THE MPS MAGAZINE

Society for Mucopolysaccharide Diseases

Support Research Awareness

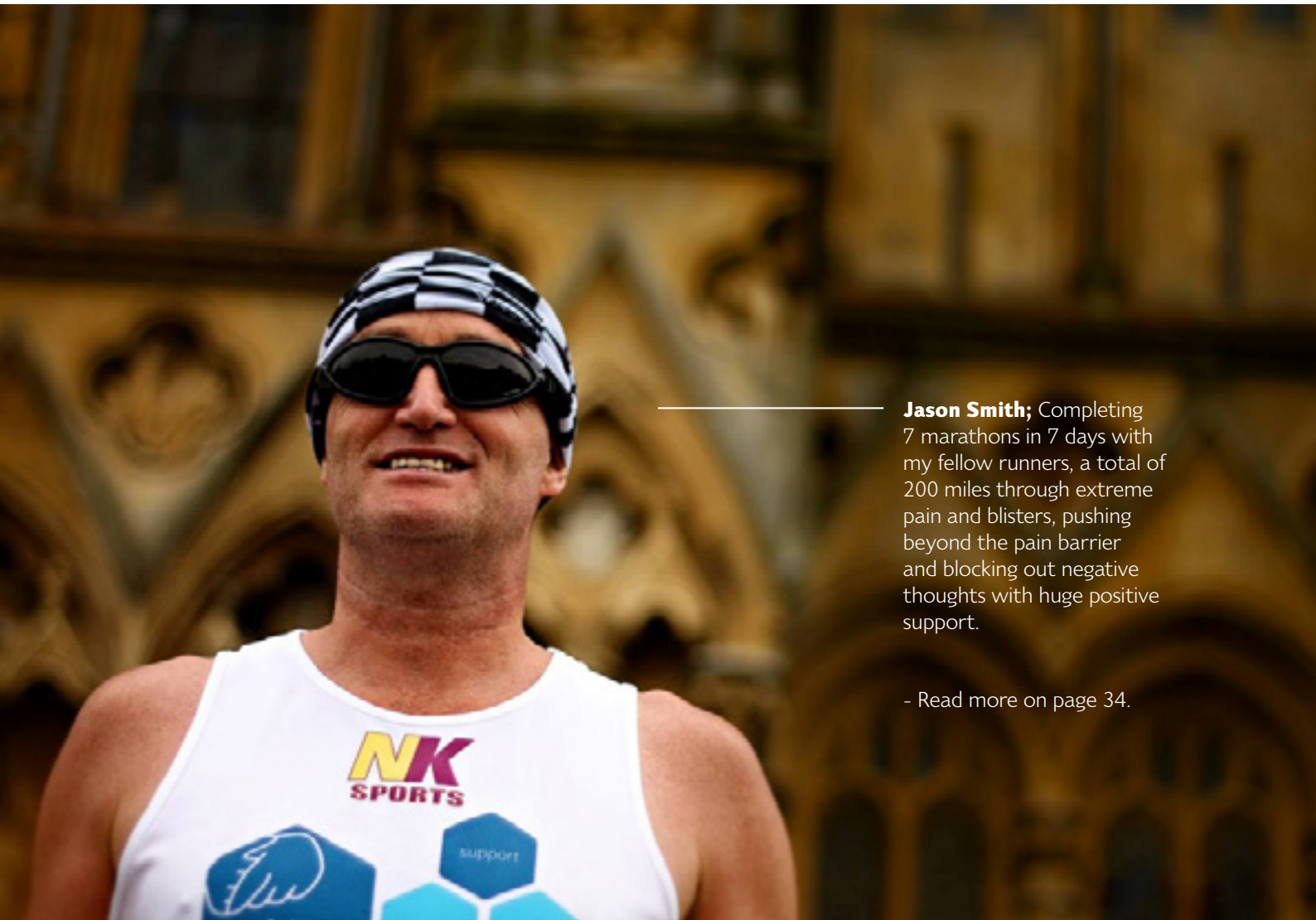
Autumn 2014

www.mpssociety.org.uk

13th International MPS and Related Diseases Symposium
Held in the idyllic location of Bahia, Brazil, the conference brought together sufferers, their families and professionals to share knowledge and experience. Our Ambassadors spoke to some prominent people and these interviews can be found on p38.

The Genistein Trial
Update on the clinical trial which aims to evaluate the clinical effectiveness of a treatment for Sanfilippo Disease. Researchers at The Royal Manchester Children's Hospital and The University of Manchester have recruited Jack Watson as their first child into a new study. You can read Jack's story on p57.

Wear it Wicked for MPS
Calling all witches, vampires, werewolves and mummies: get ready for some Halloween fundraising for the MPS Society! Whether you want to dress-up, bake some grisly treats or organise a Zombie Walk, we would love to see our supporters get Wicked! For some inspiration for the spooky season visit p36.



Jason Smith; Completing 7 marathons in 7 days with my fellow runners, a total of 200 miles through extreme pain and blisters, pushing beyond the pain barrier and blocking out negative thoughts with huge positive support.

- Read more on page 34.

Front cover photo: Jason Smith outside Wells Cathedral

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.

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Wear It Wicked!

Last year we saw a fang-tastic response to our ***Wear It Wicked*** campaign over Halloween, with our supporters getting into the spirit of things and becoming witches, wizards, werewolves, ghosts and all manner of ghoulies to raise funds and awareness for the MPS Society.

This year we would love to see more monsters! We've included an article in our fundraising section of this issue to help you ***Wear It Wicked,*** or you can always drop us an email at:

fundraising@mpssociety.org.uk

For more information or to request a fundraising pack.

The MPS Society

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

Our Aims:

To act as a support network for those affected by MPS and Related Diseases.
To promote and support research into MPS and Related Diseases.
To bring about more public awareness of MPS and Related Diseases

MPS and Related Diseases

Mucopolysaccharide (MPS) and Related Diseases affect 1:25,000 live births in the United Kingdom. One baby born every eight days in the UK is diagnosed with an MPS or related disease.

These multi-organ storage diseases cause progressive physical disability and in many cases, neurological deterioration can result in death in childhood.

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

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

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facebook.com/mpssociety

twitter.com/MPSSocietyUK

Welcome

With summer officially over, and the nights drawing in, we have ensured that we have included plenty of reading material to keep you all going!

As usual Christine's report highlights latest news from the MPS Society, touching on the subjects of the Brazil Symposium as well as the Genistein Trial – both of which are covered in their own articles further on in this issue.

Angela Paton describes her experience and ultimate effects of participating in the Elosulphase Alfa clinical trial, and how you can assist in securing future funding for this important drug for those suffering with MPS IV.

Our fundraising section is full of our supporters' wonderful stories, and we have offered some advice on how to support the MPS Society over the Halloween period by Wearing it Wicked!

Best wishes,

The MPS Team

Visit our online shop

www.mpssociety.org.uk.

Purchase our information resources and MPS merchandise including our T-shirt!

Chief Executive's Report

Christine Lavery

Sitting down to write this report, I can't but reflect on having just days ago returned from the biggest and one of the most successful MPS International Symposium's in their 24 year history, with over 1,000 registered delegates. This Symposium was held in Bahia in North East Brazil. I was joined in Brazil by five MPS young ambassadors, two members of the Advocacy Team and the Society's Chairman, Sue Peach. Everyone had important roles to play and you will read the fruits of our information gathering later in this MPS Magazine.

It was at the two day International MPS Network meeting of representatives of MPS Societies from 19 countries that preceded the Symposium, that after 10 years leading this International Network, I announced my intention to step down and enable others to lead. Lost in translation I have heard I have retired from CEO of the MPS Society! This is not so. Having said this, change has been afoot at MPS House over the past months.

After 14 years of total commitment to the MPS Society, Antonia Crofts has decided to make some personal changes and left MPS at the end of July. In the months leading up to her leaving she led a very successful recruitment process resulting in Charlotte Roberts joining as Communication Officer at the beginning of August and Mahboob Ahmed Khan joining as Business Development Officer on the 1 September. Antonia was a popular member of the Senior Management Team and both I and her colleagues are very sorry to see her go, we send her our very best wishes for the future.

This report would not be complete without offering a huge thank you to all those who raised money to fund the Genistein Clinical Trial for MPSIII at Manchester Children's Hospital. It has been a huge effort by the MPS Stem Cell Group led by Dr Brian Bigger at the University of Manchester and Dr Simon Jones, Consultant Paediatrician at Manchester Children's Hospital to bring the science to the clinic room. On Monday 11 August 2014 Jack Watson from Sunderland became the first child to be recruited to this important clinical trial. The downside is that with the National MPS Society in the USA withdrawing their second tranche of grant, £71,000 previously awarded, only 24 children can be recruited to the clinical trial instead of the planned 30 children. The MPS Society is still committed to raising the last £100,000 for the study but did not bargain on having to raise the extra £71,000. We would really want to have 30 children on the clinical trial as it will strengthen the science coming from the study and make the results of the clinical trial more robust. We owe this to the whole Sanfilippo community and ask for your help.

To conclude many of you asked for an update on clinical trials particularly those for Gene Therapy. My time in Brazil allowed me to catch up with many of the researchers and companies involved and the information is towards the back of this MPS Magazine.

Governance

News From the board of Trustees

The Society's Trustees meet regularly. Here is a summary of the main matters discussed and agreed at the Trustee Board Meeting held 4-5 April 2014 at the Copthorne Hotel, Manchester.

Governance

Apologies were received from four Trustees and one could only join the meeting on the 5 April 2014. No conflicts of interest were declared and following a welcome by the Chair the minutes of the Board Meeting held 24-25 January 2014 were agreed as a true and accurate record of the meeting.

The Chief Executive confirmed the Trustee Induction for one Trustee took place involving structured sessions and a Q&A session that proved very useful. Due to insufficient numbers of Trustees being available the Finance Awareness Training was rescheduled for the 4 July 2014.

It was agreed that a complete review of all the MPS policies take place over the coming months and progress would be reviewed at the next Board Meeting. The Chief Executive confirmed that an anti bribery policy was currently being drafted.

The Trustees discussed the matter of Board communications. The Chair proposed a draft Trustee protocol outlining professional standards expected of Trustees. It was agreed that this protocol be considered in greater depth at the next Board Meeting.

Treasurer's Report

The Treasurer presented her report acknowledging that the balance in the bank accounts is capable of supporting the Society's budgeted activities at this stage of the financial year. It was agreed the Finance Officer prepare a brief document outlining terms of the mortgage for the next Board Meeting.

Fundraising and communications

The Fundraising and Communications Officer updated Trustees on some significant sources of support and on the increase in the MPS Society's online audience. Approaches continue to be made to Universities and corporate donors as well as Charitable Trusts. Current emphasis is on the 'Wear It Blue' campaign for MPS Awareness Day.

Risk Management / Health And Safety

Two Trustees presented their progress working on the Business Continuity Plan (BCP). It was agreed that further discussion was needed regarding the critical box documents.

Clinical Management

The Chief Executive Officer advised that there were no matters that might necessitate changes to the Risk Management Register. The BCP is still under review and the Risk Register would in time become integral to the final Business Continuity Plan.

The Health and Safety Report was considered and agreed.

Advocacy Report

The Chief Executive presented her report highlighting particular points including the lack of progress on an independent LSD Registry; plans for the Fabry International Network Meeting that took place 10-13 April 2014; and the Shire Humanitarian Aid Programme.

The Senior Advocacy Officer's report was read and agreed.

What's On

MPS Regional Clinics

MPS Regional Clinics

MPS III – GOSH: 25th November 2014

MPS IV – GOSH: 28th October 2014

MPS I Post HSCT (under 6's) clinic - RMCH: 3rd October 2014

MPS I Post HSCT (under 6's) clinic - RMCH: 10th October 2014

Fabry clinic - BCH: 24th October 2014

Fabry clinic - QE Hospital: 11th November 2014

MPS I clinic - GOSH: 11th November 2014

MPS clinic - BCH: 14th November 2014

MPS I clinic - GOSH: 9th December 2014

Conferences and Regional Events

Scottish Family Weekend
17th – 20th October 2014

Childhood Wood Planting
26th October 2014

Lapland UK Family Day
3rd December 2014

USA MPS Conference – Florida
13th – 21st December 2014

UK MPS Conference Hilton, Coventry
26th – 28th June 2015

Welsh Information Day Marriott Hotel, Cardiff

Dates to be confirmed

Scottish Information Day – Beardmore Conference Centre, Glasgow

Dates to be confirmed

LAPLAND UK®
CHRISTMAS MAGIC FOR EVERYONE

Please see the flyer with this magazine for more information on our upcoming Lapland UK Family Day!

Announcements

New Faces at the MPS Office

Mak Khan

Hello, my name is Mak Khan and I've just joined the MPS Society as Business Development Manager to help Christine and her team run the Society's clinical trials programme.

After completing my doctorate in molecular genetics I spent a decade in medical research working in the fields of cardiac disease, transplantation and clinical diagnostics. This was followed by senior commercial roles both in the NHS for the Department of Health and in the pharmaceutical industry. These positions allowed me the opportunity to work increasingly with healthcare professionals, NHS commissioners and patient advocacy groups.

My next role with an affiliate of the World Heart Federation allowed me to work on international cardiovascular education and clinical trial programmes. Most recently, I have been working with a charity working in Libya to help provide medical training to doctors and surgeons across the country.

Outside of work I love to spend time with my three daughters and enjoy live music and going to the cinema. I've recently taken up Ceroc dancing and hope one day my skill level will match my enthusiasm.

I'm very happy to be joining Christine and her wonderful team at the MPS Society. The achievements of the Society and the stories of the families are inspirational and I hope my experience working in the NHS and the pharmaceutical industry will be an asset to the charity.

Charlotte Roberts

Hello my name is Charlotte and I joined the MPS Society mid-July as Communications Officer. I have only been in the post a short while but thanks to the friendliness of everybody at MPS House and our members I have been made to feel welcome and very much part of the team. So thank you!

It has been a busy month learning about the work of the MPS Society and the huge variety of assistance they provide and I am looking forward to getting out and about meeting our members.

Out of work I love travel and being outdoors, I enjoy playing Ultimate Frisbee and need to improve my fitness in preparation for a half marathon in September. One year I hope to run the marathon.

I am going to be involved with the production of the magazine so I will be keeping close eyes and ears on everything going on and so look forward to hearing from you soon.

Sad News

Very unexpectedly my husband Arjan passed away on Friday morning the 29 August 2014. Arjan was co-founder of VKS the Metabolic patient support group in the Netherlands.

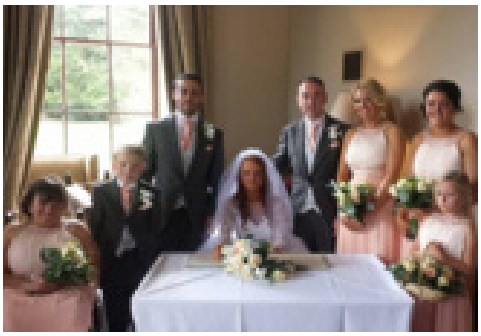
Hanka Meutgeert

Directeur VKS

Arjan and Hanka were together in Sauipe, Brazil for the MPS International Symposium only returning home two weeks ago making this news is all the more unbelievable. On behalf of the UK MPS Society we send our heartfelt condolences to Hanka, daughter Sitra and son Rick (AGU) and Hanka and Arjan's adopted children at this sad time.

Christine Lavery

See Arjan's interview on page 49



Congratulations
Mr and Mrs Watson

A big Congratulations to Mr and Mrs Watson on their wedding day – parents to Jack (MPS III) who looked very smart for this special day.

Births
Congratulations Sophie!

Older brother and sister Harvey and Imogen proudly showing off their new baby sister Freya Thomas, who was born on the 20th August. Congratulations to Sophie, our Senior Advocacy Officer, and her family.

New Members:

Mr Barnes has recently been in contact with the Society. His son Stanley has a diagnosis of Sanfilippo Disease. Stanley is 18 months old. The family live in the south west of England.

Jasmin and Gary have recently been in contact with the Society. Their son Riley has a diagnosis of Hunter Disease. Riley is two years old. The family live in Wales.

Arlene has recently been in contact with the Society. She has a diagnosis of Fabry Disease. The family live in Scotland.

Advocacy

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

STEVE

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

ALISON

Supports members living in Ireland

DEBBIE

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

REBECCA

Fabry
MPS II Hunter
ML III / ML IV
Mannosidosis Fucosidosis

MPS Christmas Cards

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

Please contact us for a Christmas Collection 2014 order form if you have not already received one.

Advocacy Feedback

The importance of Feedback

We appreciate how busy life can be and how much paperwork our lives are consumed with but please can we draw your attention to the importance of MPS feedback form following any communication with us.

The MPS Society is keen to hear feedback on how you have found this support, particularly what you found helpful or whether there are any recommendations for improvements or changes. Feedback is not only for our continuous improvement but more importantly so that we can demonstrate the work that we do in order to apply for funding applications and to respond to grants received. Without funding we wouldn't be here.

If you have lost your form please feel free to email your comments to info@mpssociety.org.uk and we look forward to hearing from you.

Thank you in advance for your support.

The Advocacy Team –
Sophie, Alison, Debbie, Rebecca and Steve

My Housing Journey

I suffer with Fabry disease and have battled with this for many years. In 2013 there was a sequence of events that turned my life upside down and now thanks to the help of the MPS Society I have moved in to my own accommodation.

I come originally from Morocco and having Fabry meant that my other siblings also had the disease. I feel lucky that due to living in the UK I can have my ERT to help with my disease.

By the end of last year I had lost all my siblings either to Fabry or cancer, the impact of which was devastating. In addition to this I was out of work and, getting divorced and was made homeless.

I was housed in a homeless hostel, in a tiny room with a shower and toilet where I had to remain for 9 months. During that time with the help of the MPS Society I was pushed up the waiting list as a priority. Without their help I would have remained in the hostel for much longer if it hadn't been for the persistence of my MPS Society advocacy worker. My mental health deteriorated and my advocacy worker got me the assistance that I needed, otherwise I don't know what I would have done.

I cannot thank the advocacy team enough for all the help and support they have given me. English is not my first language and at times I have struggled to understand the procedures.

Thankfully I am now in a flat and I have had the help to get furniture, again this has been through the MPS Society explaining about the disease and the problems that I have.

My healthcare nurse may still have to sit on the floor until the furniture arrives but at least he can turn around in the flat without knocking things over. My hostel room was so tiny that with the back pack on he could not turn round ...

I am now back to normal life and I am happy with my flat and life still goes on.

Latif

Bone Marrow Transplant Clinic

Rubina

Isac

Kiera

Sonny / Mikko

On the 4th July the Willink hosted a BMT clinic. After a very early start and a slightly more stressful journey than necessary (ticket office issues ... don't get me started!) I arrived bleary eyed at Manchester Children's Hospital. Jean and the team gave their usual warm welcome and soon families began to arrive. The clinic waiting area soon filled with children chatting and playing. Sonny and Mikko built the tallest possible tower with building blocks with help from the dads, whilst Jean nervously stood alongside ready to catch should someone fall.

The clinic was a great opportunity for me to meet with parent and carers and to offer the support of our advocacy services where required. We would like to thank the Willink Team for hosting the clinic and we look forward to the next one.

Steve Cotterell
Advocacy Support Officer

All Ireland

I am typing my article this quarter from sunny Bahia (Brazil) – a little different from my usual typing spot in Belfast City Hospital.

The team from the MPS Society have travelled across the world (it took a very long 28 hrs!) to attend the International MPS Conference; and so far we have not been disappointed. We are learning lots and meeting many influential people from the world of MPS. It is always really exciting to represent Ireland at events like this. This year, myself and Aidan Kearney (one of our ambassadors) travelled from Belfast and Mary Boushel (Irish MPS Society) travelled from Dublin. Between us we hope to bring back lots of knowledge that will benefit our MPS families across the island of Ireland.

I can't go any further without mentioning how sorry we all were that Dr Fiona Stewart was unable to attend. Despite making it to Heathrow her trip came to a sudden end thanks to a fuel leak. We missed you, Fiona! Hopefully you will have a more successful trip to the All Ireland conference in September – at least that one is close to home!

You will read lots more about Brazil in this magazine from our wonderful MPS Ambassadors.

Since June my time has been divided between providing individual advocacy support and planning for a few events that are coming up in the autumn months. At this time of year families tend to start thinking about the next school year so I've been in lots of classrooms providing important information to teachers and school staff about the needs of MPS children in schools.

If your child is of school age and you haven't spoken to the Advocacy Team about our school talks please get in touch – they really do make a difference and are a great way to ensure your child's school has only reliable information about MPS.

Educating and providing face-to-face support is at the heart of the Advocacy Support Service and we are always more than happy to pay a visit if you need us to. If you live in Ireland (North or South) and have an unmet support need please do not hesitate to get in touch!

Alison Wilson

Advocacy Support Officer

What to look out for in my next report

Disability Pride (20th September) – I am involved in organising the Northern Ireland Rare Disease Partnership's participation at this event in September.
I can promise lots of carnival style photos in the next magazine!

Your Stories

Bilal Mohammed

Bilal helping out in the community

Bilal Mohammed

I am currently a Police Support Volunteer and I have been working for Greater Manchester Police for 3 years. I play a role in calculating violent crime statistics for Bolton's licensed premises to help target enforcement action. I have produced an 'easy read' hate crime booklet to help vulnerable people in society understand what hate crime is and how to report it, also adapting the use by Mencap.

I have produced a leaflet aimed at educating young people about what to do about domestic violence in the home. I also created a power point presentation for cyber bully crime, which helps my colleagues to bring information to schools and colleges, so that young people know what they can do if they experience something similar.

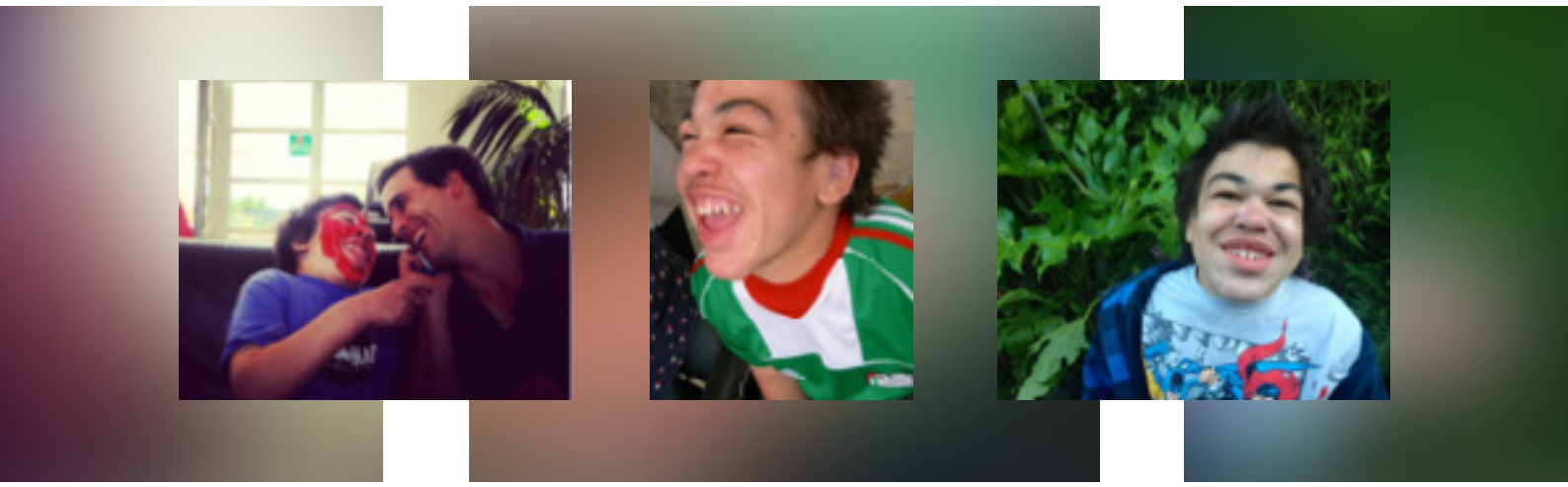
I am determined to give something back to the community by working with the police.

New School Uniform

Clark D'Alton

Clark D'Alton

New school year? New school uniform? For the next edition we would love to see photos, like Clark D'Alton looking super smart in his new uniform. Please send photos to **magazine@mpssociety.org.uk**



Jackson James Whittaker

MPS II — 15/09/1997 - 12/05/2014

Jackson had a smile that would have people stopping in the street — a smile that reflected his openness and playful spirit. He was well known around his neighbourhood of Brixton and anyone who didn't know him, soon met him. He was always striking up conversations with strangers, quickly getting to the bottom of who they lived with, where their mum's were, or whether they had children.

He was born in Melbourne and, as a young child, his three favourite things in the world were superheroes and characters from books, dressing up and his family. At family gatherings he would always note who was there and ask after whoever might be missing. And for the other two, you never quite knew which of Jackson's alter-ego's you might come across, whatever the occasion — he might go to school as Batman, the supermarket as Astroboy or a formal dinner as Aladdin.

At the age of six, Jackson left his Mimi and Grandad and his uncle Josh, who were the centre of his world, and came to London to participate in the Hunter ERT clinical trial. Each week at Great Ormond Street Hospital brought some kind of ordeal, whether that be multiple attempts to insert needles for catheters, regular observations and blood pressure checks or just running riot with the other children on the ward. But it was during these long hours in the hospital when he met his MPS family — some of the wonderful people who have helped and supported us along the way.

Jackson never let Hunters stop him from experiencing the world as fully as he could — he travelled all over Europe and north Africa, kayaked (he loved it), rode horses (not so much); and he made friends wherever he went. He had an abundance of love and affection and even when he had few words, he was still able to ask for a cuddle.

Jackson was nothing if not brave — though he'd regularly deny it with the hope of avoiding a needle. He faced the challenges of his condition, and the increasing invasive measures to combat his illness, with dignity and the most awesome smiles.

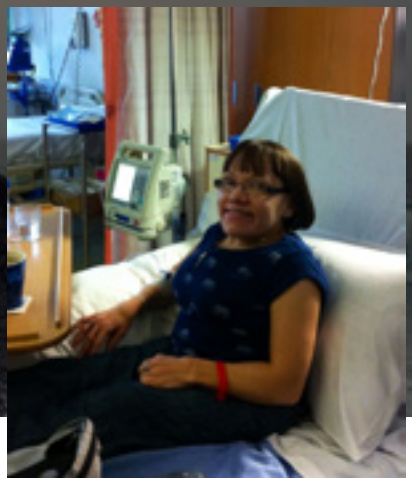
Even during his last days in hospital he shook his shoulders and danced for me — trying to make me happy and make me laugh. On May 12 2014, Jackson slipped out of the body that had been slowly failing him. There is a big community, not just in south London but around the world, who miss Jackson dearly. To his family and friends, he really is a hero.

Xanthe Whittaker

My Experience of the Morquio Enzyme Replacement Therapy Clinical Trial

As you may remember from our Summer Edition the announcement that Elosulfase Alfa (Vimizim) has been given marketing approval by the European Medicines Agency (EMA) as an Enzyme Replacement Therapy for MPS IVA Morquio disease.

Angela Paton has kindly written a piece describing what it was like to be part of the clinical trial and the impact the drug had on her.



I was diagnosed with Morquio Syndrome in the late 1970s at the age of four and grew up in the 1980s and early 1990s when a treatment for Morquio was still a far off hope and not something I expected to see within my lifetime. Despite the physical limitations that come with having Morquio, I have managed to live a relatively normal life.

I attended mainstream schools and 6th form college and was able to go away from home to university. Since leaving university I have always managed to work and although I need assistance with a number of aspects of daily life and running a home, I have been able to live alone and basically independently. As an adult I had accepted my identity as a person with Morquio and with that, the knowledge that as I got older my disease would continue to progress and my health and mobility would continue to deteriorate. Despite this knowledge, I never really gave much thought to the prospect of there being an actual treatment that would address the underlying cause of Morquio.

In 2008 I attended the MPS Society Morquio conference in Northampton where it was announced that enzyme replacement therapy had been developed by Biomarin and that it would soon be ready for human trials. I was in my 30s by this time and so, while I was thrilled that there was now hope of a treatment, I was also very much of the opinion that this would be something that would only benefit young children and future generations. I assumed that for adults like myself, the damage was done so to speak and it would therefore be too late for ERT to have any real effect. I was aware in the following years that clinical trials had begun in children but it was quite a surprise in 2011 when my consultant told me the clinical trial was now going to be extended to include adults and asked if I would be interested in taking part.

Upon reading the consent form I found the prospect of taking part in the clinical trial, at first, rather daunting. It was still an experimental treatment with a number of potential reactions and side effects. I was also very uncertain that I could physically manage the demands of the trial. Living in Leeds and the nearest trial site being Manchester meant I would have to drive myself to Manchester and back every week for infusions. This is a 120 mile round trip, making it a very long day, and in addition to which, I would still have to go to work for the rest of the week. As I was working a five day week at that time, albeit slightly reduced hours, it also meant that I would have the financial burden of further reducing my working hours in order to accommodate the weekly infusions.

It was not an easy decision but my decision to take part in the trial, in the end, came down to two things. Firstly, if I could manage it physically, then I wanted to play a part in the development of a treatment for the benefit of everyone with Morquio, especially for those who, for whatever reason, were not able to take part in the trial themselves but would benefit from a licensed treatment. Secondly, from a health point of view, I felt I had nothing to lose. My stamina, energy and ability to manage everyday tasks had been reducing for some years, but especially so in the last few years. I had recently had to move to a bungalow due to my increasing inability to manage stairs and despite having reduced my working hours a little each day, I was still struggling physically most days. I therefore decided to go ahead and give it a chance. I felt that if the treatment improved the way I felt then that would be fabulous. Even if it just slowed down the progression of the disease then that still would be a bonus and if it had no benefit at all then at least I could move on knowing that I had tried and not left wondering 'what if?'

I had my first infusion in February 2012, aware that for the first 24 weeks of the trial I may not be getting the treatment at all but rather just a placebo. My employer was incredibly accommodating, reducing my working hours, allowing me to work a four day week in order that I could have my infusions and giving me paid time off for my assessments. I was incredibly nervous for the first infusion, however, everything went fine. I had no problems or initial reactions to the infusion and so began the weekly trips to Manchester. I was very surprised at how quickly I adapted to making the trip to Manchester every week. It is surprising how something that at first seems so daunting can so quickly become routine.

For the first 7 or 8 weeks of the trial I felt no different. Naturally, I was constantly wondering and speculating as to whether I was on treatment or placebo but as I had felt no change and had no initial reaction to the infusions I was swaying towards placebo. Then, after around 7 or 8 weeks of infusions, I began to realise that I was feeling physically better. Prior to starting the trial I spent a great deal of my time feeling stiff, sluggish and generally very tired. Everyday tasks such as getting showered and ready for work in the morning would often leave me feeling worn out and I was struggling to maintain a balance between being able to work and still having enough energy for a normal social life. Even though I

was working slightly reduced hours, I rarely had the energy to do anything in the evenings, going to work was as much as I could manage and most weekends I would need to spend at least one full day just resting at home to recover from the working week.

Approximately 7 or 8 weeks after the infusions had started this began to change. The first thing I noticed was that I wasn't as tired after getting ready for work in the morning or feeling exhausted at the end of the day when I came home from work. As my energy and stamina increased I found that I was able to do more. I was able to do simple things like go out for a meal or to the cinema after work where before I would often have declined these invitations, being too tired after a day at work. I also found I was able to do more at the weekend, no longer needing to rest as much. Being at work became much easier and more enjoyable too as I began to feel less tired, stiff and sluggish. While these may seem like small improvements, just being able to get on with life and enjoy my leisure time as everyone else does without constantly feeling exhausted has had a very big impact on my quality of life.

The enzyme is not a cure for Morquio and cannot undo the bone damage that has occurred. I still have a lot of physical limitations and need assistance with many aspects of everyday life. I still have to live with a lot of joint pain and severely restricted mobility but I have found that the pain is a lot easier to deal with when it is not accompanied with the tiredness and stiffness that it previously was. Joint pain that at one time would have resulted in me taking time off work or cancelling arrangements, I now find I am able to deal with and still manage to get on with life.

There will always be a part of me that questions whether, in those first 24 weeks of the trial, I was receiving the treatment and not a placebo and that I did genuinely feel the benefit of the treatment and not just the placebo effect. Having studied it during my psychology degree, I am very aware of just how strong the placebo effect can be. These doubts are minimised however when I speak to other adults who have been on the trial or read stories on the internet and on social media about the positive impact that this treatment has had on the lives of so many others who have taken part in the trial.

There has also been another benefit from being on the clinical trial that I did not expect. I have become friends with a number of other adults with Morquio who are on the trial with me in Manchester and others at sites around the country and I cannot overestimate the value of these friendships. Having the opportunity to chat and socialise with others who genuinely understand the struggles and challenges that come with living with Morquio is invaluable and I don't doubt that these friendships will continue long after the trial has ended.

> *"Two and a half years on I can undoubtedly say that my quality of life as improved as a result of being on this clinical trial and receiving this treatment."*

Two and a half years on I can undoubtedly say that my quality of life has improved as a result of being on this clinical trial and receiving this treatment. Overall, my working week is now much longer than it was prior to starting the trial. I now work and extra hour per day on the 4 days that I do work in addition to leaving home at 6.30 am every Wednesday for a full day round trip to Manchester for my infusion. Yet despite this I still have so much more energy than I ever had previously and manage to take part in many more social activities.

The treatment, Vimizim, was licensed in Europe in April and the trial is now coming to an end. Due to the changes that occurred within the NHS last year, the end of the trial has been far from ideal. Biomarin will continue to fund the treatment in a hospital setting, for the time being, for those of us who were on the clinical trial. There is, however, a distinct lack of information for those of us still receiving infusions at the research facilities as to where we will receive our treatment, how far from home we will be or when the transfer is likely to take place. There is also now the prospect of months of uncertainty as we wait to find out whether this treatment will be funded by the NHS so that everyone with Morquio can benefit from it, irrespective of whether or not they were lucky enough to be able to take part in the clinical trial. For those of us who are lucky enough to be receiving this treatment, the thought that it could be taken away from us if the NHS does not deem it to be worth the cost is devastating. Despite all of this uncertainty, however, I am still incredibly grateful that I was given the opportunity to take part in this clinical trial and in the two and a half years I have been on the trial there has never been a single occasion when I have regretted my decision to take part in it.

Finally, I could not write about my experience on this trial without mentioning and saying thank you to the wonderful and dedicated medical staff who have cared for me at the Manchester Clinical Research Facility. Not only the doctors, nurses, physiotherapists etc who are directly involved with the trial but also those nurses, doctors and other staff on the ward who care for me week in and week out during my infusion visits. It is in no small part due to the kindness, warmth, compassion and humour of all of these people that I have managed to cope so well with both the physical and emotional demands of this trial and to all of them I will always be grateful.

Angela Paton

Thank you Angela for taking the time to write such a fantastic article.

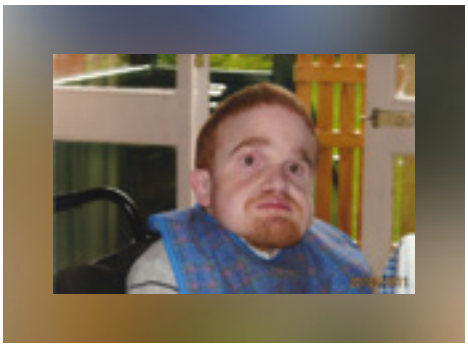
As Angela mentioned, funding by the Health Department for all four countries in the United Kingdom for Vimizim is not yet in place and the drug is currently being provided compassionately with BioMarin continuing to fund the cost.

BioMarin are continuing to keep the MPS Society up to date with developments regarding future funding and patient access to Vimizim.

We are committed to do whatever we can to bring about a successful outcome but we need to raise awareness of the clinical trial and the benefits seen to secure reimbursement.

If you are interested in helping raise awareness by writing to your MP please get in touch and we will assist you with this.

We are hopeful that by the next edition we will have some good news to report.



In Memory of My Precious Son

Richard Lewis Turner

22nd September 1981 – 7th June 2014

Richard was born on 22nd September 1981, 1:40am at St Mary's Hospital, Manchester. My daughter Claire was nine years old when Richard was born. From the very first moment Richard was placed into my arms, my gut instinct was not a good one. I was later proved correct.

Richard's first year was continuously spent in and out of our local paediatric hospital. He was a very sickly baby, had hydrocephalus, kyphosis (spinal curvature), clawed hands, and there appeared to be none of the obvious progress and development, which I had previously experienced with Claire.

One day totally out of the blue, mid Dec 1982, I received a phone call from Pendlebury Children's Hospital. It was a Dr Sardawalla, he requested to see both myself and Richard's father, together with our baby, on that very same day. We duly arrived at the unit and were met by Dr Ed Wraith (as he was then). Between the two doctors, they delivered the devastating news which every parent dreads, that our son had an inborn genetic disease. "Hunters Syndrome" MPS Type II. We were admitted immediately for five days of extensive diagnostic tests. The diagnosis was confirmed. It was explained to us that the only course of treatment was a Bone Marrow Transplant. At that time the only hospital to undertake this treatment was the Westminster Children's Hospital in London.

Firstly a suitable donor was needed; my total family was small, the chances seemed slim. However, Claire, my daughter emerged as a perfect match. Because Richard had presented with "Hunters" almost at birth and quite severely, the doctors were unclear as to whether it would be viable, also the success of transplanted "Hunters" had not been totally proven. The fact that we did have a perfect match encouraged things to go ahead. In April 1983, aged 20 months; Richard and I were admitted to Nathan Ward, Westminster Children's Hospital. The Bone Marrow Transplant was planned for the 7th May 1983. Claire was on another ward, donating while my baby waited for this life saving opportunity and gift from his sister. Richard, was reversed Barrier nursed, and sailed through the whole treatment, no temperature, totally unremarkable.

In July we returned home, Richard was under house arrest, to avoid infections, plus return visits to Westminster for multiple check-ups. Richard required physiotherapy at home, to assist him to learn to walk. He was, and remained doubly incontinent, still no speech, hearing loss, and limited understanding. We also had to take Richard into the local children's hospital for hernia repairs, and grommets for glue ear. Richard began to attend special needs pre-school. Progress and development was still not really apparent, Claire and myself devised a basic signing system to enable Richard to understand us.

When Richard was six years old, our family dynamic changed with the breakdown of my marriage. We were now a single parent family. Richard received some respite care, which helped things along. The ensuing years rolled by at pretty much the same level. Richard had been a boisterous, hyperactive little boy, very noisy, much throwing of things, not engaging very well in interactive play.

With the maturity of years, late teens, Richard was changing into a calmer young man. Nicely mobile, good appetite, very sociable and enjoyed being around people; his health in general was quite robust. At the age of 20 it was necessary for Richard to be placed in residential care, as it was becoming difficult to care for him single-handedly. We were very blessed with the care home where Richard was loved, and treated with care, respect and great dignity.

Richard attended a day centre two days per week. The staff really had their work cut out, as he would wonder off, or go meddling with things in the office. I was told on one occasion, he unknowingly shut down a lap top belonging to one of the staff, and it wasn't clear whether any data was lost. Richard was a cheeky chappie.

At age 26, myself and the staff at the Care Home noticed that Richard's mobility was slowing down considerably. He began to limp, and had increasing difficulty in lowering himself into a chair, and the same difficulty rising from the chair. After some time struggling in this way Richard gave up attempting to do either. X-rays carried out, revealed, progressive, chronic osteoarthritis in both hips and both knees. Richard had now become wheelchair bound. I was extremely saddened regarding this, as this had been his only form of independence choosing where he wanted to be, and enjoyed walking along the corridors, and in and out of rooms. Richard adapted to this great change with such patience, humility and tolerance, there was no fuss; he was such a shining example.

Richard was now under the care of a consultant at Salford Royal Hospital, Unit for Adult Inherited Genetic Diseases, via Professor Wraith. Extensive MRI and CT Scans under general anaesthetic were carried out to reveal that both Richard's hips had totally disintegrated, also both of his knees had by now contracted to quite acute un-natural angles. Extensive pain relief was given on a daily basis. Richard's dentistry was also becoming more and more problematic, and many extractions were required.

Last November (2013), on one of my many visits to Richard, I noticed that he was not looking at me, nor was he able to focus, both eyes were affected. The Care Manager and I took Richard immediately to Manchester Royal Eye Hospital. It was confirmed that both retinas had become detached. Richard was blind. The Eye Hospital rather rapidly made the necessary arrangements, and on the 13th December 2013 Richard underwent surgery on both eyes to attach the retinas. This was quite lengthy surgery; as a result Richard was really very poorly for some weeks. I feel that it had been too much for him. Hospital check-ups were out of the question, as he was too ill. It was also becoming increasingly apparent to both myself and all the staff, that this surgery had not been successful and Richard still could not see.

It was now necessary for Richard to be spoon-fed, both food and fluids, as he was unable to see things in front of him. He was also completely bedbound, as his bespoke armchair was no longer comfortable for his increasingly painful body. Things carried on in this manner, Richard receiving "one to one" care, all his needs very lovingly attended to.

The first week of June 2014, I had called as usual to visit Richard, to be told that he had been unwell that day, the GP was called out and multiple antibiotics were being given, as the GP suspected a lung infection. The GP was calling in to check on Richard everyday, however, on 6th June 2014, I received a phone call from the care manager, (I was at work, Manchester Royal Infirmary) to notify me that the GP was requiring an ambulance urgently to take Richard to Manchester Royal Infirmary as he was concerned with Richard's breathing.

The care manager and I were in "RESUS" in the A&E department with Richard from 3:40pm until 11:30pm. The staff were very caring and looked after Richard so well. At 11:30pm we were transferred to High Dependency Unit. Yet again excellent care was given, every attention given to Richard's condition. However, on Saturday 7th June 2014 at 1:40am very suddenly and sadly my lovely boy died peacefully.

My "Precious Son" leaves a gaping space in the lives of both his sister Claire and myself. We were both highly privileged to have been sister and mum to this amazing young man. Although somewhat painful writing about Richard's journey, as our grief is still quite raw, it has been quite cathartic to share it with you all.

God Bless you all

Val Turner (Manchester)

Childhood Wood

21 years of childhood wood

A warm, sunny day greeted us when we met with a number of families to commemorate 21 years of Childhood Wood. This was my first visit to the Childhood Wood and I was amazed at the size of the wood and the height of the oldest trees. It is a beautiful, calm oasis and a fitting tribute to those lost to MPS diseases.

The day began with a short memorial service led by Christine Lavery and Sue Peach and following this the families spent time finding their trees and commenting on how much they had grown. We then went to a meadow area near the pond and enjoyed a delicious picnic. It was lovely to hear the families share stories and catch up with each other's news.

The fun events took place after lunch with a choice of an exhilarating Segway ride or exciting high-level climbing completing the Go-Ape treetop adventure within the Sherwood Pines. Everyone appeared to have a great time with lots of giggles and excited chatter afterwards – a good day had by all!

Debbie Cavell

Advocacy Support Officer

MPS Family Fun Day to Flamingo Land

The gathering at Flamingo Land occurred on a remarkably sunny and warm day despite it being relatively early in the year. As the representative of the MPS Society I met everyone at the gates and distributed the tickets to the excited families who were desperate to beat the queues and trial some of the rides. Elliot and Oliver helped bring people over to the car and dish out the arrangements (so it has been an ongoing dispute as to who actually left the car boot open for the entire day, an error that will have relevance later in the report!).

As everyone raced into the theme park, we asked if they could all try to meet in the food area at lunchtime for a group photo. Most were far too excited to hear this and as you can see from the picture, not many returned as they were either far too involved or in queues for the most popular rides. Everyone later apologised but we were delighted that they were enjoying themselves so much!

One downside of the sunshine and heat was that we felt obliged to try the famous 'Log Flume' although we all completely under estimated the amount of water we would be subjected to - indeed someone remarked that we would not be so wet if we had actually jumped in and had a swim!!

As is always the case, the day passed far too quickly and as the tired families left the park they all asked us to pass on their thanks for a wonderful day - everyone left with a smile on their face!

When the last family had left we returned to our car, which was by now standing alone in the car park. It soon became clear that it may remain there for some time as we discovered the open boot (which nobody was responsible for) and a completely flat battery.

However, as we stood accusing each other and trying to find the number of a local garage, a large tractor unit from the Flamingo Land garage pulled up and a very kind mechanic hopped out with jump leads in hand saying he had seen us struggling. Within 10 minutes we were happily on our way and laughing about a really successful and enjoyable day.

Thanks from everyone to the Sir George Martin Charitable Trust for enabling such a wonderful day for our affected families!

Paul Moody

Trustee

Chessington Family Weekend

"Dragon's Fury Awesome!"

This August the MPS Society organised a fun Family Weekend to Chessington World of Adventures, where the families could enjoy the thrill of some of the country's most exciting rides, as well as taking in the wildlife at Chessington Zoo and the Sealife Centre. Whether they were brave enough to tackle 'Rameses Revenge' or preferred to go and watch the inhabitants at 'Penguin's Cove', there was something for everyone.

Advocacy Officer, Steve Cotterell, went along to help, and here is how he got on:

We arrived on Friday afternoon after battling the M25 traffic - all a bit frustrating but we soon got into the spirit of things and the usual MPS hotel takeover began: furniture was rearranged, TV channels switched and rooms swapped. Once everyone had arrived we gathered for dinner where all the children were delighted to receive chocolate and a present, all kindly donated by the Mix 96 campaign. After dinner the children enjoyed entertainment from Auntie Jo Jo with magic, comedy and superb balloon modelling, featuring Ariel the Little Mermaid, Iron Man, rabbits, penguins and more. She did a remarkable job of keeping the attention of the children for over two hours, enabling parents to mingle, chat and meet other people.

On Saturday morning we all set off for Chessington World of Adventures. Fortunately the sun was shining, and though the park was very busy, families were able to use ride access passes to allow them to get to the front of the queues. I know that the Carousel proved to be popular with the younger children whilst others enjoyed the thrill of rides such as Dragon's Fury and The Vampire Ride. Many of the children were excited to learn that the Octonauts were visiting the park with opportunities to meet and be photographed with Quasi and Captain Barnacles before continuing to "Explore, Rescue, Protect!" through the Sea Life Centre.

We asked some of the families to sum up their Chessington experience in 3 words - and here is what they came up with:

Fun, Enjoyable, Friendship

Connect, Family, Support

Fun, Friends, Talk

Excitement, Smiling, Lively

Children Having Fun

Togetherness, Friends, Fun

Dragon's Fury Awesome!

New Friends Made

Balloons, Dancing, Magic

After another comfortable night in the hotel, Sunday gave another opportunity to explore the park only with the addition of a downpour. At the end of the day, exhausted, we all headed home.

Steve Cotterell

Advocacy Support Officer

Thank you to everyone who attended who made it such a fantastic weekend!

"Please pass on our thanks to everyone at the MPS Society involved in organising last weekend's trip to Chessington World of Adventure. We all had a fantastic weekend and really enjoyed ourselves. It was also nice to catch up with some of the other families that we have met on previous outings and to meet some others for the first time. Everybody was so friendly and it was good to be in such an understanding environment. We really appreciate the efforts of the MPS Society in making these trips happen and look forward to when we can do it again."

– Chris and Hannah Brentnall

Cadbury World Family Day

Early in August we thoroughly indulged our sweet-tooth by holding a Family Day at Cadbury World in Birmingham, all of which was made possible by the kind support of a grant from the Michael Marsh Charitable Trust.

The families who attended were able to travel back in time to see how chocolate was first consumed by the ancient Aztecs, see an atmospheric replica of John Cadbury's first shop, play state-of-the-art interactive games and rides, and generally get to grips with all aspects of chocolate production.

The day proved to be a great hit, with the children loving the games and the ride in the Cadbury 'beanmobiles', and both adults and children alike feeling rather peckish after viewing vats of liquid chocolate in the 'Making Chocolate' section!

Thanks to the Michael Marsh Charitable Trust, and all those who attended, we were able to provide a fun and interesting day for families affected by MPS and related diseases.

"Thank you for putting on such a wonderful day out at Cadbury World. Paige enjoyed herself so much, and it was nice to meet up with other families that we would not have probably met."
– Catherine Cullen

Fundraising

Welcome to the fundraising section of our Autumn 2014 MPS Magazine – a place where you will find a plethora of fundraising features, from tales of endurance and adventure, to stories of resourcefulness and fun.

Our supporters never cease to surprise us with the feats they are capable of, but one of the things I have noticed from looking at the stories featured here, is that the greatest source of inspiration for these events comes from the children and adults affected by MPS and related diseases; their daily struggles, twinned with their often vivacious spirit, serve to spur on fundraisers when they have pushed their bodies to the limits with marathons, obstacle courses and cycle rides, or when they are stuck for creative inspiration.

In turn, the following stories from our supporters are themselves inspirational.

Along with our supporters' stories, we have also included a section about one of our most popular seasonal fundraisers, **Wear it Wicked,** along with an outline of a variety of ways you can fundraise with minimal effort!

A huge thank you to everyone who fundraised for us and who is planning to do so in the future: your efforts, from the biggest event to the smallest, really do make a difference and allow us to continue our work in support, research and awareness.

Our apologies if we have not included your fundraising story, but this section is intended as a snap-shot only – but we do love to hear what our supporters are doing, so please remember to tell us all about your event by dropping us an email at fundraising@mpssociety.org.uk.

Elkie Riches

Fundraising and Communications Officer

Marina & Friends: Star Fundraisers for Sanfilippo

Over the years 'Marina and Friends' has become well-known in the local Bristol community as the place where you can get almost anything – an Aladdin's Cave at bargain prices. This small shop relies solely on goods donated by the general public, and is staffed by volunteers who sort and sell, and provide a warm welcome to anyone who might come in for a browse.

To date, this tiny second-hand shop has raised a truly incredible £124,654.71 towards research into Sanfilippo!

Opened in 2003, this amazing fundraising shop is the result of Marina Foster's hard work and dedication. Marina started her fundraising back in 1996 following the diagnosis of her beloved twin grand-daughters, Francesca and Josephine, with Sanfilippo disease. Starting with highly successful car boots, Marina soon took the opportunity to take on a small independent shop.

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

Thank you so much to Marina for her exemplary work in the field of fundraising, and thank you to her team of volunteers and customers who show such wonderful support. 'Marina and Friends' have a right to be truly proud of their achievements.

Heathlands Primary Academy's MPS Fundraising Day

Three of our pupils, through their own initiative, worked hard to raise money for the MPS Society. As well as having a non-uniform day, the children, with a little assistance from their friends, held a sale of bookmarks, pencils, pens, key rings, as well as selling toys, milk shakes, cakes and Asian food.

Mrs S Lambeth-Angell, Headteacher

Many thanks to Aftab Hussain and all the children who worked so hard, and also to Heathlands Primary Academy for their support. Well done on raising an incredible £670!

Fundraising Reminder

When paying in your donation as a bank transfer, please remember to use your full name as a reference so we can link it to a fundraising event and pass on our gratitude.

What Inspires You?

If you read some of the articles in this section, you will discover what, or rather who, was the source of inspiration for our supporters in their fundraising.

So, who or what inspires you to continue to support us? Who or what is your muse in your everyday life and how have they changed the way you think?

We would love to hear about your inspiration, so please write in and let us know by emailing e.riches@mpssociety.org.uk. I will feature people's thoughts on this in a future magazine – feel free to include a photo too!

Socks, cakes and swishing!

We are constantly humbled by the goodwill and generosity of those around us. **MPS Awareness Day** was no exception this year. Our eldest son Sam has Morquio Syndrome, and we decided that we wanted to find some creative ways to raise money – and have some fun too!

So, at Sam's school they held a "Wild and Wacky Odd Socks Day". The children could wear wild and wacky odd socks and brought in spare change to fill a giant sock! They raised £150.

At home I organised a soiree for the girls. We had swishing (swapping unwanted clothes for a small donation), delicious cupcakes, jewellery and cards for sale, and a raffle. All were provided by friends with their own small businesses – all donating prizes or part of their takings to the MPS Society. One friend has even been e-baying the clothes that were left after the event, to squeeze out every last pound! The odd glass of wine was drunk too.

At work (Asda's Home Office) my colleagues and I spent the weekend baking and treated the office to a fabulous cake sale! We raised £170, and Asda generously topped this up with an extra £500!

Family got involved too, supporting the soiree, selling raffle tickets, and my mum held her own coffee morning and a raffle too. Her neighbour also asked for support from her employer, Next, and they donated £200!

Thea's Trust, the charity set up by the family of Thea Paterson who suffered from MPS I (Hurlers) and sadly died earlier this year, donated a further £390 to our funding efforts. A wonderful gesture!

And lastly, a friend of a friend decided to support the MPS Society after hearing Sam's story by running the Great Manchester Run. She raised £200.

So in total, we collectively raised £2,500. We are really grateful to all our friends and family for their support. The question now – how to top it next year!!

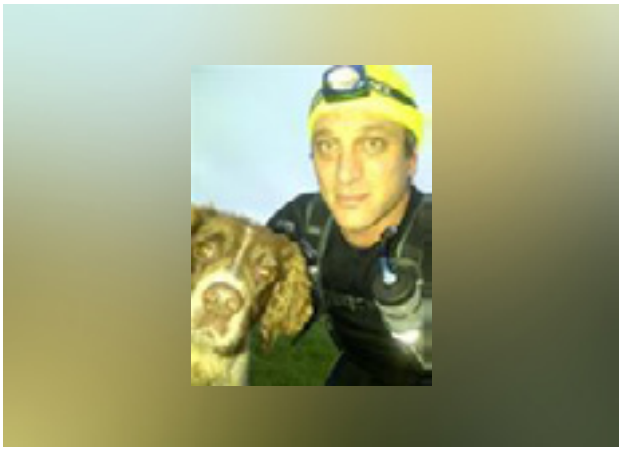
Katy Brown

Thank you to Katy, Asda, Next, Thea's Trust and all those who got involved in the Brown family's fundraising.

Thank You Ann Parsons

The MPS Society would like to acknowledge the fundraising efforts of Ann Parsons, who has been selling our MPS trolley key-rings at ASDA in Eastbourne for the past eight years! In this time it has become a regular occurrence in our office for Ann to ring up to donate the proceeds of her sale – she was seemingly able to sell a constant stream of our key-rings! On totting up all her donations over the years, we have made her grand total to be an astounding £4,003.72!
Sadly due to people helping themselves to the key-rings, Ann has decided to call her fundraising a day, but we remain very appreciative of all her efforts.

Thank you, Ann.



Marathon des Sables

If you do happen to hanker for an even bigger challenge than a gruelling obstacle course (which you can read more about further on!), you could always try entering the Marathon des Sables – ranked by the Discovery Channel as the 'toughest footrace on Earth'. This ultramarathon requires you to carry all the equipment you will need for survival on your back as you run the equivalent of five and a half marathon in 5-6 days – and all within the inhospitable locale of the Sahara Desert. Not for the fainthearted...

Charlie Wise was certainly not faint of heart, however, when he undertook and completed the Marathon des Sables for the MPS Society and raised £2,283.08 on his JustGiving page. An incredible feat of endurance, Charlie – thank you for your excellent support.

If you are interested in taking on this challenge of a lifetime, please visit the Marathon des Sables website; www.marathondessables.co.uk

Charity Spring Fete

On Sunday 18th May, Rashpal and Sandra Singh held a Charity Spring Fete, which boasted such attractions as Shaolin Warrior Monks (pictured), donkey rides, face painting, and a football tournament. Jane and Sarah from the Willink Unit at Manchester Hospital, kindly came down to run an MPS stall along with Sarah's daughter. The event was held in memory of their son, Daniel, who passed away from MPS II in December 2011.

Sandra and Rashpal recently wrote to us to report that "We had a wonderful day, with beautiful weather, and with the help of friends and family we made it a day that Daniel would have loved."

Thank you to Rashpal, Sandra, Jane, Sarah, and all involved for holding such an amazing event and raising £700 for the MPS Society. The couple also made donations to the Stars Playgroup for disabled children and Baginton Fields Special Needs School.

Towersey Morris Men

Many thanks to the Towersey Morris Men who kindly donated £600 to the MPS Society. Long-term supporters of the MPS Society, the Towersey Morris Men perform traditional English Morris dances in Oxfordshire and the surrounding area.

Thank you also to the Evans family for picking up the donation. We heard you all enjoyed the Towersey Morris Men's performance – apart from Harry Evans (MPS I) who got slightly alarmed by the loud noises!

Everyday Fundraising

It seems that we are all pushed for time nowadays, especially if you are caring for a child or vulnerable adult. So, while you may want to show your support and contribute to the MPS Society's work, you might simply not have got the time.

The good news is that there are any number of fundraising schemes that are designed to take up little of your time and can sometimes fit quite easily into your everyday life. We have outlined some of the most popular ways to engage with easy, everyday fundraising:

Easyfundraising

This fantastic shopping fundraiser allows you to raise money for free, while you shop at any of the biggest online retailers, such as Amazon, Tesco, M&S, Aviva and Thomas Cook – in fact, easyfundraising are supported by 2,700 top retailers! Just register at http://www.easyfundraising.org.uk and collect donations from these companies every time you make a purchase through Easyfundraising, with no extra cost to you. Similar schemes include Give As You Live (http://www.giveasyoulive.com/) and The Giving Machine (http://www.thegivingmachine.co.uk/).

The Weather Lottery

You can enter the Weather Lottery and support the Society for Mucopolysaccharide Diseases for as little as £1 a week. Just visit the website (http://www.theweatherlottery.com/), choose your cause and join, and you could be in for a chance of winning the £25,000 weekly jackpot! Players must be 16 or over.

Recycle

We all know the importance of recycling, but next time you wonder what to do with your old mobiles or ink cartridges, think of us! We have teamed up with Recycle4Charity to recycle these objects, while receiving 50% of the recycling value. Please email us at fundraising@mpssociety.org.uk for a freepost envelope for mobiles and ink cartridges. We also recycle used stamps!

Charity Flowers

If you are thinking of sending flowers to someone, try visiting www.charityflowers.co.uk. All of their bouquets are packed in a presentation box together with a cut flower care leaflet and your own personalised message. The flowers are sent by first class post. Order your flowers by phoning 08705 300 600 and quote 'MPS' at the time of ordering so that 15% of the retail price of each order is donated to the Society.

Thank you to all our donors including...

Barclays Bank raised another £22.33, in addition to the previous £280.69, which they raised by holding a Wear It Blue event.

Super-fundraiser, **Hannah Brentnall**, has donated a further £1115.69 which she raised over MPS Awareness Day.

Yewstock School Wore It Blue for MPS Awareness and raised £101 for the MPS Society

We have received £60 matched funding from **Barclays**

Ann Parsons sold MPS trolley key rings at ASDA, Eastbourne and raised £86.

Natasha Fairweather donated £250 as part of the money she has raised from completing the London West Tough Mudder. She is still collecting further funds on her Virgin Money Giving page.

Padraic O'Connor completed the Hyde Park Triathlon and donated £30, which he received as sponsorship. Padraic has raised further funds on his Virgin Money Giving page, which amount to almost £2,000!

Morgan O'Hara completed the Manchester 10k, while Lily, Jasmine and Olivia O'Hara did a sponsored 15 mile walk along the Wirral coastal pathway. In total the family raised £362.50, plus £336.22, which they collected on their JustGiving page.

Beauchamp College Wore it Blue on MPS Awareness Day and held a dress-down Friday and raised a fantastic £1,084.01.

Sue Jenkins sold refreshments and cream teas at her local community market and raised £103 for the MPS Society

Sarah Robson from RBS, Sunderland sent in a further £50 toward the Genistein Trial, which was kindly donated by a member of public.

The Lloyds Bank Private Clients Team in Manchester held a Wimbledon-themed 'Strawberries, cream and shortbread' event and raised £96.

Bridgnorth Women's' Rugby Club supported the MPS Society on their float at the local carnival and managed to collect £150.46

Kath Hiller sent in a further £120.41, which was collected as sponsorship from her Wicked Walkabout.

Marina's secondhand shop in Bristol raised another £9773.70 toward Sanfilippo research.

David Gosling celebrated his 50th birthday and in lieu of a present from Mr & Mrs Hobson of the Hobson Foundation, he received £5000 as a donation for the MPS Society

Pupils at St James CE Primary School Wore It Blue to support MPS Awareness Day and raised £205.20

The Year 5 pupils of Fernwood Junior School Wore It Blue for a day and raised £100. A brother of a student suffers from a MPS disease.

Antony Williams ran the Greater Manchester Marathon and raised £592.50 in sponsorship

Annie Anstey braved the Maltese sun and completed the Malta Marathon and made £1436.50 for the MPS Society

Helen Sharples of Savvy Financial Planning completed a 10k Run for Ella, raising £67.50

James Hope-Gill ran the Virgin London Marathon (featured in Summer 2014 magazine) and raised an amazing £3240.65

Kerry Fricker competed in the breath-taking Electric Run and raised £97.50

Kirsty White completed the infamous Tough Mudder London West, raising £900

Simone Piromalli ran the Virgin London Marathon for the MPS Society and raised £1985.58

Stuart Ellison ran the Greater Manchester Marathon and received £180 in sponsorship

Rachel Tobin of Savvy Financial Planning ran 10k on the local trailway and raised £627.50

Richard Sleath ran the iconic Virgin London Marathon, raising £296.25

Sumeet Garara, Chandra Varsani, Bhavin Garara and Kaajal Raghvani completed the 'Nightrider', a 100km moonlit ride around London's landmarks and raised over £1,000 for the MPS Society

Claire Kaye ran the Brighton half marathon and raised over £300

Donations

James Roberts, The Blackburn Trust; Next plc; Mrs A. Baker; Springwell School; 5 Ruxley Court; Pharmaco Foundation Ltd; Oliver Drewery; Susan Hollidge; F.A. Sandow; Mr & Mrs Arrowsmith; Justin Berry; Jean Mossman; Carol Copsey; Mr & Mrs Lavelle; Mr & Mrs Eaton; Mrs E. Mason; Keith & Shirley Bown; Joan Crespin (for MPS III research); Old Trafford Christadelphian Church Sunday School; Knitt & Natter St Mellons Group; Ray & Aileen Webber (for MPS IV); Mr & Mrs John Keenleyside (for MPS IV); S A Swayne; Alan & Ann Barker

In Memory

Clifford Murray; Mr Alec Evans; Richard Lewis Turner; Miss Elsie Winifred Nowell; Gethin Robins; Mrs Sheila Robinson; Mr William Stanley Belding; Gracie Bella Sims; Alec Evans; Mrs Ingrid Lewis

Collection Boxes

Stamps, foreign coins, mobile phones, ink cartridges, jewellery

Your Derwent & Solway; Damien & Dinah Adair; Keith & Shirely Bown; The Croft family;

The Society would like to thank the following donors for their regular contributions by either Standing Order or Give As You Earn

R & K Dunn; S Littledyke; N Saville; M Tosland; M Barralet; L Twaddle; S Bhachu; C Cullen; L Brown; I & A Hedgecock; D Forbes; P Shrimpton; G Simpson; A Sullivan; A Byrne; D Robinson; N Cadman; J York; J Wilson; J Wood; A Tresidder; Mr Thompson; E Cox; K Robinson; K Osborne; M Rigby; M Peach; C Garthwaite; R Arnold; John Scott; C Hume; E White; A Weston; I & V Pearson; J Ellis; R Arnold; C Garthwaite; M Peach; W Cavanagh; A Sabin; B Harriss; L Brodie; A Ephraim; J Dalligan; M Malcolm; E Mee; Mr Hahner; K Brown; E M Brock; J Casey; J & V Hastings; E M Lee; C & M Gibbs; D & S Peach; S Bhachu; C Cullen; S Brown; I & A Hedgecock; D Forbes; P Summerton; R & N Gregory; K & S Bown; S & J Home; V Little; S & D Greening; Z Gul; M Reeves; E Moody; M Fullalove; G Ferrier; E Parkinson; M Leask; R Taylor;

Claire Dew raised £198 in sponsorship by running the BUPA Great Manchester Run

Jemma Thomson ran the Liverpool Spring 10k and raised £380

Megan Hooper ran the Watford 10k for her big brother, Jamie (MPS III), and raised £100

Kate Farran ran the BUPA Great Manchester Run for her neice, Hannah, who suffers with MPS VI, and managed to raise £458

Linda Bell Wore It Blue and fundraised, raising £385 on her justgiving page.

Stuart Ellison ran the Manchester Marathon and raised £155 in sponsorship

Susan Payne bravely had the MPS Society's logo tattooed on her ankle and raised £800! Well done, Susan!

Barry Quant ran the Torbay half marathon and has so far raised £340

Pauline Pike took part in Kath Hiller's Bridport Wicked Walkabout and raised £132

Fiona Curtin was inspired by the Brown family to complete the Great North Swim. So far Fiona has raised £60 on her JustGiving page

Ned Stringer, whose daughter suffers with MPS I, completed a Manchester to Cumbria cycle and raised an amazing £627

Kelly & Dale from **Storm Hair Design** completed the Leeds Total Warrior event on behalf of Lee-Anne Tosland and her daughter who suffers from Fabry. So far the pair have raised £20 Lee-Anne's JustGiving page

Friend to the Hillers, Madeline Warren hosted a cream tea event, complete with a raffle and cake stall, to raise money for MPS. Madeline served 60 cream teas to friends and family who braved the changeable weather to attend the event held in Madeline's garden in Charmouth, Dorset, raising a fantastic £502.65.

Iris Hitter held an open house event where she sold cakes and held a raffle and a quiz. Iris wanted to remember her daughter, Jacqueline, who sadly passed away from MPS III, and also she wanted to raise awareness for the disease. In total Iris raised £206.34

AFC Bridgnorth Football Club raised £205 during their Little Spartans Creche fundraising morning, which included a football session and a party with "Disco Daddy"! Thanks to Emma Hiller for passing this on.

Courtlands Lodge nursing home raised £308.01 from their summer fayre. The housekeeper at Courtlands has a granddaughter who suffers from Hurler Disease.

Polly Anna's Day Nursery held a fundraising event in support of Blake Knaggs (ML II) and raised £100

Susan Lowry walked the Cotswolds Way in memory of her daughter, Sarah (MPS VI), and raised £100.

Jacob Singleton sent in a further £35 for his Coast to Coast event (featured in Summer 2014 magazine). This was donated from staff at Harrogate District Hospital.

Fundraising

Raising Awareness on the Isle of Wight

Family of Avah Flynn, who suffers with Hurler Disease (MPS I), took advantage of the carnival season to raise awareness for MPS. Avah's great-grandmother, Sue, passed on her summer holiday to spend her money designing and decorating a beautiful 'Frozen' themed float, which has been taken to carnivals across the island, each time winning an award. Avah's family donned their MPS t-shirts and did a fantastic job raising awareness at each event.

Thank you to Sue, Gemma and the rest of Avah's supporters for doing such a great job of raising awareness for the MPS Society. We're already looking forward to hearing about your next fundraiser!

Jacks Day at Keenans Estate Agents

Elizabeth Heath and the staff at Keenans Estate Agents in Lancashire held a fabulous cake sale throughout their six stores – everything from mouth-watering Victoria sponges to tempting cupcakes were on offer alongside the usual houses, flats and apartments.

Headed by Elizabeth in memory of her son Jack, who lost his brave fight to MPS II last year aged just 13 years, Jack Day was a resounding success thanks to the dedication of Elizabeth and the team at Keenans, and was even featured in the Lancashire Telegraph.

The cake sale bought in a wonderful £550, which the big-hearted estate agents then doubled to make £1,100! Thank you to Elizabeth, Keenans and all involved.

Fun at the Cambridge Arbury Carnival

Once again it was time for the Cambridge Arbury Carnival. This year's theme was musicals - Zack who has Hunters is mad on High School Musical, so that was us sorted (he had to be Troy Bolton, number 14 for those who do not know). He is particular so surfing the net for the right colour, suitable size, name and number began. Two weeks to go and we were in need of prizes for the tombola, "Don't Panic" Amanda (Zack's mum) and I made little notes and put them through our neighbours' doors up and down our street, asking for donations to raise funds for the MPS Society. The response was fantastic with over 50 prizes coming in.

On the day of the carnival, cars were loaded and we headed off. A little drizzle as we were setting up stall was soon followed by sunshine, which lasted for the rest of the day. We were positioned more centrally this year which gave us perfect views of the 15 to 20 floats as they went passed. Zack arrived and went off with Nana to spend his money on the fair rides. He enjoyed the go karts and wet sponge best, and came back with his face painted as Batman. The crowds were swelling and the stall was constantly busy from 11am to 5pm.

At the end of the day we had about 8 prizes left over. We were confident we had made a good sum.
On counting out we made £478.41. A good day for all!

The Carnival Team: Jamie, Amanda, Barbara, Shannon, Izzy, Mark, Phil and Eddie

Joan's Theatrical Fundraiser

Grandmother to Seren-Rose (MPS I), Joan Davis took some time out of her performance in Oklahoma in her local theatre to collect funds for the MPS Society. Helped by Seren-Rose's sister, Bethany, the pair, dressed in eye-catching costumes straight out of the Wild West, spoke to theatre-goers about MPS and the work of the MPS Society and collected £200!

Many thanks to Joan and Bethany for putting on such a show!

Charity of the Year

One great way to support the MPS Society, and raise awareness for MPS and related diseases, is to approach your employer to vote for the MPS Society to become the company's Charity of the Year. Quite often, charity of the year status is decided by the employees, so we need you to stand up and get us heard!

You could even arrange for a fundraiser involving your colleagues – anything from quiz nights to dress-down days have proved very popular in the work place.

Of course, we would be more than happy to provide you with any information you might need, just give us a call or email fundraising@mpssociety.org.uk.

Team Hugo Climb Mount Kilimanjaro

A huge thank you to Paul O'Donnell, Brian D'Souza, Bernard Xavier and Giles Brend – AKA Team Hugo, who climbed Mount Kilimanjaro and raised an incredible £7,622.50 for the MPS Society!

Brian D'Souza, whose son, Hugo, was unfortunately diagnosed with Hunters Disease in 2010, wanted to support our work towards the advancement of treatments of these rare genetic diseases – and he certainly did succeed in doing just that.

Congratulations, Team Hugo!

London 10k Thankyou

A huge thank you to all our London 10k team who completed the iconic race in July! We hope that you enjoyed the day and soaked up some of the great atmosphere that the London 10k is known for. Your efforts and your training are very much appreciated!

If you would like to enter the British London 10k as part of the MPS Society team, please email us at fundraising@mpssociety.org.uk and we would be happy to put your name on the list for 2015. If you are successful, you will enjoy one of the world's greatest road races, passing many of the capital's iconic location, such as St James's Palace, the Golden Jubilee Bridge and Big Ben.

Jane's Cycle Challenge

Having lost 3 stone, I began cycling to help keep the weight off and to build on my fitness. I then decided that I needed something to aim for and the Macmillam Cycletta was in nearby Woburn. My daughter Emilly agreed to join me as moral support and, boy, on the day I needed that support!

We chose the MPS Society as our friends the Coopers' 10 year old daughter is affected by MPS. We first got to know Hannah and her family when Hannah started at the school where I worked and my daughter, Emilly, worked for a while as Hannah's lunchtime one-to-one. She then began babysitting and we have since become great friends.

Hannah is an inspiration to many people, always attacking life with enthusiasm and humour. Hannah and her sister, Emily, joined us our training rides and were there to support us on the day.

The ride itself was much harder than I expected, with lots of hills and at times I nearly gave up but with my daughter's encouragement and the knowledge that every day Hannah has her own hills to climb, helped me carry on and complete the challenge. I felt so proud of myself and couldn't have done it without the encouragement of people on the day and the generous sponsorship of so many friends and family.

MPS Awareness Day Coffee Morning

Teresa Jeffery and her son Corey (MPS I), organised a coffee morning with a difference at the Preston Baptist Church to mark MPS Awareness Day. Coffee was not the only thing on the agenda, but the morning also included a tombola, a bouncy castle, bring and buy, face painting, tattoos and a blue cake sale. Corey's cousin Scott even bravely withstood a sponsored leg wax, and Corey's other cousin Steve had his head shaved. We commend their dedication!

Everyone there embraced the cause and Wore It Blue, and the morning also held a 'Guess the Weight of the Blue Cake' competition, which was kindly supplied by local entrepreneur Karen's Cakes.

This fun-packed coffee morning bought in a spectacular £639!

Well done to all involved!

Charlotte & Ben's Fundraising Adventures!

Brother and sister, Charlotte and Ben Davis began their fundraising by washing cars in their local area – but not content to stop there, the intrepid duo then undertook a sponsored climb of Pen y Fan, which is situated in the Brecon Beacons National Park. The peak is the highest British peak south of the Snowdonia mountain range, but the imposing landmark did not stop Charlotte and Ben, who trekked all the way to the top and back – needless to say they were very tired afterward!

The pair finished off their fundraising by collecting outside ASDA in Aberdare – giving them an incredible total of £234!

In their own words, Charlotte and Ben's mission was "to raise awareness for this genetic condition because our sister Seren-Rose was diagnosed with it and we wanted people to understand it. We know she's not very well but to us she's our little sister and we love her to bits".

A massive thank you to Charlotte and Ben for their heroic efforts in raising important funds and awareness for the MPS Society.

Growing Star Montessori Nursery School

Growing Star Montessori Nursery School in Malawi supported MPS Awareness Day by Wearing It Blue!

Teacher, Raihana Seedat, encouraged her students to wear blue for the day and pledged to pay £1 to the MPS Society for each child who did just that. Raihana organised the event for the first time this year, inspired by her niece, Aisha Seedat, who suffers from Morquio and who is an active fundraiser herself in this country. One of Raihana's aims was to also raise awareness in Malawi, where very few people know of MPS, or understand its effects.

We think that Raihana did brilliantly, and it was lovely to see pictures of students from Growing Star Montessori Nursery School wearing blue for the day! Altogether Raihana managed to raise a fantastic £128.

7 marathons in 7 days

Inspired by a 6 year old called Archie

I stood at the summit of Mont Blanc, the tears that had fallen from my eyes had already frozen before I realised why my eyes were burning. Another dream was complete but this challenge was a big tick off my bucket list of life, I knew it was going to take something very special to reward me with such a challenge as this, as I slowly descended with my fellow climber I was already thinking about my next challenge ahead.

I'm somewhat fortunate to work from home, located in a small cul-de-sac of just four homes, I tend to see who comes and goes up the driveway on a daily basis. I live next door to a young six year old lad called Archie Pearson. He's not just any normal boy growing up rapidly outgrowing his clothes, but he is my inspiration to live my life with no boundaries. Archie has a rare genetic disease called MPS, sadly a life limiting disease that comes in various forms but this doesn't stop Archie from living his life with limitations, wearing a brace to support his spine and weekly treatments to try and prevent the disease getting worse. In the six years of knowing Archie I've never heard him complain of his disabilities, he runs, jumps, cycles and plays in the driveway like any normal fast growing boy without a care in the world. It puts the daily complaints or worries my wife and I have into true comparison and these are the reasons why it's an honour to be able to fundraise for the MPS Society as Archie continues to inspire me to attempt other great feats, large and small.

The 7 Marathons in 7 Days took nine months of preparation, training, logistics, compiling a support team and then there was the route I was going to take with a group of chosen runners. During my training and a discussion with Archie, I found out his dream was to meet the superhero Batman, so I made a few calls to Gotham City and explained Archie's dream. My fingers were crossed as my sixth Marathon was to be in aid of the MPS Society and we hoped Batman could visit when Archie would see us off at the Grand Pier of Weston-super-Mare, making our way to Wells.

The morning had arrived, my legs ached, arms sore from my previous five marathons, but I had made it to mile 135 and we had just two more marathons to complete the dream. It was a Saturday morning on 31st May, the sun was breaking through and on arrival and a crowd of friends and family were already stood at the start line. Archie was dressed in his Batman costume and his friends played without knowing what was about to happen.

Suddenly a beautiful convertible Bentley arrived, pulling up onto the promenade and with our superhero Batman perched on the back of the car. Archie looked up in disbelief as Batman pointed to him and said "You're Batman". Archie replied, "No, you're Batman". The cameras clicked as Batman helped our own little superhero onto the back seat of the car and Archie was handed a gift from the

Caped Crusader. As I was completing my dream of running 7 marathons in 7 days, another was complete as Archie's dream had finally come true, meeting Batman. Archie sat chatting to Batman as once again the tears began to fill my eyes: I now knew I would push through my own pain barrier that day without complaining with my fellow runners and reach the final marathon. I thanked Batman for coming to visit us and as I turned to face the crowd I had never seen so many smiling faces, including the biggest smile I've seen from Archie himself.

I'm delighted to say I arrived at my destination on the Sunday evening, completing 7 marathons in 7 days with my fellow runners, a total of 200 miles through extreme pain and blisters, pushing beyond the pain barrier and blocking out negative thoughts with huge positive support from our drivers, our medic, fellow runners, our family, friends and local residents across Somerset, a once in a lifetime memory while also getting to run seven consecutive marathons.

For me, I was following in the footsteps of the great Adventurer Sir Ranulph Fiennes, but totally inspired by a young six year old boy called Archie Pearson, achieving what some said was impossible, completing the unthinkable of running 7 marathons in 7 days.

My next Adventure challenge is ... well, that would be telling, but I'm sure to let you know when it's confirmed, with such inspiration like Archie Pearson, life has no boundaries.

Jason Smith
www.jasonsmith.me.uk
@ChallengerWSM

Wear It Wicked

With autumn here, the nights getting darker and Halloween just around the corner, now is the perfect time to embrace the spirit of the season to Wear it Wicked for MPS!

In recent years we have heard many people bemoaning that Halloween has become a money-making scheme, but what if we could make it into a fundraising scheme? Talk about being Wicked to do some good!

So if you would like to support the MPS Society over the autumn months, why not don a scary, spooky or crazy costume and Wear it Wicked! Whether you want to be an angel or a demon, Frankenstein or Dracula, or even the Lone Ranger and Tonto, all you have to do is wear your wicked costume to work, school, college or university and make a donation to the MPS Society – and don't forget to get your colleagues, friends and classmates involved too! The more the scarier! And don't forget those Halloween treats!

Or, if you want to get out and about, why not terrify your town by organising a Zombie Walk and shuffle your way around local landmarks? Just get friends, colleagues and family to join your legion of the walking dead, stock up on fake blood and start practising your best moan!

- Choose any of our easy ways to donate:
- Text MPSS01 £2/£5/£10 to 70070
- Call 0845 389 9901 to donate over the phone

Send a cheque (made payable to the MPS Society) to this address: Society for Mucopolysaccharide Diseases, MPS House, Repton Place, White Lion Road, Amersham, Bucks. HP7 9LP

Donate online via JustGiving or Virgin Money Giving – just look for the MPS Society

If that sounds like fun but you're unsure of where to start, drop us an email to fundraising@mpssociety.org.uk and we can send you out a fundraising pack and sponsorship forms, which will give you all the information you will need.

Remember to send us in your ghoulish photos and stories for our Winter magazine, or post them up to our facebook or Twitter pages.

Happy Halloween to all our supporters!

Mud & Glory

This past year, we have seen increasingly more of our supporters not just running, swimming or cycling, but also leaping, sliding, swinging, and generally getting very muddy, as they participated in various challenging obstacle courses throughout the country. With obstacles boasting such names as “Ladder to Hell” and “The Plunge”, it is not hard to imagine that far from being a walk in the park, these events are built to challenge a participant’s strength and stamina, as well as his/her resolve and fortitude – all things our supporters seem not to be lacking!

For those brave and resilient enough to want to take on one of these monumentally challenging events to raise money for the MPS Society, here are a few places to start looking:

The Tough Mudder has so far proved to be the most popular with our fundraisers, with locations in London and the Midlands. As a 10-12 mile obstacle course, this gruelling event requires a steely determination and fosters a great sense of camaraderie.

Book early for a place on the Tough Mudder for 2015.

www.toughmudder.co.uk

Designed with the help of ex-military personnel, the Total Warrior team allows you to choose between a 10k or a 10 mile course in Leeds or the Lake District, complete with 25-30 fiendishly challenging obstacles. Will you unleash the warrior within?

Total Warrior places can sell out months before the event, so book early and receive an early bird discount.

www.totalwarrior.co.uk

Voted as the world’s leading obstacle race, the Reebok Spartan Race takes place in locations around the world, and more than 500,000 have taken part in Spartan races to date. A global ranking system is a nice touch which will appeal to those with a competitive streak.

Choose between ‘Sprint’, ‘Super’ or ‘Beast’, each course varying in length and number of obstacles, but with no easy option!

Again, book early to avoid disappointment.

uk.spartanrace.com

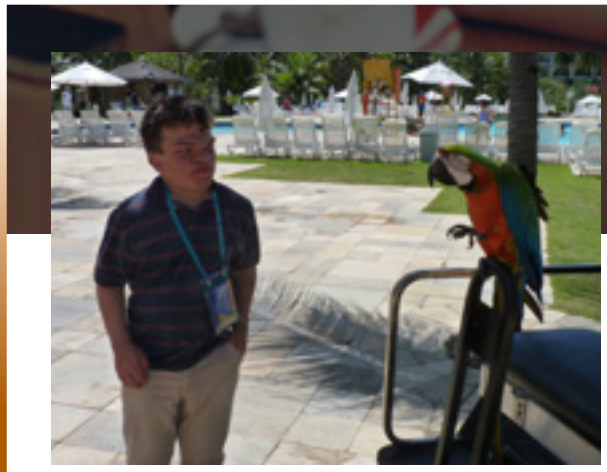
So if you want to fundraise by completing something challenging but satisfying (and be able to forever boast to friends and family), why not get off the couch and sign up for an experience you won’t forget!

If you do decide to undertake an obstacle course, remember to drop us an email at fundraising@mpssociety.org.uk for a fundraising pack and any advice on how to make the most of the event.

International

Brazil Ambassadors Interview those in the know

It wasn't all sightseeing, swimming and fun; our Ambassadors in Brazil were tasked with interviewing six prominent people of the MPS World; despite being hard to track down.



Jessica Reid

I had a brother Daniel who had MPS II (Hunters). He was diagnosed when he was about 3 years old: back in around 1980. My parents, Jane and Neil Reid were introduced to Christine and Robin Lavery whose son Simon had the same condition as Daniel and was a similar age. They found it a great support and comfort to meet a child just like their own and to be able to talk to parents going through similar struggles. Sadly, Daniel died when he was aged nearly 9 – myself and my twin brother, Josh (thankfully unaffected) were only very little (2½ years old) so I don't have many memories other than people's stories and photos.

I have always supported the MPS Society, through attending the family and sibling days as a child while my parents attended the conferences, then onto fundraising in my later years (lots of crazy cycling competitions and charity quiz tournaments) and more regularly, becoming a Trustee of the MPS Society, bringing my legal skills (in my capacity as a solicitor) to the board.

Thomas Garthwaite

Thomas Garthwaite is 20 years old and is currently studying at Oxford University. Thomas and his younger brother Louis both suffer from Hunters, but have both been very active supporters of the MPS Society.

Jessica Reid and Thomas Garthwaite interviewed:

Barbara Wedehase

Barbara is a Genetic Counsellor and has a Masters in Social Work. She has run the National MPS society for the last 14 years and is based in the USA.

She believes ERT has been the most advanced development in the last decade: especially now there is financial backing for it.

She is excited that we are on the cusp of significant change as there are more companies developing drugs for the treatment of MPS disorders and believes that this competitive market will bring benefit to members.

Barbara believes the next decade will bring exciting developments in Gene Therapy.

She says "it is critical that all families have hope from the time of diagnosis." The fact there is the current possibility of different clinical trials for MPS III for which there was previously no treatment, is an "incredibly exciting time."

She believes Social Media brings both benefits and pitfalls to families. On the one hand there is the possibility of sharing information and exchanging ideas in a rapid, international fashion, but this may also mean families become fixated on a treatment option that may have worked for someone else's child, but will not necessarily work for their own child. She stressed that as each MPS disorder is so unique (as within each specific disease there are inordinate differences in attenuated forms) families need to bear this in mind.

Barbara praised Christine Lavery for all her hard work in MPS and said the coming together of professionals and families, at the International Symposium is all down to Christine.

Dr Elizabeth Neufeld

She was described by many at the conference as "The Bible" or "Queen of MPS" being the person to discover the basic defect causing MPS diseases over 40 years ago. It was thus a privilege to interview her. She liked to draw diagrams in order to explain her research to us and visualise how her results showed significant comparisons between an unaffected person and affected person.

She worked at the school of Medicine in California and has been involved in MPS since 1967 when she conducted some of the very first experiments. She and a friend from Belgium believed a number of diseases were linked to LSD's and then she had an idea of what might cause an LSD. She explained how she set up an experiment, showing that fibroblasts cultured from Hurler and Hunter patients could correct each other's biochemical defect. She discovered they were exchanging something - either enzymes or DNA. In order to make pure enzyme they had to clone it, which back then was very difficult to do.

She believes genetic sequencing (looking at the whole genotype) with unknown diseases is a marvellous idea because that is how you can truly find the mutant gene otherwise you just look at the gene of interest or the enzymes.

Jessica Reid and Thomas Garthwaite interviewed:

Professor Andreas Gal

Professor Gal has now retired as a Professor but is still doing genetic diagnostics. He is now chair of the committee of pre-implantation diagnostics.

He believes ERT has been the most innovative development in the MPS world in the last decade. However he accepts that in the early days there was too much enthusiasm in modifying the nature of the disease; ERT does not eliminate/cure the disease. There is a need to diagnose earlier so the treatments are effective.

He enjoys travelling, music and playing chess if he ever gets any spare time.

Dr Paul Hamatz

Paul is Associate Director of the Paediatric Clinical Research Centre at UCSF Benioff Children's Hospital Oakland, California.

During the last 15 years, he participated in clinical trials with MPS I, MPS II, VI and IVA. He started clinical trials on MPS VI but as there were only 5 affected patients in the USA with this disorder he had to go worldwide and recruited from Austria too.

The International Symposium, with the attendance of affected individuals and their families gives him new ideas about what to change for the patients, such as how to shorten infusion time.

He believes the next decade will be exciting and active to include discovering how to treat the brain more, advances in intrathecal methods and gene therapy.

The focus of the next 5 years should be on new born screening, early diagnosis and awareness of MPS diseases so that treatment can start much earlier and be more effective.

He described his main hobby as travelling to meetings for MPS and participating in these conferences. Otherwise he loves gardening.

> **"I'm excited by genetic testing these days- it is so much easier now to find a mutation. It used to take 2-3 years but now they can do it in under a week."**

Professor Joseph Muenzer

Joseph is a Professor of Paediatrics and Genetics at the University of North Carolina, USA.

He has had a long-standing interest in MPS for 25 years. He did a PhD in Biochemistry. He thought he should then go to medical school in order to get exposure to bio-medical research. He worked in a federally funded big clinical research department. In the early days he volunteered as one of the "normal medical" subjects spending an hour and a half each day in a metabolic room at 45 degrees centigrade to participate in a study about how humans sweat! He then (thankfully!) moved on to paediatric testing and did a research fellowship. It was there that he met Professor Elizabeth Neufeld who asked him to research purifying the Hunter syndrome. He was the first person to work in her lab who was both clinically trained and had a PhD. He is grateful to her for the promotional backing she gave him through his publications.

He learnt by default - he showed that amneon membrane implantation didn't work in MPS patients.

Whilst undertaking his research he came into contact with 20-30 MPS I, II and III patients. He said "MPS picked me, I didn't pick it." Something we can all probably relate to. He learnt about MPS by talking to affected families - not through a book.

He believes that we are at the cutting edge of research especially in relation to neurological advances.

In his spare time he loves to walk and can mostly be found doing the gardening.

Simon Heales

Simon runs the Lysosomal enzyme laboratory at Great Ormond Street Hospital.

He has been interested in inherited metabolic diseases for the last 25 years, but only worked specifically in LSD's the last 6 years. He said "you can't truly understand diagnosis and monitoring without knowing about LSD's.

He described being "bowled over" by the International Symposium because of the presence of patients and families. His team only ever work with urine and blood samples - so this "brings his work to life." He feels coming together like this "brings the barriers down and tells us more about the patients." After all, patients are what it is all about. The MPS International Symposium is unique- "good at bringing families, scientists and doctors together". "It should be how conferences are in the future in other fields." He would like to be able to take his whole team to these conferences, so they can see for themselves the people affected by the work they are doing.

Simon is pleased with inroads being made to gradually get enzyme into the brain. He described Chaperones (which trap the enzyme) as "floating my boat."

He loves that he gets to travel with his job and he enjoys "bailing out" his three children. His eldest daughter is now a Pharmacist.

Dr William Sly

Dr William Sly is a physician who described the first patient with MPS VII (Sly syndrome) in 1973. It was him who discovered the specific enzyme deficiency in a new storage disease called MPS VII.

Whilst he appreciated MPS VII is a very rare disease indeed (there are currently no known patients in the UK with it) his research was able to benefit and apply to other diseases too. His research thus led to developments in Gaucher disease too as well as many clinical trials.

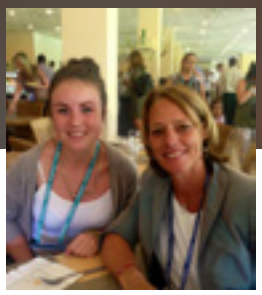
He described himself as being "emotionally attached" to MPS even though he officially retired one month ago.

When asked about what he benefits from attending the International Symposium he said he "used to worry there would be no one to replace the oldies but through the international networks he can see new generations of clinicians, scientists and doctors who are "just as intelligent and passionate about MPS as he and his colleagues were.

He said he was "excited by genetic testing these days - it is so much easier now to find a mutation (it used to take 2-3 years but now they can do it in under a week)."

He told us he has 7 children - and thus drew our attention to the irony that he discovered MPS VII.

When not busy looking after his 24 grandchildren (!) he enjoys baseball.



Rhoswen McKnight interviews

My sister Sarah has MPS I. I have been involved with the MPS Society for many years through my sister and volunteering at UK and International MPS Conferences, and was asked to join the Young Ambassadors Team out in Bahia, Brazil for the 13th International Symposium. This has been such an opportunity for me as it has been the first conference I have been to where I was able to participate in the program and talk to the people involved in the care of the patients suffering with MPS and the people who are researching into new treatments. It was an amazing experience where I met many people with the condition and their siblings, expanding my ever-growing MPS family.

Featured are six very inspirational people that I interviewed over the course of the conference, who gave me an insight on their involvement within the MPS world and their expertise in their field.

Jane Roberts

Jane Roberts is a Paediatric Nurse Specialist for MPS II at the Royal Manchester Children's Hospital, alongside Jean Mercer.

She also worked with Professor Ed Wraith in the 90's and this is how she became involved with the MPS. She studied children's endocrine functions and was encouraged to help Jean on the clinical trials. She then carried out her own studies on MPS II and MPS VI, which lead her to becoming the Clinical Nurse Specialist for MPS.

She believed that the greatest advance in the last ten years is having a designated team to look after the patients. She said it's a more cohesive team with better communication.

Her hobbies included Zumba and socialising.

Jean Mercer

Jean Mercer is a Paediatric Nurse Specialist for MPS I at the Royal Manchester Children's Hospital, which is the National Commissioning Group site for the UK.

She became interested with MPS when she worked with Professor Ed Wraith in the 90's on the metabolic ward. She then went away for a while, and returned to the Willink to run clinical trials for MPS I.

When asked what she felt was the most relevant advance in this field in the last decade she said "the improvement in the outcome of patients after treatment."

When asked about her hobbies, they included spinning, reading and quiz shows.

Lorraine Thompson

Lorraine Thompson is the Adult Nurse Specialist for MPS and works at Salford Royal Hospital Manchester in the Centre for Adult Inherited Metabolic Disease.

Lorraine explained that it was completely accidental how she got involved with MPS as she once worked on a metabolic ward at a children's hospital. She then got a job at Salford as a Specialist Nurse for Fabry, which lead to her to becoming the Clinical Nurse Specialist and Team Manager.

She believes that in the last decade the greatest advance was the growth of an adult service for the patients and a better multidisciplinary team.

In her spare time, she enjoys Northern Soul Dancing and holidays.

Ans Van der Ploeg

Ans Van der Ploeg is the head of the subdivision of Metabolic Diseases at the department of Paediatrics and Chairman of the Centre of Lysosomal and Metabolic Diseases. She comes from Rotterdam in the Netherlands and is based at Erasmus MC University. She told me that she has been working with lysosomal disorders for the past 25 years.

When asked about how she got involved with MPS, she said it was completely accidental, as she was offered to do her PhD in lysosomal storage disorders and it went from there. What she found most beneficial from coming to the conferences was meeting with colleagues to find out what is currently going on in the field and exchanging information. She has also enjoyed the interaction of the scientists and patients in a non-clinical setting.

When questioned about her views on the new advances in genetic testing she replied that she "thought it was interesting, that we would be able to find new patients with metabolic diseases who had not yet been diagnosed, however it may provide challenges in finding the actual defects in the patients and then not knowing the required treatments."

We asked about her hobbies, and she enjoys playing tennis, listening to music, reading and diving.

Shona Tearney

Shona Tearney is a Clinical Psychologist at Birmingham Children's Hospital. She became involved with MPS as she got a new post at Birmingham Children's Hospital for neuropsychology. Before this she hadn't worked in this field. Since then, she has been involved in clinical trials for Intrathecal Enzyme Replacement Therapy (ERT).

She thinks that the most relevant advancement in this field is Intrathecal ERT and after being involved in trials, says "the early signs of the treatment are promising."

Shona's hobbies are tennis, wine tasting and cycling.

Dr Hugh Lemonde

Dr Hugh Lemonde is a doctor at Great Ormond Street Hospital in the Metabolic Medicine Department. He works alongside all the patients meeting a range of needs for a number of different conditions.

He became involved with MPS as he did his PhD in Metabolic Medicine; however it was not direct to MPS, although it was focused around that patient group.

This was his first MPS Conference and he said that what he benefitted from most was "learning more about MPS as a condition and about the current and upcoming treatments for the patients."

When I asked him "what he thought was the most relevant advance in the last decade" he said, "the availability of the therapies and how there is now the knowledge of enzymes and how this understanding has advanced." However, he also noted that he thinks better therapies may eventually become available in the future that may be superior to ERT such as Gene Therapy.

When I asked him about his hobbies, he said "motorbikes and running around after his children."

Simran Bhachu's interviews

Simran has MPS I Hurler-Scheie, and has been an active member of the MPS Society for many years now, with all her family getting involved to support her. Currently, Simran is very busy studying at university. She joined our team on the trip to Brazil as an MPS Ambassador, and below are six professionals that she stopped to speak to.

Maurizio Scarpa

Q1. Which institution do you come from and how does it relate to MPS?

Maurizio is the Director of the Centre for Rare Disease at the Horst Schmidt Klinik (HSK) in Germany. He received his medical degree, paediatric residency and doctorate from the University of Padova in Italy, where he was working with MPS for 25 years. He has now set up a new centre for diseases in Germany, which are related to MPS disorders; he has a lot of patients for follow up and provides them with infusions.

Q2. How did you become interested in this field and what is your main research interest?

He was interested in MPS as he had finished his medicine degree and research working with gene therapy in metabolic diseases. Maurizio was fascinated by the diversity of the diseases and its links with the unique genetic code. His major interest is how to make early diagnoses and how to treat them, new-born screening methods and developing new drugs for neurological diseases.

Q3. What and how does it benefit you from coming to these conferences?

Maurizio saw it as a great opportunity to meet people (patients in particular), exchange ideas, network for collaboration and expand knowledge from other people's experiences.

Q4. What do you think has been the most relevant advance in this field in the last decade?

"The availability of therapy is the major breakthrough, the new systems to make diagnosis and the possibility of administering the drug to the brain."

Q5. What are your views in advances in new genetic testing (sequencing of the whole gene)?

"It is very important, especially when diagnosis is not immediate and difficult to detect. Gene sequencing is becoming cheaper which will help make the diagnoses quicker."

Q6. What are your hobbies?

"Unfortunately I don't have many hobbies as I work too much but my major hobby is playing music and I like horse riding."

Elsa Shapiro

Q1. Which institution do you come from and how does it relate to MPS?

Elsa is a Professor of Paediatrics and Neurology at University of Minnesota and expert in research on Neurobehavioral and Neuroimaging in genetic neurodegenerative disorder. Elsa has been studying MPS disorders for over 30 years.

Q2. How did you become interested in this field and what is your main research interest?

Dr William Tirivit was the first person in the USA to do a bone marrow transplant for Hurler syndrome. He asked if Elsa would collaborate with him, she accepted his offer and that is how she became interested in MPS disorders. Elsa's research is on the longitudinal natural history of these disorders, looking at brain function and things such as how physical symptoms and treatments affect the brain.

Q3. What and how does it benefit you from coming to these conferences?

Here she is able to share results and findings, learn from others about what they have been doing, and receives new ideas for what she can do next.

Q4. What do you think has been the most relevant advance in this field in the last decade?

The most relevant changes are that "there are so many treatments now, enzyme replacements, transplants, substrate reduction and gene therapy is coming. There are many new treatments and some are going to be very effective. That is what is very exciting."

Q5. What are your views in advances in new genetic testing (sequencing of the whole gene)?

She "thinks it is a good thing".

Q6. What are your hobbies?

She loves to travel, likes to cook and is interested in genealogy (family tree).

Lynda Rigby

Q1. Which institution do you come from and how does it relate to MPS?

Lynda works for Genzyme, located in Australia. Her role is senior product manager for MPS. Genzyme are one of the pioneering companies in Enzyme Replacement Therapy.

Q2. How did you become interested in this field and what is your main research interest?

She applied for a job in Genzyme, as part of her interview she had to deliver a presentation on Mucopolysaccharidoses. She had never heard of the condition before, so researched it and put a presentation together. She became really interested and thought that this was an amazing offer to be a part of.

Q3. What and how does it benefit you from coming to these conferences?

There are a lot of benefits from coming to these conferences; she gets to meet a lot of people like me, learn about the diseases and how it impacts on my life and family and the progress that the enzyme has made, which is fantastic. She has the chance to catch up with other clinicians and other departments that look into MPS and how the future is going to be.

Q4. What do you think has been the most relevant advance in this field in the last decade?

She gets to see more and more patients being able to access the medications and not just Aldurazyme. ERT is now also available for MPS II and VI and having lots of other areas to research.

Q5. What are your views in advances in new genetic testing (sequencing of the whole gene)?

"It is amazing as it has implications for all diseases in mankind. We have only scratched the surface."

Q6. What are your hobbies?

She likes running, swimming, reading and walking.

Lorne Clarke

Q1. Which institution do you come from and how does it relate to MPS?

He is Professor, Department of Medical Genetics at University of British Columbia and Head of Genetics & Health research cluster and Senior Clinician Scientist,

As well as researching MPS he also diagnoses and treats patients with MPS.

Q2. How did you become interested in this field and what is your main research interest?

In the late 1980's people were beginning to isolate the genes that cause MPS and that is when he became interested.

Q3. What and how does it benefit you from coming to these conferences?

He says it benefits him in millions of ways. At this conference he gets the chance to meet patients as well as their families and see how the therapy is helping and any issues they may have. He also gets to meet outstanding scientists, who complement and enhance his work.

Q4. What do you think has been the most relevant advance in this field in the last decade?

Lorne believes that the biggest advancement was when all the genes from all the MPS diseases were discovered and isolated and the other advance is the use of enzyme replacement therapy.

Q5. What are your views in advances in new genetic testing (sequencing of the whole gene)?

"The genetic advances are actually fantastic as it allowed us to make early diagnosis that was precise and accurate, so I think the genetic advances in MPS and other LSD's have been outstanding."

Q6. What are your hobbies?

He skis and surfs as well as hikes. He has a huge garden at the back of his house and so does a lot of gardening and a lot of things outside.

Simran Bhachu's interviews

Paul Orchard

Q1. Which institution do you come from and how does it relate to MPS?

Paul comes from the University of Minnesota USA and is a Professor of Paediatrics in the division of Blood and Marrow Transplantation at University of Minnesota.

Q2. How did you become interested in this field and what is your main research interest?

Paul became interested in Paediatrics and was most immersed in the patients with higher needs and decided to train in Oncology (bone marrow transplants). From there, he started to work in the field of bio-chemistry and genetic diseases. His niche became bone marrow transplant in inherited metabolic storage diseases.

Q3. What and how does it benefit you from coming to these conferences?

Paul believes that the conferences are great for a number of reasons. It is great to see the patients, "that is why we are in this field". Meetings that have families are always beneficial. He feels it is good to get together with other people that are in the field to find out what new things are being developed, hear updates on clinical trials and the research that is being done which bring you up to speed on everything going on.

Q4. What do you think has been the most relevant advance in this field in the last decade?

"In the bone marrow transplant field, they are always trying to make things better. As other therapies are being researched, they are being incorporated with transplants for example as enzyme therapy became available in 2003, it was incorporated with transplants. There are also emerging techniques and gene therapy, which will help to open doors."

Q5. What are your views in advances in new genetic testing (sequencing of the whole gene)?

Paul says it's a great question! For most of the diseases it influences how patients are going to do and how other genes are involved. In some way the technology is too advanced and that we don't know what to do with some of the information at the moment. There are so many genes to get information about and to know what is relevant and what is not is an area to focus on.

Q6. What are your hobbies?

He tries to get outside a bit, does fishing here and there and has taken up Karate, although the job keeps him busy.

Brian Bigger

Q1. Which institution do you come from and how does it relate to MPS?

Graduated from Bath University with a degree in Applied Biology and completed a PhD in Gene Therapy from Imperial College London. He received an MPS Society fellowship and in 2006 and set up the MPS Stem Cell Research Laboratory at University of Manchester. With colleagues he works on pathology, diagnosis and clinical development of treatments for neurodegenerative Lysosomal storage disease glioblastoma.

Q2. How did you become interested in this field and what is your main research interest?

He kind of fell into it by chance, he was originally working with Haemophilia, and he helped develop gene therapy, by using stem cells. He tried to get funding to take it to trial, but nobody was interested and he was told that the condition was not severe enough for funding. Through talking to Rob Wynn who ran the blood and bone marrow transplant unit in Manchester, and had a great interest in Hurler and doing transplants; it was thought that this would be a great chance to look at MPS diseases. As these are quite nasty diseases, he thought that this was a great way to take the gene therapy forward in order to make a difference.

Q3. What and how does it benefit you from coming to these conferences?

He joked, "Firstly it's in Brazil!" From his prospective it is nice to get out from the lab. It is a fantastic opportunities to come to a conference like this, talk about gene therapy and how to make it better. "It's a good to come to family conferences to meet patients and to meet individuals like you who have the diseases and to see the impact that the therapy has". Paul said that as he does not get out much it is inspiration to see how the treatments work.

Q4. What do you think has been the most relevant advance in this field in the last decade?

He says; "Without a doubt it has to be enzyme replacement therapy, as it has a huge impact on life". He thinks that ERT has made a huge difference to patient lives but does not think it is perfect and thinks that it has limitations. He thinks that it has a limited impact on Hurler diseases, particularly on neurological function. It is not as good as bone marrow transplantation for treating the bones and joints.

He is going to talk about sleep apnoea and the effect of ERT on respiratory function and how bone marrow transplant is more effective. "Enzyme treatments are not perfect and we need to make it better. So it is important to keep the pressure up. There is a whole plethora of treatments coming along, for example substrate reduction therapy and gene therapy could be on the cards so it is exciting times."

Q5. What are your views in advances in new genetic testing (sequencing of the whole gene)?

He thinks that there are some advantages but also disadvantages in gene sequencing. In many of the MPS conditions it is useful to sequence the gene. There are mutations in the genes which can be predicted and will give a specific outcomes to the patient; gene typing is able to use this. When there are too different alleles, it is more difficult. Certain mutations are clear cut, but some are vague. New-born screening can give a lot of information about who will be affected by the disease, also information about which genes modify enzyme levels. Some patients have higher enzyme levels than others, but the same mutation and this is because we are all different. All factors make a difference to how enzyme levels perform. We all have different genetic makeup and different environmental factors and these can all make a difference in the way in which the enzymes work.

Q6. What are your hobbies?

He is a passionate rock climber and scans the weather forecast to see if there is an opportunity to get out. Brian also does karate has done it for a few years with his children. It is fun and a good way to really relax at the end of a long day.

Aiden Kearney interviewes

Aidan is 36 years old and lives in Ballycastle (Northern Ireland). He is one of 10 individuals living in Northern Ireland to be diagnosed with an attenuated form of Morquio Disease. Aidan has taken part in the Enzyme Replacement Therapy Trial for Morquio Disease and has seen his health and quality of life improve since beginning treatment. Aidan has spoken at the Palace of Westminster for the MPS Society and is travelling to Brussels later this year to address the World Orphan Drug Congress about the importance of clinical trials for rare diseases. Aidan is a regular speaker at our MPS Society Conferences in Ireland.

Michaela Weigl

1. Where do you come from and how are you involved in MPS?

I am from Austria and I run the Austrian MPS Society

2. What drives you to run an MPS Society and help other sufferers in your country?

After taking my daughter Maria to our first conference and meeting other children I thought I needed to do something to help and give them a better quality of life.

3. What do you get out of the MPS conferences?

I like to meet people, and learn from scientists and hear all the talks about the new information to help the Austrian families back home.

4. What is the major factor in improving your MPS Society in the last ten years?

To have help! I have had a paid secretary for the last ten years and just this has helped the Society greatly.

5. What are your views in new genetic testing?

It's great that genetic testing is available for families especially when they are looking to start a family as they can be tested giving them peace of mind.

6. Have you any other interests outside of the Society?

I don't have much time but enjoy being with my family and also enjoy gardening and photography.

7. Where do you hope your society will be in ten years' time?

More awareness in Austria for the MPS diseases and to have more people talking about these conditions.

Michael Beck

1. Where are you from and where do you work?

I am from Germany and I work in the University of Minze in the genetic department.

2. What benefits do you get from the MPS conference?

You get a great experience, having contact with patients and families. It is great sharing new ideas and talking with people from the MPS World.

3. What has been the most relevant advance in your field in the last decade?

The advancement in treatments; there was no interest in LSD thirty years ago and therefore little knowledge for patients and no treatments.

4. What motivates you to work with rare genetic diseases rather than the more popular ones?

When I was at medical school it was always the rare genetic diseases that got my interest.

5. When did you first find out about MPS diseases?

30 years ago I saw a Hunter patient and did not know what he had, so I managed to get some money and funded a small laboratory to help diagnose him and it all took off from there.

Mary Boushel

1. Where do you come from and how are you involved in MPS Society?

I come from Ireland and I started the Southern Ireland MPS Society in 1995.

2. What drives you to run a MPS Society and help others in your country?

To help others. I care about the children and adults. It is very rewarding helping the MPS children, adults and family members.

3. What do you get out of coming to the MPS conference?

To meet members from other MPS Societies, as well as the doctors and pharmaceutical representatives. It is a great opportunity to network and pick up new ideas.

4. What are your views in advances in new genetic testing?

Genetic testing is a major advance in treatment; it will be extremely useful for the children.

5. Have you any other interests outside of the Society?

I am a member of an active retirement group. I play bridge and enjoy hill walking and table tennis and of course all the Gaelic sports for example hurling.

6. Where do you hope your society will be in ten years' time?

I am hopeful that we will have more active participation from our members, and hope that some members can join the committee and help.

Arjan Meutgeert

1. Where do you come from and how you involved in MPS?

My son Rick has a Lysosomal storage disease. My wife and I started the Netherlands MPS Society 20 years ago, a metabolic disease society. We got in contact with Christine Lavery at the MPS society and went from there.

2. What drives you to run a MPS Society and help other sufferers in your country?

It is unfair that people are born with this disease and I wanted to do something about it - to help everyone with MPS because everyone deserves happiness.

3. What do you get out of MPS conferences?

It's really great to meet up and network with each other, which is very important. I love and enjoy organising the children's programme and just love seeing all the MPS children smile and enjoy themselves. That's what I get out of it.

4. What has been the most major factor in improving your MPS Society in the last ten years?

It has to be new discoveries in treating MPS. It's still not a solution and there is still work to be done. In the past ten years there has been an improvement in life expectancy and quality of life.

5. Have you any hobbies or interests besides the MPS Society?

I love to make stain glass windows and recently made an Ajax of Amsterdam football stain glass window for my son. I also like to study philosophy.

6. What do you hope your society will be like in ten years' time?

We hope to have lots more treatments available to offer children.

Nigel Nichols & Alison Warwick

1. Which institution do you come from and how does it relate to MPS

I work for BioMarin UK and Ireland. I work most closely with Morquio patients and look after the clinical trial needs of patients and share resource information.

2. How did you get interested in this field and what is your research interest?

I was mainly interested because my son has a rare disease.

3. What and how does it benefit you to come to the MPS Conference?

It is great to meet and network with people from all over the world in the same field. It's the one time where you can learn and get a lot of new information and experiences all in one place and there is a massive reward from coming and seeing the dedication to research.

4. What do you think has been the most relevant advance in this field in the last year/ decade?

We have shown that ERT works and is safe and has a continued benefit. It took ten years to prove that.

5. What are your views on advances in new genetic testing (sequencing of the whole gene)?

The future is exciting because of the development of gene therapy. It will revolutionise the treatment for MPS patients and hopefully will be available in the future.

6. What are your interests and hobbies?

I like music and gardening, I grow mostly all my own vegetables and like to play golf.

7. How and when did you first find out about MPS?

I found out about MPS through Christine Lavery because my son was diagnosed with a rare disease and Christine was part of an organisation called Contact a Family. Christine has also written a book on rare eye disorders which is a main part of my son's condition.

Guirish A Solanki

1. Which field are you in and how does it relate to MPS?

I am involved in Neurosurgery in the spine for MPS patients

2. Where are you from and where do you work at, and which centre/hospital?

I was born in Mozambique, and went to medical school in the West Indies then came to the UK. Then I met another neurosurgeon from Dublin and worked in Ireland before returning back to the UK to finish training. I am now based in Birmingham

3. What benefits do you get from the MPS conference?

Most of the conferences that I attend are neurosurgery related but Christine Lavery kindly asked me to attend and do a talk. I always learn a lot and it's rewarding to help patients and families out at the MPS conferences.

4. What has been the most relevant advance in your field in the last decade?

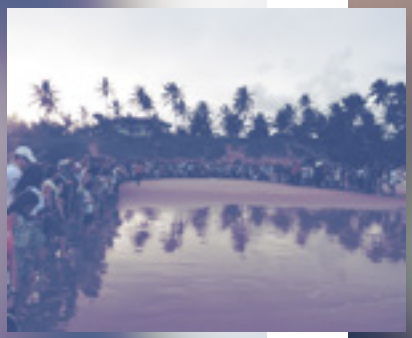
There have been huge advances in neurosurgery in the last ten years and there are huge advances in imaging which makes it easier to do our jobs. There is better communication between doctors and the hospitals.

5. What motivates you to work with rare genetic diseases rather than the more popular known diseases?

I was approached in hospital to help and I said yes straight away. I had no personal experience but wanted to learn. When I met my first MPS child they were very sick, and sadly not long after passed away. From that day on I wanted to stop the suffering of MPS patients and help.

7. When did you first find out about MPS syndrome?

Through medical school you are taught about MPS patients. I was approached to help an MPS patient at the hospital in 2005 and it has gone on from that day.



Sue Peach's Brazil Diary

Saturday 9th August.
At 7.30pm we were all gathered at Heathrow Terminal 5 London full of excitement for our journey to Brazil for the MPS International Conference.

The Delegation were Christine, Jessica, Alison, Aidan, Debbie, Jessica, Rhoswen, Simran, Tom and myself. After two flights and a taxi we arrived tired and hot at 12.30 pm on Sunday 10th in Costa da Sauipe. The exhaustion rapidly evaporated as we set eyes on the Conference resort. Palm trees bearing coconuts, huge butterflies, birdsong, white beaches and the blue ocean left us dumbfounded. This was indeed paradise.

After checking in and freshening up we met for lunch in the hotel restaurant. The range of food available was amazing and so we had a good lunch then went to explore the resort. We walked down to the beach and watched huge waves crash over the rocks, we explored other parts of the Complex and came across the Saupe Kids area which was very impressive in terms of facilities and activities offered (there was even a children's restaurant). One friendly member of staff approached us with a iguana which I had the experience of holding. He told us that the staff were expecting the children of the MPS families and were looking forward to meeting and entertaining them.

As we walked past the pool, we decided that we could do with a relaxing swim to wash away the stickiness of the journey and relax on the sun loungers soaking up the last of the sun and swimming in one of the pools. Aidan was the first to brave the water but Jessica and I soon followed. Rhoswen and Simran eventually joined us but it was beginning to get dark, the tropical rain clouds were gathering and so off we went to shower and change for dinner followed by an early night.

Monday 11th August.
We awoke to a beautiful morning and soon after breakfast clambered into a taxi to take us to Praia Do Forte, a nature reserve and tourist village famous for its preservation work with turtles and nurse sharks

The turtles were incredible! The baby turtles were very sweet. However, we could not believe how huge and old some of the turtles were, enormous and pushing 70 years old. Yet another memorable experience!

A leisurely lunch - everything but the driving is leisurely in Brazil – followed by a little shopping and it was soon time to return to our hotel. We were amazed to see huge puddles around the hotel as we had seen no rain in Praia Do Forte yet there had clearly been quite a downfall only half an hour away. For our young ambassadors the next day would bring whale watching. For Christine and myself the work would begin.

Tuesday 12th August.
Another glorious morning and after a second night's sleep we were all feeling much more refreshed. After breakfast the Whale watching group set off. With some time to spare before the Network meeting started, Christine, Fer Pidden and I decided to walk to the beach. We were delighted to find rock pools teeming with life - multi-striped fish, hermit crabs, sponges and water lizards. Christine, our very own David Attenborough was in her element.

The Network meeting (attended by representatives from 19 countries) took the rest of the day during which, amongst other things, we considered the future coordination of the Network and listened to a presentation about the future of gene therapy.

Meanwhile, the UK Young Ambassadors (Jessica, Rhoswen, Tom, and Simran) with the two advocacy officers, Debbie and Alison and a young man from the Austrian MPS Society got up early to go on a hump back whale watching trip. After 20 minutes on board the small boat, they saw their first whale, followed by a mother and her calf which was incredible.

The whales didn't want to come out of the water much, but they saw plenty of their blow holes and bodies. The weather turned stormy and so they had to turn back to shore after about an hour and a half on board. The group were then shown a presentation about hump back whales,

their preservation and learnt many interesting facts about them. They then spent the afternoon eating Gelato (ice-cream) and shopping round the tourist streets before heading back to the spa at the hotel.

Wednesday 13th August.
Following a tropical storm in the night, Wednesday dawned bright and fresh. Again Christine and I had a full day of Network meetings as well as a working lunch with a pharma company. Updates about clinical trials from 7 companies were given; decisions were made about the hosting of the 2015 Network Meeting and the 2018 International Symposium. There was also a frank discussion with industry about the funding of future symposia and a presentation from Brian Bigger about the Gene Therapy Programme in Manchester and the Genistein Project.

Meanwhile, the rest of our party enjoyed the pool and prepared their work for the Conference as some were speaking whilst others had to think of relevant questions as they had each been tasked with interviewing six prominent scientist or head of a National MPS Society or some other eminent person in the MPS World as part of their role as young ambassadors.

Thursday 14th August.
We awoke to a torrential downpour so my plan for a swim before breakfast was postponed.

The Conference started in earnest today with most of the over 1044 attendees now at the hotel.

After the opening words of welcome, we listened to an excellent lecture by Lorne Clark entitled "What we already know about MPS and what we don't yet know about MPS" although some of the science went over my head the issue about the mechanisms that underlay the disease symptoms in addition to storage raised very interesting questions about the development of therapies.

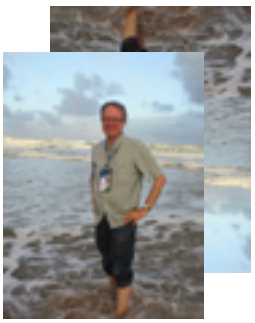
Unfortunately, for some from the UK there had been a flight delay of over 36 hours at Heathrow and by lunchtime we were concerned that they had been further delayed. Fortunately, shortly after lunch they arrived just in time for Bryan Winchester, one of our trustees, to deliver his talk on gene therapy which was very well received.

Apart from listening to presentations Christine and I attended several meetings working through both lunch and dinner. Meanwhile, our young ambassadors were interviewing their chosen victims! They were hard to track down because all the eminent scientists were in high demand and catching up with their international colleagues.

Yet another tropical storm arrived and it rained solidly for several hours. Additionally, the Wi-Fi, which was spasmodic anyway, went down completely. Several people were in a state of shock - how could they survive with no access to emails, Skype, Google? Gradually, the shock wore off and everyone started talking face to face with each other. What a novel concept! It was mooted by some that governments should introduce a law that Wi-Fi be turned off for a couple of hours during the day so that old fashioned ways of communicating survived.

It had been a very busy and mentally exhausting day so as our last meeting ended at 9.00pm Christine and I decided that we had earned a gin and tonic and an early night.

Friday 15th August.
This morning's Family Conference opened with a presentation by Uma Ramaswami about "What can be learned from Natural History Studies". It was clear to me that these studies are very important in understanding not only the development of MPS diseases but also the efficacy of various treatments and they also provides a vital platform for clinical trials. Uma also commented on the importance of Disease Registries for helping to select patients for clinical trials. In her conclusion Uma stressed again the importance of patients taking part in Natural History Studies so that the diseases can be better understood and new therapies developed.



Simon Heales

Simon Heales followed Uma and gave us a bio chemistry lesson about bio markers and metabolic pathways. He delivered this lesson in terms that the families could understand and with humour unlike some speakers who blinded us with science and left us more confused than when they started! I particularly liked his photo of the statue in Bruges of the little boy weeing to illustrate the collecting of urine.

Next up was our very own CEO, Christine. She spoke about the treatment options for Mucopolysaccharidoses. As always her presentation was well planned, clearly presented and very relevant for her audience of mainly families and patients. She addressed the benefits of HSCT for Hurlers and ERT for Hurler Scheie. She also described how ERT had benefitted Hunters and would hopefully benefit Morquio patients in the future. Whilst talking about MPS VI, she spoke of the success of one of our members who has set up a business selling useful gadgets for the home that are particularly helpful for people with disabilities.

Christine also commented on the problems in accessing treatment not only in some developing countries but also in parts of the UK. Christine spoke about clinical trials and the importance of them being done safely and quickly which meant that the right patients had to be enrolled.

Fer Pidden was the next to speak and she spoke with great passion about her experiences of living with her daughter, Natalie, who was born with Sanfilippo A - the ups and downs, the laughter and the tears. The importance of the MPS Society in her life was clear "We were not alone."

- continued next page

Fer spoke about her involvement with the MPS over the years - how she had raised funds, spoken at conferences around the world including at the Palace of Westminster. Her eloquent presentation summed up the life of many MPS families. Around the auditorium it was clear that many had shared those experiences. Natalie had changed Fer's life but she had been empowered by Natalie's diagnosis to fight for a better life for all MPS families and careers. For so many of us, after the shock of that initial diagnosis, we have also been spurred on to do things we had never thought we would. It was an incredibly positive presentation.

After a coffee break, we were treated to an exciting talk about emerging therapies for MPS I, II and III by Joseph Muenzer of the USA. This covered the crossing of the blood brain barrier using Intrathecal ERT delivery including a phase II/III trial for adult MPS II patients; anti-inflammatory treatments; Stop-Codon Read-Through Therapy; Gene Therapy trials being planned for MPS IIIB and MPS VII.

We have waited so long for possible treatments and now there appear to be a considerable number on the horizon.

Yet another UK based speaker was next, Chris Hendriksz, he informed the audience about the progress of ERT for MPS IVA. He talked about the importance of having a therapy that would improve the quality of life for patients. Improving mobility means the ability to deal with everyday tasks, and less pain (even though sometimes greater mobility means more pain, most patients were happier to have more mobility). Chris talked about the Phase 3 study which tested the safety and efficacy of Vimizim including the side effects. The practical aspects of receiving the treatment were also explained. Again this presentation was pitched at just the right level for the family audience.

At lunch Christine and I had yet another meeting so afterwards we felt that we had earned a break so we took ourselves off to the pool for some sun and a swim. The azure blue sky, the coconut palms swaying gently in the breeze, the calypso music blaring over the loud speakers and the chatter of the parrots ensured that we could really relax in this tiny spot of paradise.

However, it was soon time to get back to work. I had a report to write, Christine had another meeting and our young ambassadors had interviews to do so we gathered our towels and headed back to the hotel.

Saturday 16th August

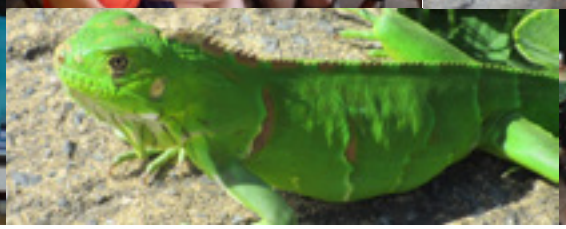
The last day of Conference and we were all a little weary as we had stayed up late talking. I had a particularly interesting conversation with Chris Hendriksz and his wife about the psychological effects on young people after transition and was fascinated and delighted to hear that he had organised a two year study and received a grant to employ a psychologist to look at this.

The first session was about surgery for MPS patients. Claire Garthwaite spoke eloquently about the correction of Louis's knock knees and the greater mobility this had given him. She continued with a description of the experimental surgical procedure carried out on Tom to widen his windpipe to aid his breathing. This had made a terrific difference to Tom's quality of life. It was a fascinating talk and many other families were keen to speak to Claire afterwards to find out more. Further talks covered surgery for carpel tunnel syndrome, spinal cord management, and leg surgery.

After coffee we were treated to two talks from Elsa Shapiro who is always fascinating to listen to. She presented data about the cognitive loss and challenging behaviour of children with MPS II and then the issues surrounding adolescence in MPS I, II and VI and in particular the key role of parenting, commenting that too many parents inhibited their children's social development by being too protective and not encouraging greater independence.








During this session Alison Wilson and Debbie Cavell presented the results of a study carried out by the MPS Society on Gastrointestinal problems in MPS Disease. The talk was very well received and elicited several questions. The final talk of the morning was by Felipe Negro who has Hunter. He described his life and how he had come to be ordained as a Catholic priest and has his own congregation. It was truly uplifting.

I spent the rest of the afternoon in a meeting. Then there was the Remembrance Ceremony on the beach. Literally hundreds of us, mainly dressed in blue and white, snaked down the path and over the narrow wooden bridges which led to the most wonderful sandy beach. It had been raining for most of the day but the clouds parted and the sun smiled on us as we placed our flowers of remembrance into beautifully decorated baskets.

A Bahian band played music and sang, a rescued turtle was returned to the wild and then the baskets of flowers were taken out to sea in a small traditional rowing boat and placed in the water. It was the most wonderfully moving ceremony and as we left we could see the baskets being tossed by the huge waves - an offering to the goddess of peace and love.

At the gala dinner, a further surprise was sprung upon us. Bryan Winchester was presented with a Lifetime Award for his scientific contribution to MPS. The British contingent sprang to their feet and cheered and applauded Bryan as he went forward to collect his award. Bryan confessed afterwards that he hadn't heard his name called out as he was too busy talking. We all felt so proud of Bryan who well deserved this accolade and it was a fitting end to an amazing week.

On Sunday as we had a 5 hour wait at Rio International airport, before our flight back to the UK, Jessica organised a mini bus to collect 7 of us and take us swiftly round the main sights in Rio. We were driven up a windy mountain and got as close to the Christ the Redeemer statue as we could – by then it was getting dark, and the statue was fully lit up glowing in the dark. We saw wonderful views over Rio – a scene our cameras could not even begin to capture. We drove past Copacabana beach and saw the cable cars up to the Sugarloaf Mountain. We all vowed to return to Rio in the future!

Jessica Reid

Trustee and Ambassador Jessica Reid, gives us her account of the International MPS Symposium.

"Have the strength to believe. Do not give up. There truly is hope in our genes."

This was my first ever MPS Conference as an adult (having previously only attended the child programme in my younger days!) I was immediately impressed – seeing MPS on this international scale was phenomenal: 81 speakers from all continents, 1044 registered participants, affected families and patients travelling thousands of miles to attend. It was a real privilege to attend the International Symposium in Brazil.

I attended the Family Conference Programme. I learnt a lot about new treatments and clinical trials on the horizon, gene therapy etc.

There were some talks given by affected family members that put a tear in my eye - in particular Fer Pidden's account of her daughter's struggles (she had Sanfilippo and died in 2009) and Mark Dant's account of his child's experience of Intrathecal ERT. However, as the conference went on I felt more and more uplifted. It was wonderful hearing how the struggles of MPS patients had been overcome by some, including fierce determination to graduate from school/college.

The Gala Dinner with everyone dancing and laughing was a unique and uplifting experience that I will never forget. So too was the beach remembrance ceremony.

The resounding message from the International Symposium was clear:

"Have the strength to believe. Do not give up. There truly is hope in our genes."

Research & Therapies

Dose Responsive Effects of Subcutaneous Pentosan Polysulfate Injection in Mucopolysaccharidosis Type VI Rats and Comparison to Oral Treatment

Michael Frohbergh, Yi Ge, Fanli Meng, Nesrin Karabul, Alexander Solyom, Alon Lai, James Iatridis, Edward H. Schuchman, Calogera M. Simonaro

Abstract

Background: We previously demonstrated the benefits of daily, oral pentosan polysulfate (PPS) treatment in a rat model of mucopolysaccharidosis (MPS) type VI. Herein we compare these effects to once weekly, subcutaneous (sc) injection. The bioavailability of injected PPS is greater than oral, suggesting better delivery to difficult tissues such as bone and cartilage. Injected PPS also effectively treats osteoarthritis in animals, and has shown success in osteoarthritis patients.

Methodology/Principal Findings:
One-month-old MPS VI rats were given once weekly sc injections of PPS (1, 2 and 4 mg/ kg, human equivalent dose (HED)), or daily oral PPS (4 mg/kg HED) for 6 months. Serum inflammatory markers and total glycosaminoglycans (GAGs) were measured, as were several histological, morphological and functional endpoints. Overall, weekly sc PPS injections led to similar or greater therapeutic effects as daily oral administration. Common findings between the two treatment approaches included reduced serum inflammatory markers, improved dentition and skull lengths, reduced tracheal deformities, and improved mobility. Enhanced effects of sc treatment included GAG reduction in urine and tissues, greater endurance on a rotarod, and better improvements in articular cartilage and bone in some dose groups. Optimal therapeutic effects were observed at 2 mg/kg, sc. No drug-related increases in liver enzymes, coagulation factor abnormalities or other adverse effects were identified following 6 months of sc PPS administration.

Conclusions: Once weekly sc administration of PPS in MPS VI rats led to equal or better therapeutic effects than daily oral administration, including a surprising reduction in urine and tissue GAGs. No adverse effects from sc PPS administration were observed over the 6-month study period.

Phase 2 Proof of Concept Clinical Trial for MPSI Patients Naïve to ERT or HSCT who have One or Both Disease Causing Nonsense Mutations is Recruiting

Based on an evaluation process and in discussion with outside experts, PTC Therapeutics based in New Jersey has selected Mucopolysaccharidosis type I (MPS I) as the next indication to pursue for Translarna. It is PTC's goal to initiate a Phase 2 proof-of-concept study for MPS I in the fourth quarter of 2014.

Globally, MPS I occurs in about 1 in every 100,000 births. It is estimated that 60-80% of MPS I patients have their disease as a result of a nonsense mutation. There is no cure for MPS I and enzyme replacement therapies do not sufficiently address the central nervous system, skeletal or cardiac symptoms associated with the disorder. PCT recognise there is urgent need for the development of new treatments targeting the underlying cause of MPS I.

If you are a patient, parent or clinician interested in recruitment to this study please contact:

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Tel.: +49 (0) 611 43 – 2325
Fax: +49 (0) 611 43 – 3196
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or

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Phase I/II Enzyme Replacement Therapy Clinical Trial for MPS IIIB

BioMarin Announces a Phase I/II Enzyme Replacement Therapy Clinical Trial for MPS IIIB Starting June 2015 BMN-250 is an investigational enzyme replacement therapy for MPS IIIB using recombinant human NAGLU with an IGF2 tag, or Glycosylation Independent Lysosomal Targeting (GILT) for enhanced lysosomal delivery.

BMN-250 is a potential first-in-class therapy for Sanfilippo B patients has the potential to directly address the neurologic complications of MPS IIIB using BioMarin's patented technology which provides the capability for direct brain delivery.

Synageva Biopharm

Synageva Biopharm, is developing and delivering medicines that make a meaningful impact on the lives of patients. We select innovative therapeutic approaches for rare diseases based on high unmet medical need and ability to have a substantial impact on disease course and patient health. They have several therapeutics in development including enzyme replacement therapies (ERTs) for lysosomal storage disorders (LSDs) and other programs for the life threatening conditions.

One key element of their strategy is to:

Advance SBC-103 toward human clinical trials for the treatment of MPS IIIB or Sanfilippo B syndrome.

Synageva's research laboratory in Lexington, MA, has protein engineering capabilities and expertise in the rapid production of recombinant proteins for biological characterization. These capabilities allow them to engineer and manufacture proteins that are either identical to the defective protein or incorporate unique modifications to enhance the biological activity and/or therapeutic usefulness of the defective protein.

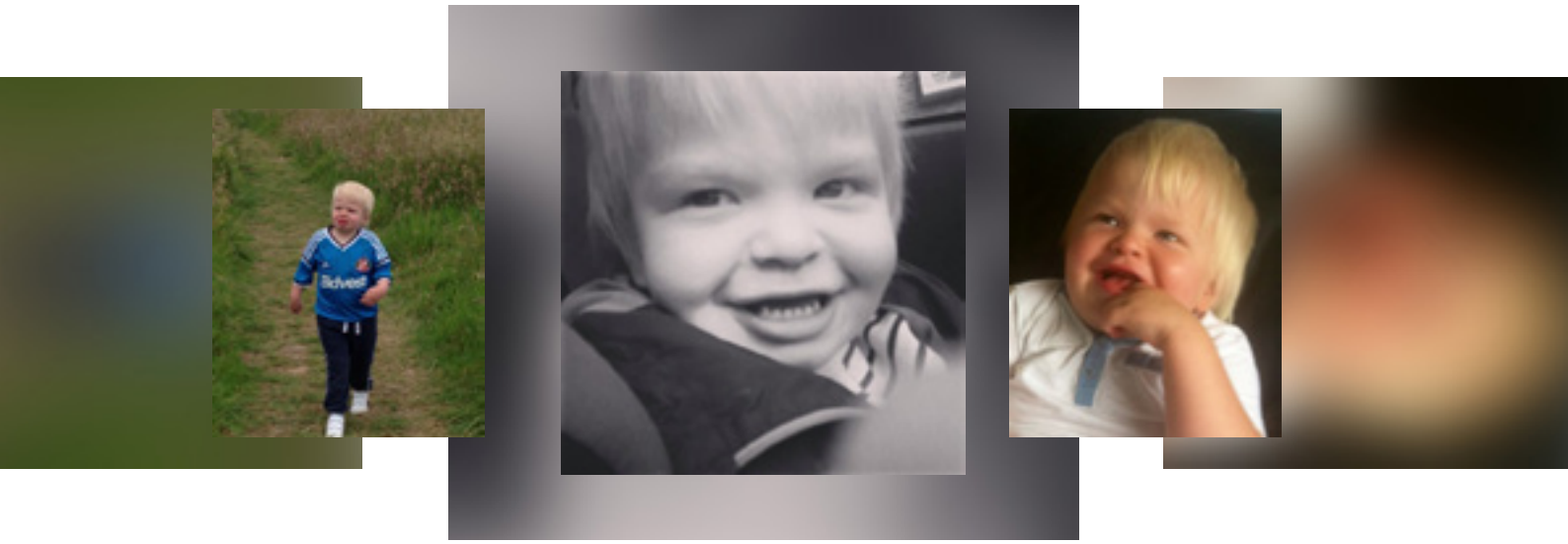
Belgium Questionnaire

What makes a new medicine valuable to you – a survey

KU Leuven, in association with King's College London and Orphanet Inserm, are currently holding a survey to find out more about the experience of those suffering with a rare disease, and how they would define the importance of a new medication. This survey is open to both patients and caregivers in order to get a good understanding of the effects of living with a rare and genetic condition.

We have selected 500 of our members at random to receive the survey. While this is optional, we would appreciate it if you could complete it and send it back as we believe these surveys will help to give our members a voice. Please be assured that your response will be anonymous, and your decision to participate in this study will not affect your usual medical care. As a survey respondent, you will have the opportunity to enter a prize draw to win €50 in Amazon vouchers. Respondents who wish to be considered for the prize draw will be requested to provide their email address, which will be kept completely separate from the answers given in the survey.

If you have not received the survey, and would still like to participate, please visit our website (www.mpssociety.org.uk) and you will find an online version under the 'New & Events' heading.



Jack Watson

Clinical Trial for Sanfilippo Disease Begins

Researchers at The Royal Manchester Children's Hospital and The University of Manchester have recruited Jack Watson as their first child into a new study, which aims to evaluate the clinical effectiveness of a treatment developed in Manchester.

Jack has the condition Sanfilippo Disease, also known as MPS III which affects around one in 85,000 people in the UK for which there is currently no effective treatment.

People with Sanfilippo Disease have too much of the substance heparin sulphate in their cells, particularly cells in the brain, because they lack the enzyme that usually breaks the heparin sulphate down. It is thought that Genistein Aglycone works by blocking the production of heparan sulphate and associated damage to the cells.

The research funded through the MPS Society in partnership with other MPS Societies from around the world have to date donated €376,000 with €120,000 still to raise to enable another six children to be part of the Genistein Clinical Trial.

Genistein is a naturally occurring chemical found in soya beans. In the study the researchers will use a synthetic version, Genistein Aglycone, to maximise absorption through the gut.

Previous research in patients with Sanfilippo Disease has shown that low doses of Genistein reduce the heparin sulphate in the blood and urine, but are not sufficient to be effective in the brain. However, research at The University of Manchester using higher doses of Genistein Alygone in the mouse model of Sanfilippo Disease has shown that this is effective in reducing neurodegeneration.

"This new study is open to children aged 2-15 years old. Patients taking part in the study will receive either Genistein Aglycone or placebo (an inactive substance that looks like the treatment) with food, over a period of 12 months. After 12 months all children will receive Genistein Aglycone for a further 12 months" explains Dr Jones who is also Honorary Senior Lecturer at The University of Manchester.

It is anticipated that the study will take around three years to complete with patients attending up to nine clinics. We are looking forward to hearing the results of the trial and hope that the treatment will benefit patients.

Jack's Case Study

Jack Watson, who is four years old and from Sunderland, has the condition Sanfilippo Disease, also known as Mucopolysaccharidosis (MPS) III, which affects around one in 85,000 people in the UK.

Jack, who goes to Sunningdale School, was diagnosed with MPS III when he was 16 months old. Gemma Nelson, his mum noticed that something wasn't right from the day Jack was born – the way he slept, the way he breathed (and stopped breathing), and the way he opened his bowels.

Gemma, explained: "Jack's condition means that he doesn't speak much, is very hyperactive and has to be sedated to go to sleep. Jack isn't very interested in toys, but he loves going to school, and out elsewhere. He enjoys going on the bus and going to Asda, where everybody knows him.

"It took some time to diagnose Jack. Initial visits to my GP suggested that he was tongue tied, had asthma, and had a chest infection. When his fontanelle (soft spot of the skull) didn't close up, we were referred to a doctor at the Niall Quinn Children's Centre at Sunderland Royal Hospital, who knew instantly that Jack had a Lysosomal Storage Disease, but we weren't sure immediately which one.

"We found out about the clinical trial through The MPS Society, which was working with Dr Jones at The Royal Manchester Children's Hospital. We got involved initially in 2012, by helping to fundraise for the study and have raised around €20,000 to date. Family, friends and the community have been brilliant at fundraising and Jack is now a bit of a celebrity in the area. We have had sponsored skating (7 miles on roller skates), bag packing at the local supermarket, parachute jumping, charity nights and one friend, who had long hair down to the bottom of her back shaved it off to a grade 2.

"I was dancing around the kitchen when I found out that Jack was an eligible candidate for the trial. After waiting for so long the trial is now at stage where we can begin treatment. We know that the treatment is not going to save Jack, but if the drug works we will have more time to make more memories, and memories are priceless."

MPS IIIA and IIIB Gene Therapy Research Report from Nationwide Children's Hospital, Columbus Ohio

Nationwide Children's Hospital, Columbus Ohio have developed two effective gene therapy approaches for the treatment of MPS IIIA and IIIB. These approaches target the root cause of these diseases, the genetic defect of the missing enzymes, by delivering human NAGLU or SGSH gene using AAV9 vector. AAV9 vector can transduce broad tissues, including the CNS and somatic tissues. More importantly, AAV9 has the ability to cross the blood-brain-barrier.

Therefore AAV9 provides an ideal vector for MPS III gene therapies because the lysosomal storage pathology manifests cells in virtually all organs, although the CNS is the major concern in MPS III disorders. In the preliminary studies in mouse models, by a single IV AAV9 vector injection, they were able to achieve lifelong restoration of NAGLU or SGSH activity and the correction lysosomal storage pathology throughout the CNS and in the majority of the somatic tissues. Importantly, the treatments led to the functional neurological correction and extended survival. To expand the clinical relevance, they have also tested the AAV9 gene therapy approaches in older MPS IIIA and IIIB mice and demonstrate that both MPS IIIA and IIIB are reversible to a certain extent. A strong team at Nationwide Children's Hospital has been formed to move their MPS IIIA and IIIB gene therapy projects forward for Phase I/II clinical trials in patients and are currently in the process of interacting with the FDA.

Stem Cell Gene Therapy for MPS IIIA only Two Years from the Clinic

Sanfilippo disease is a devastating paediatric lysosomal storage disorder (LSD) caused by mutations in the SGSH gene. SGSH enzyme deficiency leads to the accumulation of complex sugars that become toxic to cells, particularly in the brain. This causes progressive mental decline and death in early childhood. Less severe LSDs can be treated with enzyme replacement therapy, but for Sanfilippo disease the enzyme is unable to get into the brain where it is needed. Currently there are no effective treatments.

The Stem Cell & Neurotherapies group led by Dr Brian Bigger at the University of Manchester are conducting some exciting research developing a new stem cell gene therapy treatment for Sanfilippo. The technique utilises pioneering technology where lentiviral vectors (modified viruses) are used to introduce correct copies of the defective SGSH gene into patients own stem cells. Following bone marrow transplant the modified stem cells are able to transform into cells that can migrate to the brain where they can produce SGSH enzyme and correct disease in neighbouring brain cells. This innovative approach holds great promise and the group have already shown that the treatment can correct the disease in mice with Sanfilippo. Furthermore, other researchers using a similar strategy have seen promising results in ongoing clinical trials for other genetic conditions including: Metachromatic Leukodystrophy (MLD), Adrenoleukodystrophy (ALD), X-linked Severe Combined Immunodeficiency (SCID) and Wiskott-Aldrich Syndrome (WASP).

The Stem Cell & Neurotherapies group have recently secured a prestigious grant from the Great Ormond Street Hospital charity in collaboration with Prof. Bobby Gaspar at Great Ormond Street Hospital, for the pre-clinical workup of the lentiviral vector that I will be working on. Over the next two years my work will continue to complete pre-clinical safety, efficacy and toxicity testing where the vector will be rigorously tested to ensure safety in readiness for a phase I/II clinical trial. With this grant we aim to have everything in place, including a large batch of clinical grade lentiviral vector, to perform a subsequent clinical trial in two years time that we will shortly be seeking funding for.

Dr Stuart Ellison

ArmaGen Announces Strategic Licensing and Collaboration Agreement with Shire to Develop AGT-182 for Treatment of Hunter Syndrome

ArmaGen, a privately held biotechnology company focused on developing revolutionary therapies to treat severe neurological disorders, have announced that it has entered into a worldwide licensing and collaboration agreement with Shire plc to develop AGT-182, an investigational enzyme replacement therapy (ERT) for potential treatment of both the central nervous system (CNS) and somatic (body-related) manifestations of Hunter syndrome.

Under the agreement, Shire receives worldwide commercialisation rights for AGT-182. The collaboration between ArmaGen and Shire on AGT-182 will be managed by a joint steering committee, with representatives from both companies. ArmaGen will be responsible for conducting a Phase 1 / 2 study for AGT-182 and expects to initiate the trial before the end of 2014. Shire will be responsible for further clinical development, including Phase 3 trials, registration and commercialization of AGT-182 worldwide.

"The agreement with Shire validates the clinical potential of ArmaGen's lead therapy and its ability to cross the blood-brain barrier to treat the progressive and devastating neurological complications of Hunter syndrome," said James Callaway, PhD., Chief Executive Officer of ArmaGen. "Shire is the ideal partner for AGT-182, based on the company's international research and expertise in serving patients with Hunter syndrome. We look forward to beginning our Phase 1 / 2 clinical trial of AGT-182 in collaboration with Shire and, in parallel, advancing ArmaGen's propriety pipeline of innovative therapies."

"Our agreement with ArmaGen strengthens our long-standing commitment to the Hunter syndrome community to bring forward novel therapies that have the potential to dramatically redefine the treatment paradigm and address the most critical unmet needs," Said Philip J. Vickers, PhD, Global Head of Research and Development at Shire. "AGT-182 has the potential to be an important new therapy to our existing portfolio of Hunter syndrome programs. We plan to apply our proven ability to develop therapies for rare genetic disease to progress AGT-182 as a potential treatment that offers hope to patients with Hunter syndrome and their families."

AGT-182

AGT-182 is a novel, investigational ERT that has received orphan drug designation from both the U.S. Food and Drug Administration (FDA) and the European Medicines Agency (EMA). Using ArmaGen's proprietary technology, AGT-182 is designed to take advantage of the body's natural system for transporting products across the blood-brain barrier (BBB) by binding to the same receptor that delivers insulin to the brain. AGT-182 is engineered by the fusion of the replacement IDS enzyme to an antibody that is attracted to a receptor of the BBB. The IDS enzyme is designed to travel through the BBB attached to that antibody.

AGT-181

AGT-181 is a novel, investigational enzyme replacement therapy (ERT) engineered for the treatment of Hurler syndrome, also known as Mucopolysaccharidoses type I, or MPS I. ArmaGen plans to begin a Phase 1/2 study of AGT-181 before the end of 2014 in patients with this lysosomal storage disorder.

Hurler syndrome is caused by a deficiency or absence of the enzyme iduronidase (IDUA). Hurler syndrome affects the brain and spinal cord in children, resulting in debilitating signs and symptoms that include developmental delay, progressive mental decline, loss of physical function, impaired language development (due to hearing loss and an enlarged tongue), corneal and retinal damage, carpal tunnel syndrome, and restricted joint movement.

Available treatments for Hurler syndrome are unable to penetrate the blood-brain barrier, and therefore do not address many of the severe and progressive neurological complications of the disease.

AGT-183

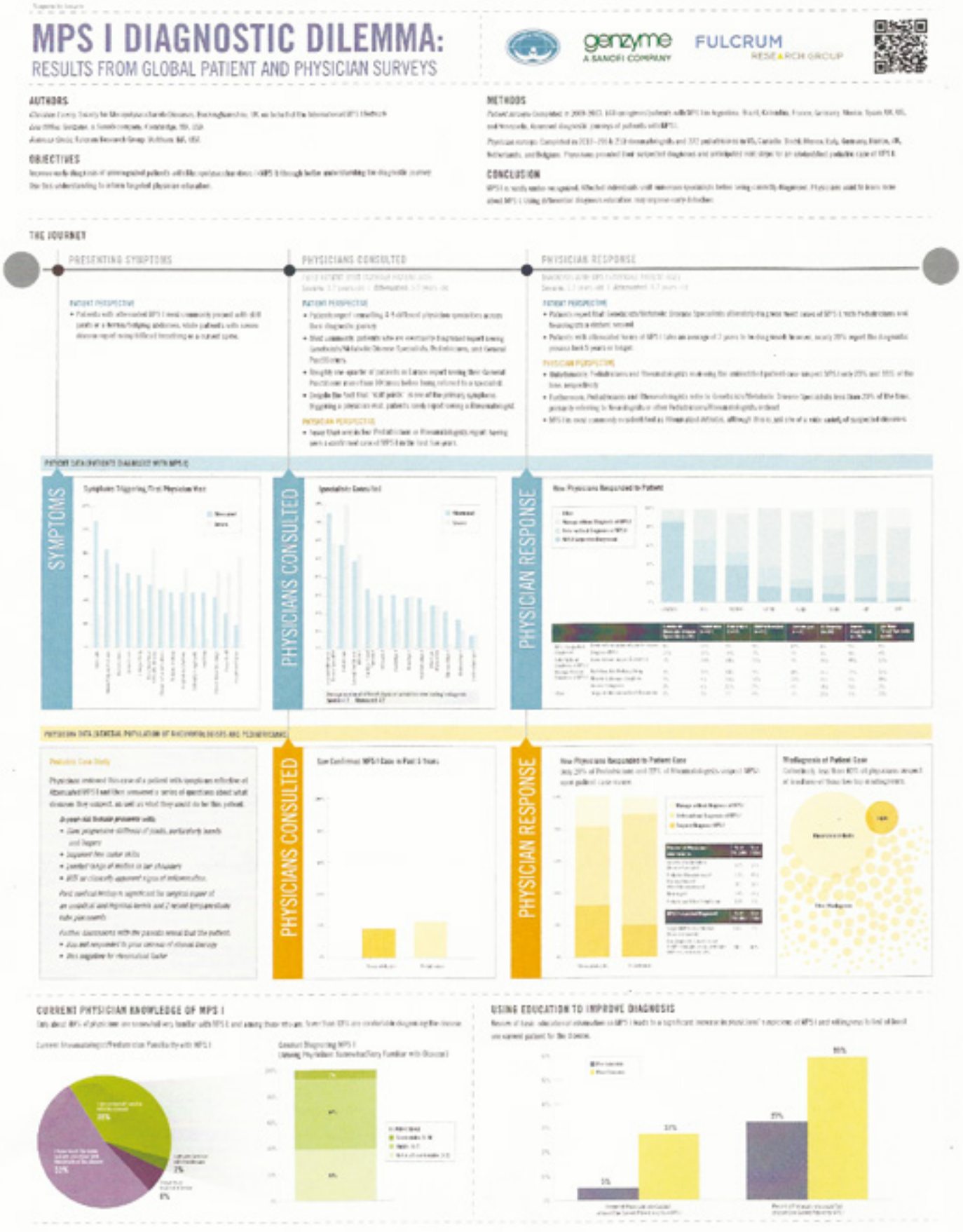
AGT-183 is a novel, investigational enzyme replacement therapy (ERT) engineered for the treatment of Metachromatic Leukodystrophy (MLD), a lysosomal storage disease that arises from a deficiency of the enzyme arylsulfatase (ASA).

This deficiency results in build-up of sulftatides (a type of lipid) in the white matter of the central nervous system (CNS), causing demyelination (destruction of the myelin sheath, the protective cover of nerves in the brain and spinal cord). Demyelination causes a breakdown in communication between the nerves and the brain, leading to paralysis, blindness, seizures, and eventually, death. Currently there are no approved treatments for MLD.

AGT-184

AGT-184 is a novel, investigational enzyme replacement therapy (ERT) engineered for the treatment of Sanfilippo A syndrome, also known as Mucopolysaccharidosis type IIIA (MPS-IIIA), a lysosomal storage disease that arises from a deficiency in the gene encoding for the enzyme N-sulphoglucosamine sulphohydrolase (SGSH), which results in a build-up of complex sugars.

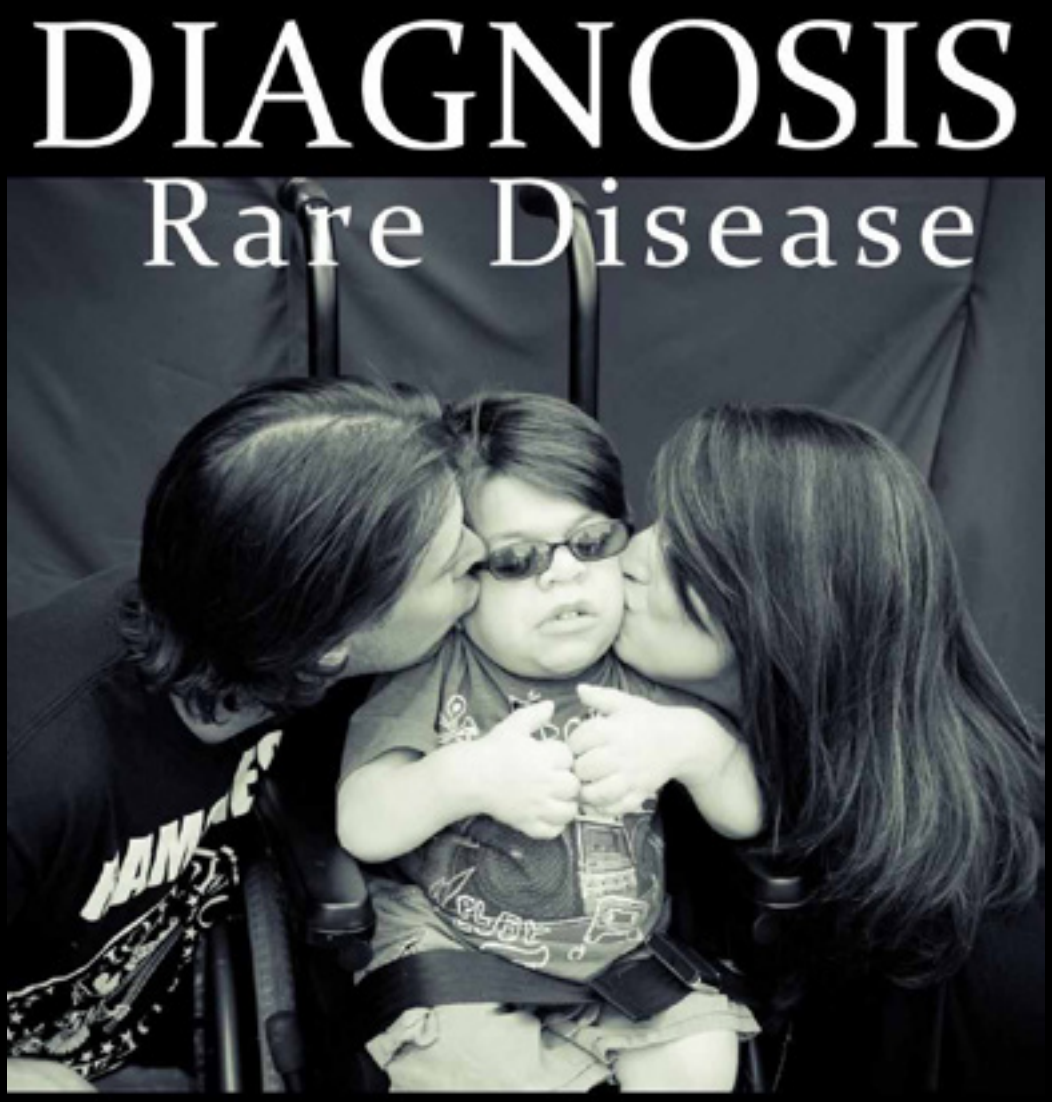
Sanfilippo A syndrome causes progressive intellectual disability and the loss of previously acquired skills (developmental regression). In later stages of the disorder, people may experience seizures and movement disorders. Currently there are no approved treatments for Sanfilippo A syndrome.



MPS I Diagnostic Dilemma poster

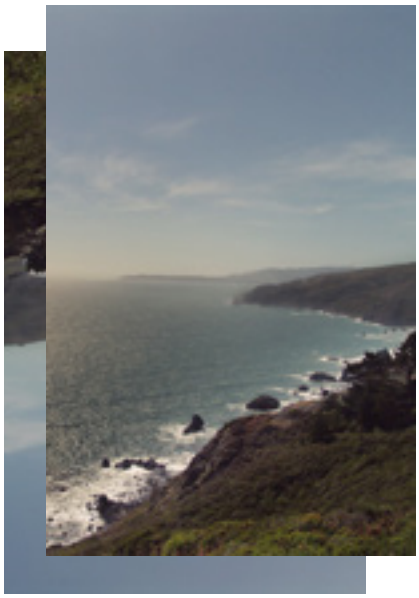
As created by Christine Lavery, Erin Wilkie from Genzyme, and Rebecca Gould from Fulcrum Research Group, in order to improve early diagnosis for patients with MPS I by educating physicians. This was presented at the International Symposium in Brazil earlier this year

Information and Resources



Diagnosis Rare Disease by Denise Crompton recounts the journey of 13 families affected by Mucolipidosis as they deal with the issues involved. The book has been published as an E-Book available at Amazon, Barnes and Noble and Authorhouse, and will eventually be available in a hard copy. The links can also be found on the ISMRD website under news and events.

Other families with a rare disease will recognize themselves in this book. It is a must for medical professionals, especially those who want to understand the families and how to best work with them during these difficult times.



Expression of Hope III

Inviting novice and budding artists from every country to submit art

In 2005, Genzyme launched the Expression of Hope Program to give those impacted by Lysosomal Storage Disorders (LSDs) the unique opportunity to shine a light on these rare genetic diseases. Numerous patient organisations from around the world became involved and encouraged their members to submit artwork that shares feelings of hope and explores the realities, perceptions, and experiences of living with an LSD. More than 100 artists participated, bringing their individual stories of triumph over adversity, and hope for the future to a global audience through art.

Building on that success, the second Expression of Hope Program was held in 2009, providing another opportunity for global collaboration in expressing the impact of living with Lysosomal Storage Disorders. To date, over 300 pieces of art have been created and collected in the Expression of Hope series, increasing awareness for Lysosomal Storage Disease.

This year, Genzyme is pleased to announce Expression of Hope III, inviting novice and budding artists from every country to submit art that explores the experience of living with an LSD. Individuals living with LSDs as well as caregivers, family members and friends are invited to create original works of art that reflect the personal challenges and successes they experience every day.

Care partners and family members can also participate. Artists can submit a digital image of their creation to the Expression of Hope III website at: **www.expressionofhope.com.**

All of the submitted artworks that meet programme guidelines will be exhibited on the Expression of Hope website. Several pictures that reflect the spirit and determination of those affected by LSDs will be selected as Featured Artwork and displayed through global exhibits and materials. All artists who participate in Expression of Hope will be eligible to receive a book of featured art.

You can create a work of art that can help thousands of people around the world learn more about living with an LSD!

Being a Part of Expression of Hope is as easy as 1-2-3:

1) Create an original work of art.

2) Photograph it.

3) Submit a digital image of the art to the Expression of Hope website.

Entry period starting in September.

For more information, visit www.expressionofhope.com.

Genzyme is proud to announce Expression of Hope III

COMING SOON!

A Global Program of Awareness and Inspiration Featuring Works of Art by the Lysosomal Storage Disorder Community

Expression of Hope III is a program that uses the power of art to generate awareness and understanding of the inspirational strength and courage of people living with lysosomal storage disorders (LSDs). Through this special program, people touched by any LSD are invited to create and submit an original work of art that reflects their feelings of hope and explores the realities, perceptions and experiences of living with an LSD.

To enter, original artwork should be photographed and digitally submitted to the Expression of Hope III website (www.expressionofhope.com), where they will be displayed for a global audience. Several works that reflect the spirit and determination of those affected by LSDs will be selected as Featured Artwork and displayed through global exhibits and materials. Artists who participate in Expression of Hope will be eligible to receive a book of Featured Art and information about the artists.

Being a Part of Expression of Hope is as easy as 1-2-3:

1. Create an original work of art
2. Photograph it
3. Submit a digital image of the artwork to the Expression of Hope website.
   (Entry period will be fall 2014. More information to follow.)

For more information visit www.expressionofhope.com.

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