Enzyme replacement therapy, Aldurazyme. This is the first specific treatment approved in the European Union for children and adults with a diagnosis of MPS I.

In 2003 for the 50 or so MPS I patients who had not been treated with bone marrow or cord blood transplant hope was now on the horizon. Already 10 MPS I children in the UK had been receiving weekly infusions of Aldurazyme as part of the MPS I ERT phase III/IV clinical trial and reported anecdotally that their physical condition had improved. Nothing is that easy and in 2003 funded Enzyme Replacement Therapy was at the behest of individual Primary Care Trusts in England. A taste of the post code lottery.

The MPS Society's finest hour was when it successfully persuaded the Minister of Health at the time, to award National Specialist Commissioning to the Lysosomal Storage Diseases. This decision in 2004 (implemented in April 2005) brought to an end total misery for MPS I families who supported by the MPS Society were subject to humiliating funding refusals and appeals.

From April 2005 all MPS I Hurler Scheie and Scheie patients have been able to access Aldurazyme in England and Northern Ireland. In Scotland and Wales accessing Aldurazyme continues to be a battle and support of the MPS advocacy team is always on hand. **Christine Lavery**

Aldurazyme really has changed my life

When I was asked to do this interview I started to reflect on where I'd be now if I wasn't on Aldurazyme and can only conclude that my life would be completely different and certainly for the worse.

I was diagnosed with MPS I Scheie aged 10 years by which time I had already had four operations for carpal tunnel, trigger release and an umbilical hernia. In my teens I had osteotomies on both knees and by 16 my condition was really deteriorating to the point where I was physically an old lady before my time. I was in so much pain and so tired.

I started Aldurazyme after a battle with the local PCT at 17 years old. Before Aldurazyme I felt like a 70 year old and needed my parents to help me with getting up stairs, dressing and washing my hair. Aldurazyme has changed everything.

It is difficult to think about those times, but reflecting back I could never have gone to University to pursue a career as a radiographer and become the active person I am. When Alex proposed to me I told him that it would only be him and me – no children! I never envisaged that Aldurazyme would transform my quality of life and that I would have not just one child but two. Abigail was born on 15th April 2011 and we are currently expecting another daughter in January 2014. Alex married me on 16 August 2008!

Aldurazyme has given me my independence. I still get tired easily but in every other way I live life the same as a woman of my age without MPS I. Alex canulates me for my weekly infusion and four hours is a small price to pay for my much improved health. It is what you make of it. Alex and I get Abigail to bed and make infusion night special having dinner and just spending time together.

Aldurazyme gives me hope that I can continue to carry on the way I am being the healthiest I have ever been. Looking to the future I just hope that the funding for Aldurazyme continues so that Alex and I can enjoy our family and I can carry on working.
Joanna Wilson-Smale

A mother's experience

Vicki Brockie is Samantha's mother. When Sam was 9 years old she was told that Sam has MPS I. She recalls a picture of doom and gloom with the doctor telling her "Sam's got this condition and we will watch and monitor. There is no treatment or cure." Unimaginably hope came very soon after Sam's diagnosis with news of

an Enzyme Replacement Therapy Clinical Trial starting at Manchester Children's Hospital. Sam was recruited to the trial and later learnt she'd been on placebo for six months before crossing over on Aldurazyme. Sam has continued on Aldurazyme for 12 years and her latest tests and scans including her heart scan show no deterioration since she was diagnosed. Sam did well at school getting 5 good GCSE's and going to college to achieve City and Guilds in Hospitality.

10 years after Aldurazyme received marketing approval, on Saturday 7th September 2013 Samantha Brockie walked down the aisle to marry her finance Ashley. Sam's mum is sure that without her daughter receiving weekly infusions of Aldurazyme it would almost certainly be a very different story. Vicki Brockie says, "As a mother Aldurazyme is a godsend. It is hard to explain an MPS illness to anyone and is confusing. Aldurazyme gives positivity and hope, without it Sam would not be where she is now." **Vicki Brockie**

UPDATE: MPS I - Hurler post HSCT ERT Clinical Trial

Fantastic news! The three UK children on this trial have had their baseline tests at the University of Minnesota in the USA and two of the children received their first weekly infusion of Aldurazyme on 7th August 2013.

A charitable foundation has remarkably ring-fenced funds for this important clinical trial but if you can help with a donation or fundraising this would be appreciated.

Is Bone marrow (BMT) or cord blood transplant (CBT) a cure?
No. These treatments are proven to preserve the brain in young children with Hurler disease but not to be effective in preserving the musculo-skeletal disease. Therefore there is an urgent unmet clinical need.

Why aren't Hurler children treated long term with Enzyme replacement Therapy instead of BMT or CBT?
Hurler disease causes rapid neurological degeneration and Enzyme replacement therapy (ERT), Aldurazyme, does not cross the blood brain barrier.

What is the reasoning for this clinical trial?
Aldurazyme is already a licenced treatment for the attenuated MPS I diseases, Hurler Scheie and Scheie disease. With over six years of experience treating children with these diseases there is evidence of Aldurazyme stopping or improving musculo-degeneration. This is one of the hypotheses of this Phase I/II clinical trial.

Where will the Clinical Trial be carried out?
The children will receive their weekly Aldurazyme at Great Ormond Street Hospital and Manchester Children's Hospital. After 6-8 weeks the children will then receive their enzyme replacement therapy at home with home care nurses. The three UK children will travel five times to the United States during the two year trial for assessments.

How will funding this clinical trial help other Hurler children?
At the end of the two year clinical trial the results will be analysed and the results published. If the results demonstrate safety and significant benefit to the children a multinational Phase III/IV clinical trial will take place. The Phase I/II children will continue on ERT and if this Phase III/IV clinical trial also demonstrates significant benefit Aldurazyme can be prescribed and paid for by Departments of Health across Europe. **Christine Lavery**

Amicus Therapeutics Announces Chaperone-Advanced Replacement Therapy in Development for MPS I

Grant Awarded to Support Preclinical Studies of Next-Generation Therapy

On 25th June 2013 Amicus Therapeutics disclosed a preclinical Chaperone-Advanced Replacement Therapy (CHART™) program for MPS I. Amicus is developing a proprietary human recombinant IDUA (rhIDUA) enzyme co-formulated with a novel pharmacological chaperone as a next-generation therapy for MPS I.

The pharmacological chaperone is designed to improve tissue uptake and reduce the immunogenicity of rhIDUA by stabilizing the enzyme in its properly folded and active form. In addition, in support of its development of a proprietary rhIDUA enzyme, Amicus has received a grant of up to approximately $250,000 from a private U.S.-based donor that provides medical research grants to find better treatments and cures for rare genetic disorders, including lysosomal storage diseases.

David J. Lockhart, Ph.D., Chief Scientific Officer of Amicus Therapeutics, said, "This grant expands our preclinical work and supports our development of a next-generation therapy for MPS I. Our goal is to engineer a proprietary, potentially bio-better rhIDUA enzyme that may be further improved by the addition of a pharmacological chaperone stabilizer. First-generation ERT for MPS I has helped to improve certain manifestations of the disease, however, rhIDUA is highly immunogenic. Directly co-formulating rhIDUA with a chaperone has the potential to increase enzyme stability in the infusion bag and in circulation, enhance uptake of active enzyme in disease-relevant tissues, and potentially reduce immunogenicity."

In people with MPS I, missing or deficient IDUA leads to the accumulation of complex carbohydrates that can affect physical abilities, organ and system functioning, as well as mental and skeletal development. First-generation ERT (laronidase) with rhIDUA has been shown to improve walking capacity and pulmonary function in people with MPS I. However, most patients who receive laronidase develop anti-rhIDUA antibodies, and the laronidase label carries a black-box warning that cites life-threatening anaphylactic reactions in some patients during infusion.1 Amicus believes it may be able to improve upon the properties of the rhIDUA enzyme itself, while also incorporating a pharmacological chaperone stabilizer.

John F. Crowley, Chairman and Chief Executive Officer of Amicus Therapeutics, stated, "Our MPS I program is an additional step forward for our CHART platform technology to combine proprietary enzymes with novel pharmacological chaperones across the lysosomal storage diseases. Importantly, the grant funding supports our internal biologics capabilities at Amicus, which we are leveraging alongside our expertise with chaperones to develop next-generation therapies for people living with lysosomal storage diseases."

UPDATE: Phase III Double Blind, Placebo-controlled Clinical Trial of High Dose Oral Genistein Aglycone for children with MPS III Types A, B and C.

September 2013

The UK MPS Society awards first grant of £210,000 to the University of Manchester to fund a 'Phase III Double-Blinded, Placebo-Controlled Clinical Trial of High Dose Oral Genistein Aglycone in MPS III, Sanfilippo Disease'

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.

To date, the MPS Society with the amazing support of a small number of affected families in the UK, the National MPS Society in the USA, and many other MPS Societies around the world we are guaranteed to raise £450,000

With only £200,000 to go, the UK MPS Society is therefore delighted to announce we have awarded two grants totalling £210,000 to the University of Manchester to begin the first stage of the Genistein clinical trial, recruitment for which begins this autumn. In starting the trial whilst still securing the remaining funds, it is recognised that children with Sanfilippo do not have time on their side.

This clinical trial has come about because of dedicated funding and support of pre-clinical research on Genistein and Sanfilippo disease over the past six years funded primarily by the MPS Society in the UK.

On behalf of the Board of Trustees, the MPS Society would like to thank its members, Share A Gift and the National MPS Society in the USA for their support in enabling us to start this important clinical trial.

There is still much to be achieved and the MPS Society and its members are committed to continue fundraising for the remaining £200,000.

It is anticipated that further awards will be made as donations are received and as fundraising continues. **C Lavery**

Gene Therapy for MPS IIIA

Lysogene is a sole clinical stage biotechnology company working on Sanfilippo A disease.

The SAF–301 AAV gene therapy product developed by Lysogene has recently successfully undergone a clinical trial in France involving four young patients affected with the disease.

This clinical trial P1-SAF-301 is an open label, single arm, monocentric, phase I/II clinical study evaluating the tolerance and safety of intracerebral administration of adeno-associated viral vector.

Preliminary data collected during the clinical trial support an excellent safety profile from a product and a surgical perspective. The study's principal investigator, Dr Marc Tardieu, head of neuropediatry, Le Kremlin-Bicêtre in Paris will present full data for this study during the annual meeting of the European Society of Cell and Gene Therapy at the end of October 2013.

Additional information about this clinical trial can be found at http://clinicaltrials.gov.ct2/show/NCT01474343

LYSOGENE

Behavioural phenotypes of the MPS disorders - A systemic literature review of cognitive, motor, social, linguistic and behavioural presentation in MPS disorders

E.M. Cross – D.J. Hare

Materials and methods: MEDLINE, PSYCINFO and Embase databases were searched, alongside manual screening, to identify relevant literature. Papers were included in the review if they were published in a peer reviewed journal and conducted empirical research into cognitive, motor, social or linguistic development or behaviour in one or more MPS disorders.

Results: Twenty five papers were reviewed. Two papers used methodology of a sufficiently high standard to demonstrate a behavioural phenotype; both found sleep disturbance to be part of the phenotype of MPSIII. Fearfulness and sleep disturbance were frequently observed in people with MPSI and MPSII. Cognitive and motor impairment and decline, and challenging behaviour were highly prevalent in the severe form of MPSII. Cognitive decline and severe behavioural problems relating to aggression, hyperactivity, orality, unusual affect and temper tantrums were seen in MPS III.

Conclusions: Sleep disturbance is part of the behavioural phenotype of MPSIII and challenging behaviour is highly prevalent in MPSII and MPSIII, therefore the efficacy of behavioural interventions for these populations should be investigated. Further research into the behaviour and adaptive skills of children with MPSIII and MPSIV is required.

J Inherited Metabolic Disorders (2013) 36:189-200

GM1 Gangliosidosis and Morquio B Disease

GM1 Gangliosidosis and Morquio B disease both arise from beta-galactosidase deficiency. They are very rare Lysosomal Storage diseases with an estimated incidence of about 1 : 200,000 live births worldwide.

GM1 Gangliosidosis is a neurodegenerative condition for which three main clinical forms have been identified: type I (infantile), type II (late infantile / juvenile and type III (adult).

Morquio B disease is a Mucopolysaccharide disease also known as MPSIVB that is associated with significant skeletal changes, corneal clouding and impaired cardiac function.

In recent times, particularly since ERT has been in clinical trial for MPSIVA families have been asking for clarification on differences. In a NIH Public Access Paper Biochim Biophys Acta. 2011 July; 1812(7): 782-790. doi:10.1016/j.bbadis.2011.03.018. the authors set out to address the problematic partition between Morquio B and Juvenile GM1 Gangliosidosis phenotypes using mutation analysis.

The author's conclusions were as follows:

"The tertiary structure of human GLB1 has not been resolved and only a previous homology model of human GLB1 was derived from the structure of the Bacteroides Thetaiotaomicron GLB1 protein has been reported [21]. Here we have used two structures of GLB1 from Penicillium and Bacteroides to produce and improved homology model of human GLB1.

Three-dimensional analysis and in silico outputs of mutated GLB1 proteins are helpful tools in defining patients phenotypes. However, a clear-cut phenotype classification between GM1 types I, II, III and Morquio B can be difficult. The considerations raised from the clinical and genetic assessment of our patients' cohort together with the description of a neurological Morquio type B form [6], and with previous evaluations on the convergence between the different forms of GM1 gangliosidosis and between GM1 gangliosidosis and Morquio B [7,22,25,34,36], warn physicians about the complications of defining disease severity in each case, and therefore of recommending any treatment that may be available . Polymorphisms could also play interesting roles on resulting enzyme activities and/or phenotypes.

At a glance, a continuum of phenotypes can be remarked in all carefully examined patients in whom GLB1 enzyme activity is deficient."

Acknowledgements
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Some samples were obtained from the "Cell Line and DNA Biobank from Patients Afected by Genetic Diseases" (G. Gaslini Institute) – Telethon Genetic Biobank Netwprk (Project No. GTB07001A).

Medco Health Solutions

We have recently learnt that Medco has made the decision to explore strategic alternatives for non-core parts of its pharmacy benefit management business. Express Scripts is considering the closure of its Northampton-based subsidiary, Medco Health Solutions Limited and is continuing to explore whether the business can be transferred.

A collective consultation has commenced with those currently employed by Medco Health Solutions Limited.

We understand Medco Health Solutions Limited is continuing to work closely with NHS England Highly Commissioned Services to deliver the highest quality service to its patients and will work with these parties to transition the care for these patients to other companies safely and securely.

If you are affected by the closure of Medco Health Solutions please do contact your LSD Specialised Centre or the MPS Society. If you are about to onto ERT with Medco as your healthcare provider you may wish to consider starting with another provider from the start. **Christine Lavery**

Preimplantation Genetic Diagnosis

From April 2013 Preimplantation genetic diagnosis (PGD) has been nationally commissioned giving our members across England an equitable and fair service that is no longer subject to the post code lottery.

The MPS Society working with the Genetic Alliance UK has regularly provided the Human Fertilisation and Embryology Authority (HFEA) with statements of opinion supporting PGD for specific MPS licence applications for PGD.

Over the past few years a number of our families have benefited from PGD, enabling them to have a child that is free from MPS or related Lysosomal storage disease.

If you would like further information on PGD please do ask your LSD Specialist, geneticist or seek the help of a member of the MPS advocacy team.
Christine Lavery

Review of Welsh Appraisal Process

The Welsh Government announced a review of their appraisal process for orphan and ultra-orphan medicines earlier this year. Mark Drakeford, Minister for Health and Social Services requested a group be established to review the appraisal of orphan and ultra-orphan medicines in Wales. On August 27th three patient organisations were invited to present oral evidence to the review panel regarding access to funding for treatments, the experience of patients that had experienced delays from the local health boards for processing funding applications and the opportunity to share experiences (good and not so good) and identify systems that work well elsewhere and what makes this happen.

On behalf of the UK LSD Patient Collaborative, Tanya Collin-Histed, Chief Executive 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 (ERT) and Substrate Reduction Therapy (SRT) compared to England where patients are able to access these treatments through the LSD highly specialised service, if they meet the eligibility criteria.

As a result of Tanya's involvement in the meeting at the Welsh Assembly in August, she will continue to represent the Collaborative by providing advice and feedback on draft polices and being a member of implementing the Rare Disease Strategy for Wales.

12th International Postgraduate Course on LSDs

Nierstein, (Mainz) Germany, 3-7 June 2013

I was delighted to be invited for a 12th time by Professor Dr Michael Beck to give the patient and patient organisations perspective of Living with a Lysosomal Storage Disease. Each year there is an enthusiastic group of postgraduates from many corners of the world. This year was no exception and over 5 days the post grads learnt everything there is to know about cell biology, pathophysiology, lysosomal membrane defects, the blood brain barrier, clinical management and treatment of lysosomal storage diseases from a very distinguished international faculty.

Despite Prof Dr Beck's forthcoming retirement I was delighted to learn that Prof Dr Beck will be organising a 13th International Postgraduate Course of Lysosomal Storage Diseases during the first week of June 2014. If there are doctors or scientists working in the LSD field or interested in working in the LSD field this masterclass is a must. Over dinner on the last night as each post graduate was awarded their certificate members of the faculty observed that we have never had any doctors from India on the course.

If you know anyone interested in participating in the 13th International Postgraduate Course of Lysosomal Storage Diseases in June 2014 please do let me know and I will pass your details on. **Christine Lavery**

Fundraising

Our Community - An unstoppable lot!

You've held Wicked Walkabouts, fun days and quizzes. You've worn blue, run marathons and baked cakes; you've even fallen out of planes all to make a difference to the lives of MPS families.

A big thank you to all of you who have fundraised for us. On the following pages you will find some of your stories.

If you are still looking for some inspiration for fundraising ideas here are 10 things you could do. Together we can make a difference...

- Its **Back to School** time - Would your local school like to support the MPS Society? We've lots of great ideas and downloads available on our fundraising section at **www.mpssociety.org.uk**
- **Wear it Wicked** at Halloween - A great time of year for some 'fangtastic' fundraising, email us for a poster **wickedgenes@mpssociety.org.uk**
- **Get Wicked at work** - Nominate the MPS Society as your Charity of the Year, have a Bake Off, ask your company about payroll giving.
- **Get Wicked at Uni** - New terms, new RAG teams, please nominate the Society for support.
- **Get Wicked in your community** - Hold a family fun day, or pub quiz
- **Run, walk, swim or cycle** for MPS - Great Northern/Southern runs, London marathon, London to Brighton Cycle plus many more!

- **Wicked Walkabout** – Honestly it's easy to set up, great fun and helps to raise local awareness.
- **Donate** - regularly or one off to MPS - Set up a standing order, direct debit, set up a legacy, donate in lieu of presents. Regular giving enables the Society to plan ahead.
- **Recycle** - Phones/stamps/cartridges, every penny counts.
- **Wear it Blue, Wear it Wicked, Wear it anyway you want, let's just put the MPS Society on the map!**

Priory School raises £1750

Thank you to the Priory School who have raised £1750 for the MPS Society. The cheque was presented to Ita Vickery at the school's final assembly. The School have supported the MPS Society for many terms and hopefully will do for many more. "The Head of the Juniors, Mrs Clark, has been a driving force behind the fundraising since she met and taught Luke (MPS IV) in Year 2. Sadly Mrs Clark retired last week after 28 years teaching. Both Luke and I will miss her greatly. She is a great teacher and friend", writes **Ita Vickery**.

Countrywide Admin Centre supports Wear It Blue

I went to bed just before midnight on Tuesday 14th of May after finishing decorating buns and cakes for the fundraiser to be held at work on Wednesday.

The day started with a sale straight away of a fresh cream fat free sponge cake! - the dieters dream and went from strength to strength when people turned up with further donations of bought and homemade goodies including an American style cheese cake which I dished up and sold within about 4 minutes!

The day progressed with a boost in sales over the dinnertime and break time periods and I answered many questions after people had read some of the information I had displayed.

People kept trying to congratulate me for my efforts but all I wanted was to raise awareness and raise some much needed funds for the Society after they have been so helpful and supportive of my family.

I was so pleased when we had more than doubled the amount raised last year as my place of employment has become a national administration centre and the generosity of my colleagues has really touched my heart. The final amount raised for the day was £206.76. **Elizabeth Heath**

Marina and Friends reach £100,000 - a Reason to Celebrate

A day to remember – Tuesday 11th June 2013 – The day of 'Grandma's Do'. The day when we publicly acknowledged that Marina Foster of 'Marina and Friends' charity shop had achieved the magnificent feat of raising in excess of £100,000 for research into Sanfilippo disease (MPS III) since opening her shop in April 2003.

Marina's incredible fundraising journey began in 1996 following the diagnosis of her beloved twin grand- daughters, Francesca and Josephine, with Sanfilippo disease in 1995 aged 3 ½.

Marina with Francesca and Josephine (MPS III)

One of the words in the girls' vocabulary was 'Grandma' when referring to Marina, hence she became known to us all as 'Grandma' and still is.

It all started suddenly when 'Grandma' declared to her family that she was 'going to do a car boot sale to raise some money for research'. Quite a surprise because not only was she not a driver, she did not have a car! However, 'Grandma' desperate to do something to help recruited her husband and brother and set about 'car booting' at least twice a month with all proceeds going to the MPS society and specifically towards Sanfilippo research. Their car boot efforts raised in excess of £22,000 over seven years.

In April 2003 Marina took the opportunity to open a small independent shop in Sandy Park Road, Brislington. A courageous move as she only had her car boot experience and very little stock. Ten years on, Marina who is 78, runs the shop with a small dedicated team of volunteers and is extremely grateful for the support the local community has shown, both as providers of stock and as customers.

Francesca died in 2007 aged 15 and Josephine in 2009 aged 18, leaving the family devastated. On top of this Marina also lost her husband in 2011. It has taken great courage and determination on her part, assisted and encouraged by the efforts of her voluntary team, to keep the shop open.

'Grandma's Do' had its beginnings in January 2013 when a cheque sent to the MPS Society brought the cumulative total donated from the shop just a few thousand pounds short of £100 000. We felt that 'Grandma's' hard work and dedication should be recognised and decided to arrange a surprise party at 'Marina and Friends' shop once the magic figure had been reached.

Marina sells donated clothing, bric-a-brac, toys, books and other items for small amounts of money – skirts/ tops/ trousers can be bought for 50p - £1, T-shirts – four for £1, paperbacks 20p each or 8 for £1. Some things cost more, for example a pushchair or children's bike for around £5 - £10. However, these small amounts have amassed to the very significant figure of £100 000. Over the years 'Marina and Friends' has become well known in the local community as the place where you can get almost anything and always at a bargain price. The shop is tiny, so goods need to be sorted and sold without delay. Since all the stock is donated, customers never know what they will find and this leads some of them to visit daily or even several times a day. As well as finding a bargain, there's always a warm welcome and someone to chat to.

By late April 2013 we were able to start turning our plans into actions. We contacted the MPS Society to discuss a special celebration certificate and Christine Lavery kindly offered to come along to present it. The date was set for Tuesday 11th June 2013 and fingers were firmly crossed that the day would be fine. It was essential that all the plans remained a secret as Marina doesn't like any fuss and may have refused to attend if she'd found out about it!

We enlisted neighbouring shop keepers to help and they were wonderful, providing storage facilities, decorations and forecourt space. We also approached the Avon Veterans Brass Band. When they heard about Marina's amazing achievement they were keen to help us make the day go with a swing. Behind the scenes, Marina's volunteers issued invitations and tidied the shop. Everyone involved kept the secret – Marina didn't have a clue.

Marina and her daughter Julie,(Fran and Jo's mum) always spend Tuesdays together, so the plan was to pick 'Grandma' up as usual then to bring her to the 'Do'. Beforehand, some of the volunteers decorated the shop with banners and balloons. We brought refreshments and the party essentials, setting everything up outside the shop. Chris, (Fran and Jo's dad), welcomed the band members and greeted Christine Lavery and Wilma Robbins while Julie went off to pick up her mum. Meanwhile an excited crowd started to gather, everyone keen to support Marina on her special day.

Everything went to plan, Julie arrived with Marina right on time, with the brass band striking up as they approached the shop. The throng of people focused on Marina and it was obvious from the expression on her face that she was bewildered as to what was going on. At this point Christine Lavery approached and as Marina recognised her they exchanged a warm embrace. Others moved in to give hugs and greetings and it took some time before we could usher Marina towards the decorated shop window and provide her with an answer as to what was actually happening!

Chris spoke to the supporters who had gathered, announcing that 'Marina and Friends' Charity Shop had actually donated £103,066.15p and that he was delighted to introduce Christine Lavery, Chief Executive and Founder of the MPS Society. Christine spoke about the importance of research into MPS conditions, particularly mentioning current research into Sanfilippo disease, and how grateful the MPS Society is for Marina's ongoing financial support. She then presented Marina with a framed certificate and a bouquet of flowers. By this time everyone was very emotional, especially 'Grandma' who was in complete shock!

Following applause, the band played 'Congratulations' – a very fitting tune.

Various photographs were taken including one of Marina with some of her team of volunteer helpers. They all work so hard in the shop to support Marina's fundraising efforts. They are wonderful and we were pleased to share the celebrations with them and to thank them. Hopefully Christine and Wilma enjoyed meeting them.

As we enjoyed refreshments and celebratory cake there was a lot of excited chatter and sharing of stories about Marina and her shop. As the band played and everyone relaxed, it became clear that Marina is loved and respected for all that she does. She really is at the heart of the community. Her enthusiasm, demeanour, dedication and determination have attracted many admirers and have been instrumental in attracting funds to the MPS Society in other ways. For instance, people have run marathons, half-marathons and 10ks, groups have been inspired to make the MPS Society their Charity of the Year, donations have been given in lieu of wedding anniversary gifts and a former customer gifted money to the MPS Society in her will. Marina has given out hundreds of leaflets and has raised the profile of the MPS Society in her locality.

It should not be forgotten that in addition to the money Marina has donated, she has also had to raise money to fund her shop rental, insurance, council tax and utility bills, altogether in the region of forty thousand pounds, making her total fundraising in excess of £165,000. That first car boot sale has turned out to be like Mary Poppins' bag – BOTTOMLESS!

In May 2012 Marina was invited to the MPS 30th Birthday Celebration at the Palace of Westminster, where she was thrilled to meet Dr. Brian Bigger who is currently undertaking research into Sanfilippo disease. She enjoyed an informative discussion with him about his work and this reaffirmed her commitment to continue her fundraising efforts for as long as she can. What a positive way to express her love for Francesca and Josephine and to honour their memory.

Our love and respect for her made the celebration of these achievements particularly special as we are Julie and Chris, parents of Francesca and Josephine and Marina's daughter and son-in-law.

She may be our 'Grandma' but we have been truly inspired by her and trust that you are as well.

Chris and Julie Kembrey

Marina with her certificate and fantastic team of volunteer helpers outside her shop, Marina and Friends

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. So far, the cumulative total raised by Marina and Friends for research into Sanfilippo disease is **£107,291.66.** 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.

Will you be part of our Wicked Walkabout?

When Wicked Walkabouts was first launched we hoped it would be the beginning of something Wickedly wonderful and we haven't been disappointed.

Opening your emails with pictures of families and friends coming together in Derry, in Bucks, down in Dorset and across Surrey we have been inspired. Rain or shine, you have toughed it out to Wickedly Walk around your local communities raising money and awareness for MPS and related diseases.

This summer the fantastic Kath Hiller held a Wicked Walkabout with friends and family from the beautiful Bridport to West Bay. Meanwhile, in Surrey the unstoppable Steve Brooks had teams of 3-legged racers, Wickeding their way through a Surrey pub crawl. (A special mention should go here to the Surrey supporter who donned a rather revealing Mankini - all in the name of charity!)

If you think you'd like to hold a Wicked Walkabout in your local community then please drop us an email **wickedgenes@mpssociety.org.uk** and we can send you our Wicked Walkabout Guide, a simple, easy to follow collection of suggestions to get the most out of your event.

Watch this space for more Wicked Walkabout news and pictures.

Bridport Wicked Walkabout

Kath Hiller, family and friends on their Bridport Wicked Walkabout

As a family we wanted to support the MPS Society by organising our own "Dorset Wicked Walk" on 19th May. We thought we might raise £100 - £200 but were completely overwhelmed by the wonderful response we received from family, friends and work colleagues.

The local parish magazine and the Bridport News published articles about our dear Joseph who was diagnosed with Hunter Disease (MPS II) at the end of last year.

Donations rolled in and about 25 friends joined us on a drizzly but warm morning to ramble down the old railway line from Bridport to West Bay (Broadchurch in the TV series) and back across the fields to our starting point.

We were delighted to discover that the total amount raised exceeded £2000!
Kath, Terry and Louise Hiller

Frimley Green Wicked Walkabout

If you were walking through the village of Frimley Green on one of two consecutive Saturdays in August, you may well have thought some tom foolery was amidst. With 80 pairs of seemingly grown adults bound at the leg and taking a well-deserved pint or short at each and every pub on this warm summer's day in the Frimley Green and Mytchett area. But this was not some 'stag' do or 'hen' party and it was certainly a bit early for celebrating Oktoberfest, but it did have a deserved meaning to these antics. It was the first annual 3 legged race for the MPS Society, a charity event which raises money for research, awareness and understanding for children and families who suffer from one of several mental and physical degenerative diseases of these relatively unknown conditions which the Society akins its self too.

The event was born from what was a seemingly simple conversation down at the local pub on the green, the 'Rose and Thistle'. A few locals had gathered as they usually do for their weekend 'ritual' for one of the locals to say that his son suffers from MPS II, 'Hunter' disease. This created curiosity and some frustration at the fact that everyone in the group was unaware of 'Hunter' disease and the fact that all this time they were unaware that one of their own had a family member who suffered from this degenerative disease.

From this conversation endeavour was born to highlight these conditions and for a community to show their true worth as several of the 'locals' whom also happened to run local businesses put their minds together to come up with an event that would raise awareness and raise money for the MPS Society and thus we have the first annual 3 legged race. For several weeks before the event Idesk proprietor Steve Brooks, one of several of the 'locals' down the Rose and Thistle diligently gathered support with the help of others, signing up teams for the race with the entry money going towards the MPS Society, by race day 80 teams had entered this real local event. Due to legal issues and the overwhelming support for the event the race start had to be staggered with timings finally determining the winner. The route was The Rose and Thistle, Kings Head, Potters, Miners and back to the Rose and Thistle the whole day and final presentation was 'electric' and heart-warming to see.

Now most would believe due to the support shown to this event that may well have been enough to highlight and make aware the MPS Society and the conditions it stands for, but no more was to follow. Parallel to the organisation of the 3 legged race was a further event, the MPS ball held at the Rose and Thistle pub on the green. With thanks to the Management Owen and Sarah of the pub, it was closed for one night only for an all ticket event to continue to raise money and awareness.

The ball started at 6pm to the sounds of guests arriving to Dixieland Jazz, this was the first 'eye turner' of the evening, as people strolled past the pub looking to see what was going on, you could have quite easily have been in the summer sunshine of New Orleans. Once again the 'Dixie' band was 'donated' by one of the 'locals' who just happened to play in a 'Dixie' band. However the evening did not stop there, it was just getting started with a whole evening of entertainment set up for this incredible cause. The organisation was outstanding and again the atmosphere 'electric'. Along with the ticket was a superbly catered for choice of curries for those moments when you felt peckish, arranged and overseen by James Tucker local businessman from Britcar Racing and the outstanding Chefs and management of the Rose and Thistle, all served outside under a marquee and outside catering unit of Britcar racing and washed down with a live Cabaret.

As the evening progressed and the sun went down even more entertainment was due with a live band playing both original and 'cover songs' to keep the party moving. This was shortly afterwards followed by a Casino night in the Conservatory for those who were feeling lucky which continued until close in the early house of Sunday morning.

All in all an amazing and outstanding two weeks of events which really shows how a community can pull together for a good cause and a 'local' cause. I for one will eagerly await next year's events for what should be an annual event. Money and awareness is still being raised with the total currently at £7500.00 and rising. If anyone would like to donate please go to www.mpssociety.org.uk
Frimley Green Supporter

Events and Challenges

London to Brighton cycle ride

MPS Society Trustee, Jessica Reid and her partner Alex Kafizas and friends Krystle O'Meara and Rob Pile cycled London to Brighton on Sunday 15th June 2013 in aid of the MPS Society.

This was a very challenging route of 55 miles, including the epic Ditchling Beacon towards the end of the course - which Jessica and Krystle managed to beat the boys on and cycle up rather than join the others walking! The cycling was completed in 4 hours and 38 months after many months of hard training.

Jessica lost her older brother to MPS II in 1986 and was cycling in his memory.

Jessica raised £765 plus gift aid, and Alex raised £620 plus gift aid, but because he works for Lloyds TSB they generously matched the first £500 of his sponsorship - so collectively the total raised was just over £2,000. Not bad for a bit of peddle power!

Cycle ride in memory of Mark

Hi, my name is Sarah Ashworth and on the 14th July I rode the Manchester to Blackpool bike ride with my friend Nicola Tyrer and uncle Tony Postle. We decided to do the ride, and to raise money for MPS, in memory of my brother Mark.

Mark was diagnosed at the age of 11 years old with mild Hunter disease and XYY Syndrome, Mark was referred to Professor Ed Wraith and we were told Mark was one in ten million to have both conditions. Mark visited Pendlebury Children's Hospital for regular check ups and also had holidays with the MPS society at Primrose Valley, where we met many families with children with MPS diseases.

Mark passed away on 2nd October 2006 and is loved and remembered by his family everyday.

In total we raised £442.00 for the MPS Society to help with the research into treatment and therapies.

Thank you to all our fundraisers who raised money through justgiving. Here are a selection...

Megan Hooper did a sponsored 10k run in May raising £121. http://www.justgiving.com/MegansMiniMarathon

Michelle Brooker and family and friends who raised £1518.50 by running in the British London 10k in July. http://www.justgiving.com/Michellebrooker2

Caroline O'Driscoll's page for Erin's Baptism has raised £540 http://www.justgiving.com/ErinsBaptism

Scott Wilkins ran the Virgin London Marathon and has raised £678.20 http://www.justgiving.com/scott-wilkins2013

Jeni Abbotts ran the Manchester 10k raising £276 http://www.justgiving.com/jja

Martin Balestri raised £ 1675 from a sponsored 62 mile walk from London to Brighton http://www.justgiving.com/Ella-Bella3

Chloe Collins and friends climbed the Three Peaks and Brecon Horseshoe for MPS raising £515 http://www.justgiving.com/Chloe-Collins1

Thank you to the **DC10 Yorkshire 3 Peaks Team** who walked 25 miles in 12 hours across Yorkshire 3 highest peaks. Pen-y-Ghent - Whernside - Ingleborough on 27th July 2013. They raised £650 on their Justgiving pages.

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!

Lauren's skydive and Colour Run

Lauren Thompson is good friends with Oliver Moody (MPS VI). Lauren and friends did The Colour Run and Skydive recently.

She writes: 'These were both amazing experiences. The Skydive was so insane, after conquering the nerves just as I was getting my head round it we travelled 10.000ft into the sky above the clouds and soon the feelings were nauseousness and what am I thinking haha! After the initial freefall to 5.000ft it became an incredible experience and the views were out of this world! I am so happy I did it!' Lauren raised £580 on Justgiving.

12 hour Darts Marathon

The marathon was held on 29/6/2013, in which my brother Dan, father Martin and myself competed in.

It was a fantastic day with support and generosity from so many people. We started at 11am and went right on through until 11pm without a break.

People bought raffle tickets throughout the day and we managed to raise almost £200 through this. People were also very keen to donate to our charity bucket, raising well over £500!

We had the local paper come down to take some photos and a write up of the day way published in the paper the following week.

All in all, including the donations made on my Just Giving page, we managed to raise a grand total of £1300 through the event which we are really proud of.

We are now hoping to make this an annual event as we were overwhelmed by the amount of support we received.
Mat Sims

Nick's bike ride

Grace, our daughter was born on 11th January 2007, diagnosed with the genetic disorder MPS VII Sly disease.

We were told her outcome was very poor and that she would be still born or live anything from a few minutes to a few hours. We praise God that Grace was a big part of our family for 5 weeks, but then sadly died 17th February 2007.

For their support and in memory of Grace and the ongoing research into MPS disorders, we would like to raise a little bit of cash from Nick's little bike ride. Nick raised £378.50 on justgiving.

Danceathon at Meadows Primary School

Here at the Meadows Primary School we held a Danceathon to raise money for the MPS Society on behalf of one of our much loved pupils, four year old Emily Bradshaw and her family. As Emily loves to sing and dance to her favourite music we felt a sponsored Danceathon would be most fitting and lots of fun.

Pupils and staff came to school dressed in outfits based on different types of dance styles. Throughout the whole afternoon pupils were dancing to a variety of music. Prizes, kindly donated by Phones 4U, were awarded to the best dressed, best dancers and pupils who raised the most money. A grand total of £632.39 was raised.

Emily looking very proud in her school uniform

Thank you to all our donors including . . .

Nailsea Lions Club held a 'Cream Tea' afternoon raising £400.

Jenny Dagnall raised £1,600 from donations and the proceeds of a garden party. This money is to go towards the MPS I post HSCT ERT trial.

Ann Parsons donated £83 being the proceeds from the sale of trolley key rings.

Sophie Longley raised a further £320 sponsorship from her London Marathon 2013.

Freemasons who are members of Mercurius Lodge L4262 kindly donated £1050.

Savvy Financial Planning raised £50 towards the Genistein MPS III Clinical Trial and £1,820 from a parachute jump.

Draylow Park Golf Club held various golfing days raising £300.

Luke Blignaut's school, All Saints Junior School, held a cake and coffee sale raising £426.50 for the MPS I Clinical Trial appeal. Ross Travel donated £100 to the MPS I Clinical Trial appeal as the owner's daughter is in the same class as Luke.

Dr Anil Bansal kindly donated £151 in memory of his son's birthday on 2nd August.

Carole Bradshaw donated £60 as her friend's baby has been diagnosed with MPS.

Barry Simner and the ladies of Tywyn Probus Club donated £50.

Mr Roger Lightfoot sold his car to GIVEACAR and donated £137.20.

Heaton School raised £130 in memory of Blaise Whittle a former student.

Graham & Danielle Warren raised £349 at a pub quiz.

Tony and Annie Walker of the James Walker Foundation donated £1000 to Project Sanfilippo.

Moyle Primary School, Larne raised £155 at their Wear It Blue event.

Sally Amos donated £25, the proceeds of a Wear It Blue Day from Ella's old nursery.

Sarah Miller sold £40 worth of MPS merchandise.

£435 was donated in memory of the late Mr Duncan Alastair Stevens (Wife Mrs Jill Stevens)

Next Retail Ltd donated gifts for the MPS Christmas raffle.

Gilchrist and Soames donated 260 small gifts for the MPS conference.

Trustee Wilma Robins generously donated £2000.

The Rotary Club of Yeovil donated £425 being the proceeds from a tea and coffee stall at the Kingsbury Episcopi Fayre.

The Lloyd's Under 30's Non-Marine Group donated £1000 being the profit from an event for the young people within the Lloyd's community.

Rashpal and Sandra Singh organised a family fun day in memory of their son Daniel who suffered from MPS II, Hunter disease. They donated £60 from the sale of MPS merchandise.

11 year old Ruby Barnes raised £82 for the MPS Society over lent by giving up sweets, chocolates, biscuits and crisps.

St Thomas of Canterbury R.C Primary School raised £333 with their Wear It Blue Day.

Alan Byrne retired from teaching and requested donations to the MPS Society in lieu of leaving gifts from colleagues. He raised £370.

Shirley Jamil Raised £260 at a pamper night.

Mrs Jean Devine donated £300 which was given in lieu of flowers after the passing of Jean's husband George.

St Thomas Primary school, Bury School raised £163 with their Wear It Blue Day.

Dorothy Robinson held a fourth craft day raising £200 for MPS.

Rentokil Initial donated £83.85 as matched giving in respect of Ashley Chattin.

Mr and Mrs J Keenleyside donated £100 towards support to children affected by Morquio. Their neighbour Mr R Webber has two grandchildren affected by MPS IVA.

Waitrose, Amersham selected MPS as one of their local charities to support and raised £200.

Lauren Hartwell and her fiancé, Gio donated £150 in lieu of wedding favours at their wedding.

Marie Shepherd and colleagues at Barclays raised £450 at their Wear It Blue event for MPS Awareness Day.

Dave Joyce, manager at Oceania Nightclub, Brighton raised £2268 for the Genistein Trial.

Barnfield College in Luton raised £147 at their Fashion Show.

Amicus Therapeutics in the US raised $62 (£39.22) at their Wear It Blue event for MPS Awareness Day.

Ian Holloway at TJX donated £1,500 to MPS.

Phil Lewis from Velocity donated £1250 for the Genistein Clinical Trial.

Martin Durbridge raised £2470 for the Genistein Clinical Trial.

Brynglas Primary School in Newport raised £41 at their Wear It Blue day.

Students at St Elizabeth's School in Hertfordshire did a sponsored bike ride raising £144 for MPS. They have a young boy at their school who has Sanfilippo.

Mark Hughes raised £410 with his family at the Arbury carnival.

Great Sankey High School in Warrington raised £700 from their Wear It Blue Day.

The children and staff in foundation Stage and year 1 at James Elliman Academy in Slough raised £206.61 at their Wear It Blue event for MPS Awareness Day.

Bedonwell School in Kent (Bobby Gill's old school) raised £121.99 for the Genistein Trial from a collection after a school play.

The Staff Consultative Committee at Teva, Runcorn donated £200 for the Advocacy Service.

A friend of Jenny Hardy recently got married and donated £10.

Christine Bishop donated £50 to say thank you to the Advocacy team for their recent help in winning her ESA tribunal.

The Rotary Club of Durham donated £285 and kindly presented the cheque to Peter Conlin, father of Ben (MPS I).

Mrs Eira March donated £100 from the sale of knitted toys. She chose to support the MPS Society because of Archie Pearson.

Yvonne & Phil Pearson recently renewed their wedding vows and asked guests for donations to the MPS Society in lieu of gifts, they raised £600.

£200 was donated by a friend of David Gosling.

Savvy Financial Planning donated £190 to the Genistein Clinical Trial.

Louise Lucas at the Hampden Arms ran a guess the weight of Princess Kate's baby competition for the MPS Society raising £20.

Glyne Gap School held a cream tea raising £200. There are two pupils at the school with MPS.

Martin Balestri raised £815 with Wicked Genes.

Shirley Bown's friend Lyn held a fun day for Sanfilippo research and raised £215.

Jonathan Bayliss donated £100 in lieu of a gift James Garthwaite's birthday.

Vanessa Stottor at Be A Butterfly donated £150 raised at evening for the MPS Society.

Shire Pharmaceuticals Group donated £500 raised at their Wear It Blue event for MPS Awareness Day.

Claire Waters donated £50 in lieu of a gift for James Garthwaite's Birthday.

Julie Kennett donated £50 towards the Simms Family Fun day held in aid of the MPS Society.

Penny von Spreckelsen donated £50 in lieu of a gift for James Garthwaite's Birthday.

Sullivan Upper School in Co Down, Northern Ireland donated £1100.

Jacqueline Mount ran the Glasgow Women's 10K and raised £230.

Project Sanfilippo

Project Sanfilippo Family Fun Day - thank you to all involved!

Project Sanfilippo was developed by Karen Robinson, mother of Oliver who has Sanfilippo. Project Sanfilippo raises vital funds for the MPS Society to put towards research into Sanfilippo Disease.

Thank you to Karen Robinson and Project Sanfilippo for raising a fantastic £2560.90 from their Family Fun Day and Auction of Promises on Saturday 22nd June 2013. The MPS Society would like to thank everyone who helped to organise the event and supported Project Sanfilippo and the MPS Society. For more information please visit www.projectsanfilippo.co.uk

Joanne Humphreys organised a fundraising event including a raffle draw and the selling of artwork which coincided with her Open Studios event on 8th June. The event raised £100.

Wear it Wicked for some Fangtastic fundraising for the MPS Society

With Halloween just around the corner we want to get as many of you as we can Wearing it Wicked.

Halloween is a fantastic time of year to have fun and raise money and awareness for worthy causes. So we have lots of gruesome ideas for you...

You could hold a small ghoulish gathering with apple bobbing and pumpkin carving at home or perhaps you feel brave enough to hold a broomstick ball, or a deadly disco. You can even get Wicked at work or school and get everyone to donate a £1 and get fiendish with their wardrobe.

Here are some ideas...

- Donate a £1 and Wear it Wicked to work or school
- Hold a dusk Wicked Walkabout taking in local spooky haunts
- Guess the weight of the pumpkin
- Hold a fright night film evening and ask your audience to 'pay per view'
- Hold a Wicked cake sale with morose morsels

Email us at **wickegenes@mpssociety.org.uk** if you would like a Wear It Wicked poster. Don't forget to send us your fright night pictures and stories so we can share them.

Grants, CharitableTrusts, Grant-giving Foundations and Community Groups
Rotary Club of Mold; Rotary Club of Erith; Rotary Club of Wallasey; Rotary Club of Durham; The Albert Hunt Trust; The Griffin Club, Shauna Gosling Trust, The Reuben Foundation, Fitton Trust, Bernard Sunley Charitable Foundation;

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In Memory
Paul Franklin; Thomas Fisher; Miss Edith Doreen Boundy; Hilda Marks; Margaret Joan Newell

Collection Boxes
Ian Evans – Local Post office; L Barr

Stamps, foreign coins, mobile phones, ink cartridges; jewellery
Ian Evans and Arriva Shires; Susan Swayne; Karen Robinson; Your Derwent and Solway Housing Association; Liz Hardy; Nicola Drew and family; Maria Heap

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

Wear It Wicked this Halloween

This October we're **Wearing It Wicked** in aid of the MPS Society.

Join us in our **fangtastic** fundraising to support individuals and families affected by 24 Mucopolysaccharide (MPS) and related diseases. These are rare genetic conditions causing progressive physical and in many cases neurological deterioration for which there is no cure.

Over the past 30 years the MPS Society has been involved in supporting families, raising awareness and funding vital research in developing new treatments.

So please, Be Batty... Get Ghoulish... and Wear it Wicked this Halloween

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

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

Email wickedgenes@mpssociety.org.uk or check out our website to download our Wear It Wicked Halloween poster

MPS

support

research

awareness

Wicked GENES

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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