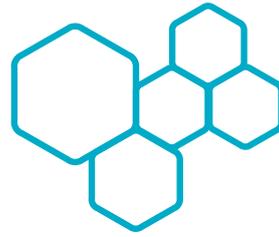


# THE MPS MAGAZINE



<b>Society for Mucopolysaccharide Diseases</b>	<b>Support Research Awareness</b>	<b>Winter 2014</b>
		<a href="http://www.mpsociety.org.uk">www.mpsociety.org.uk</a>

### *Lapland UK Family Day*

Father Christmas and his elves took a group of families on a festive and magical adventure in Lapland UK. Both adults and children alike enjoyed making toys with the elves, meeting huskies, and ice-skating!

Read more on pages 22 - 23

### *The Fight for Vimizim*

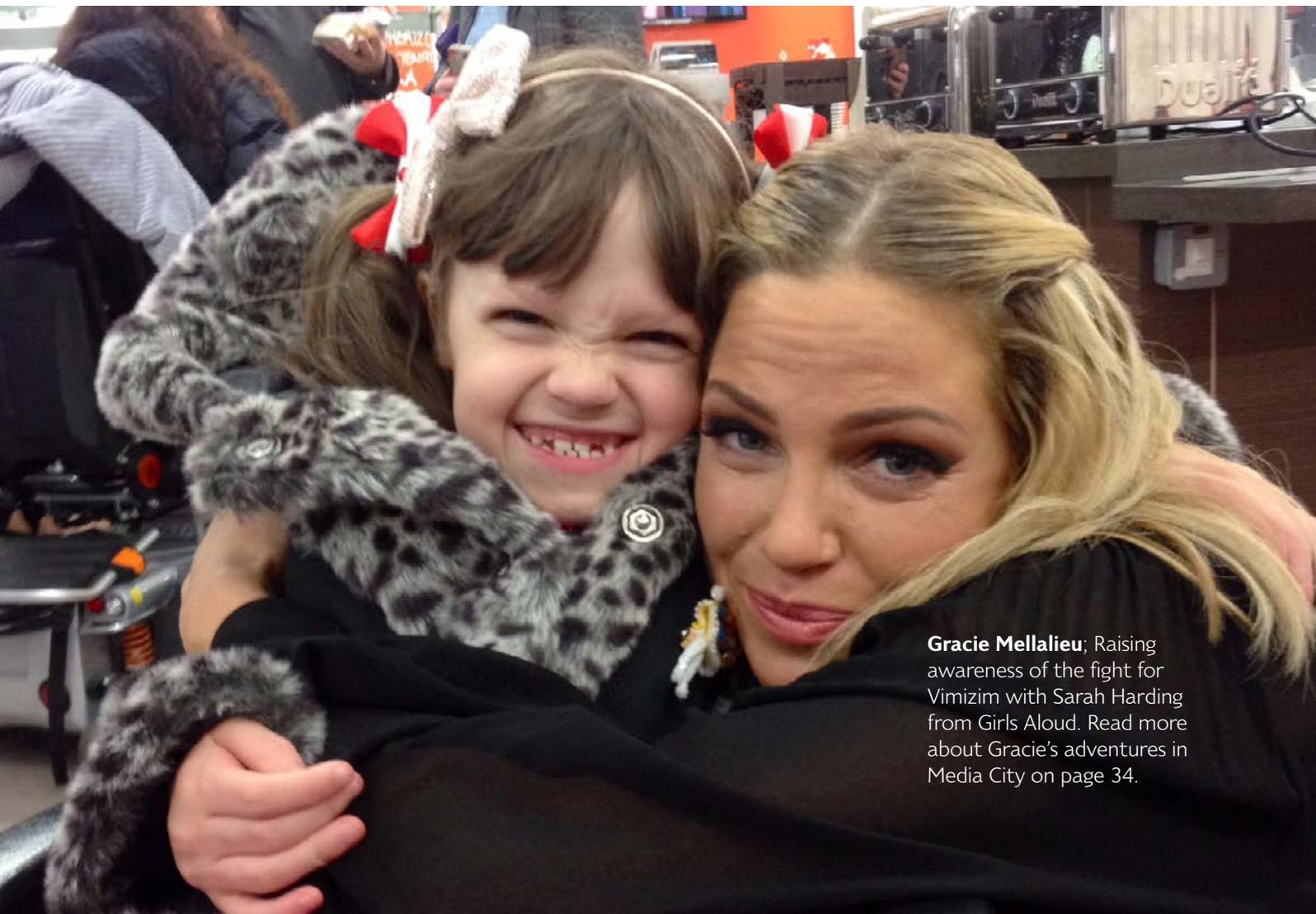
The ongoing fight to get funding for Elosulfase Alfa, and the importance of getting your voice heard is covered by Charlotte Roberts in the 'Treatment' section of this magazine.

Read Charlotte's report on pages 32 - 33

### *Thea's Trust Charity Ball*

On behalf of the MPS Society, James Hope-Gil attended this star-studded ball, organised by Thea's Trust. James received a cheque of £5,000, which was raised from a raffle and auction.

You can read James's account of this fabulous night on page 44



**Gracie Mellalieu**; Raising awareness of the fight for Vimizim with Sarah Harding from Girls Aloud. Read more about Gracie's adventures in Media City on page 34.

Front cover photo: Gracie Mellalieu with Sarah Harding from Girls Aloud

## Society for Mucopolysaccharide Diseases

### Registered Address:

MPS House, Repton Place, White Lion Road, Amersham, Bucks, HP7 9LP, UK

www.mpsociety.org.uk

T: 0845 389 9901  
Out of Hours: 07712 653258  
F: 0845 389 9902  
E: mps@mpsociety.org.uk

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

### Board of Trustees

#### Chair

Sue Peach

#### Vice Chairs

Wilma Robins

Paul Moody

#### Treasurer

Judith Evans

#### Trustees

Tim Summerton

Judy Holroyd

Bob Stevens

Bryan Winchester

Jessica Reid

James Garthwaite

Katrina Fanneran

### Magazine Deadlines

Spring 1 March 2015  
Summer 1 June 2015  
Autumn 1 September 2015  
Winter 1 December 2015

To submit content email:  
[magazine@mpsociety.org.uk](mailto:magazine@mpsociety.org.uk)

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

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

© 2014 Society for MPS Diseases (UK) Rights Reserved

### Staff

Christine Lavery  
Chief Executive  
[c.lavery@mpsociety.org.uk](mailto:c.lavery@mpsociety.org.uk)

Rebecca Brandon  
Advocacy Support Officer  
[r.brandon@mpsociety.org.uk](mailto:r.brandon@mpsociety.org.uk)

Debbie Cavell  
Advocacy Support Officer  
[d.cavell@mpsociety.org.uk](mailto:d.cavell@mpsociety.org.uk)

Steve Cotterell  
Advocacy Support Officer  
[steve.cotterell@mpsociety.org.uk](mailto:steve.cotterell@mpsociety.org.uk)

Sue Cotterell  
Trust & Grant Fundraising Officer  
[s.cotterell@mpsociety.org.uk](mailto:s.cotterell@mpsociety.org.uk)

Toni Ellerton  
PA to CEO  
[t.ellerton@mpsociety.org.uk](mailto:t.ellerton@mpsociety.org.uk)

Joanne Goodman  
Clinical Trial & Patient Access Officer  
[j.goodman@mpsociety.org.uk](mailto:j.goodman@mpsociety.org.uk)

Liz Hardy  
Trust & Grant Fundraising Officer  
[e.hardy@mpsociety.org.uk](mailto:e.hardy@mpsociety.org.uk)

Mak Khan  
Business Developer Manager  
[m.khan@mpsociety.org.uk](mailto:m.khan@mpsociety.org.uk)

Elkie Riches  
Fundraising and Communications Officer  
[e.riches@mpsociety.org.uk](mailto:e.riches@mpsociety.org.uk)

Charlotte Roberts  
Communications Officer  
[c.roberts@mpsociety.org.uk](mailto:c.roberts@mpsociety.org.uk)

Gina Smith  
Finance Officer  
[g.smith@mpsociety.org.uk](mailto:g.smith@mpsociety.org.uk)

Sophie Thomas  
Senior Advocacy Support Officer  
[s.thomas@mpsociety.org.uk](mailto:s.thomas@mpsociety.org.uk)

Martine Tilley  
Office Administrator  
[m.tilley@mpsociety.org.uk](mailto:m.tilley@mpsociety.org.uk)

Alison Wilson  
Advocacy Support Officer  
[a.wilson@mpsociety.org.uk](mailto:a.wilson@mpsociety.org.uk)



## New Look Magazine

As you all may have noticed, our last MPS magazine was somewhat different - the entire look of the magazine had changed, although it still contained all the usual up to date news and articles that you expect from us. We would like to assure our supporters that the magazine continues to be produced in-house and at no added cost.

So far we have received some amazingly positive feedback from members and MPS Society Trustees, so thank you to all who have shown their support.

*If you have any comments on our magazine we would love to hear from you. Just write to us at our office address or email [magazine@mpsociety.org.uk](mailto:magazine@mpsociety.org.uk)*



### 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.

**If you are receiving this magazine by post but would prefer to receive our quarterly magazine by email, please let us know by emailing [magazine@mpsociety.org.uk](mailto:magazine@mpsociety.org.uk).**

## Contents

04 × Chief Executive's Report

06 × Governance and What's On

07 × Announcements

08 × Advocacy

14 × Your Stories

18 × Events

26 × International

30 × Research

32 × Treatment



[facebook.com/mpsociety](https://facebook.com/mpsociety)



[twitter.com/MPSSocietyUK](https://twitter.com/MPSSocietyUK)

36 × Fundraising

## Welcome

As you will be able to tell from the extent of our 'Events' section, we have been busy organising and running lots of special days and weekends for our families. We hope everyone who attended these events enjoyed themselves and took home some lovely memories.

If you follow us on facebook or twitter, or if you are affected by MPSIVA Morquio disease, you will be aware of our desperate campaign to secure funding from the NHS for Elosulfase Alfa (Vimizim). We have made progress, with NHS England admitting that their scorecard system was unlawful, but we still have much work to do. If we can get this important drug funded, the chances of getting other treatments for other rare diseases funded, improves dramatically, so we urge all our members to help in our fight.

As always thank you so much to all our fundraisers - without you we could not continue our work.

Best wishes,

The MPS Team

Visit our online shop

[www.mpsociety.org.uk](http://www.mpsociety.org.uk).

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





## Chief Executive's Report

Christine Lavery

For some of us at the MPS Society and a handful of individuals and parents of children with Morquio disease Christmas just gone was definitely on hold till the very last minute.

As many of you will know Vimizin, an Enzyme Replacement Therapy for Morquio disease, MPSIVA, was approved by the European Medicines Agency (EMA) at the end of April 2014. We worked with NHS England's Women and Children's Clinical Reference Group (CRG) to develop a Commissioning Policy for Vimizin to go forward to the Programmes of Care (PoC) committee on 28 October 2014. A few days before the PoC Meeting I received an email via the CRG from NHS England with a 'scorecard' attached. It beggared belief that a first world government thought they could prioritise its citizens health needs based on a piece of paper more resembling the questions in a family Christmas quiz and where children and adults with Morquio disease would score 'Nul Points' reminiscent of the Eurovision Song Contest in three questions. The difference and devastating truth was that this scorecard was to determine whether children and adults would receive reimbursed Vimizin and look forward to a longer and healthier life ahead or dispensed with as 'Too Rare to Treat and Let Live'.

We wrote to Jeremy Hunt, Secretary of State for Health, ourselves to raise concerns but this letter fell on deaf ears. Consulting with our lawyers, Leigh Day & Co we agreed the only way forward was to launch a judicial review. We were very fortunate that 10 year old Kamal qualified for public funding. Supported by

his Mum, Suzanne, Kamal through Leigh Day & Co sent a letter of intent prior to going to Judicial Review to Jeremy Hunt on 21 November alleging:

- (i) no proper public consultation on the use of the score card
  - (ii) no published policy as to the use of the score card
  - (iii) no published Guidance as to how the CRG or POC should score rare diseases on the score card,
- Therefore the commissioning process currently being undertaken is unlawful.

We could only give 7 days for a response as the next stage of the prioritisation and appraisal process through the Clinical Priorities Advisory Group (CPAG) was due to meet the following week to decide the destiny of our children and adults with Morquio disease using the 'unlawful' scorecard. At one minute to the 5pm deadline on Friday 28 November 2014 we received a letter through Kamal's lawyers from NHS England's admitting that:

1.1. NHS England was itself considering the appropriate approach to prioritisation and specialist commissioning. As part of that consideration it has concluded that further work remains to be done, and that the CPAG meeting scheduled for next week should be deferred

1.2. herefore your concerns about the use of the score card fall away. No decision to use the

scorecard has yet been taken, and no decision of any sort will be taken next week.

1.3. The next step is for NHS England to consider in December the decision-making process(es) to be used in specialised commissioning and how treatments should be prioritised. . The use of a scorecard may be retained or jettisoned entirely, and if retained the form of the scorecard may be varied. The position of rare conditions and "orphan" illnesses will be considered. Before making a decision on the process(es) to be used, NHS England will carry out a public involvement exercise on its proposals.

1.4. Once a decision-making process has been decided upon, NHS England will go on to adopt a policy on elosulfase alpha for mucopolysaccharidosis IV Type A, in accordance with whatever that process may be.

Therefore no process and timelines in place. Cabinet Office guidance requires a minimum of 12 weeks for public consultation.

With the legal challenge over we are focussing on a publicity and media campaign that may need to extend up to the General Election to ensure that we achieve reimbursement of Vimizin and a highly specialised commissioning process for appraising ultra-orphan drugs. We are also asking as many of those affected by Morquio disease as possible to pursue contact with their Member of Parliament. We would also invite everyone in the Society to support this campaign. The voices of members, friends and family who are benefitting from reimbursed enzyme replacement therapy and those who are or have children clinical trials for Fabry, MPSI, MPSII and MPSIII A and B who may face similar challenges if MPS can't secure reimbursement for MPSIVA are really

### MPS online survey using Survey Monkey

The MPS Society is always keen to hear your views on the services we provide, particularly what you have found helpful or whether there are any recommendations for improvements or changes.

Feedback is vital as we are regularly asked to provide monitoring information when making funding applications and to include evidence of service satisfaction when reporting to those grant-making trusts and foundations from whom we have received grants and donations. Without doubt, we could not continue to provide the level of support services that we do without the help and generosity of these charitable organisations.

In October we emailed the link to a short survey (run by Survey Monkey) to ask you for just such feedback

valuable. Later in this Magazine you can read about the remarkable efforts of our members including Sam and his parents from Otley in Yorkshire and 8 year old Gracie from Mold in North Wales who has become the media stars so far of this campaign. Please watch the MPS website for updates on meeting with NHS England and Minister of Innovation and Health, George Freeman.

Away from Campaigning and politics the MPS Society has been preparing to appoint a new Advocacy Officer to support children and adults with Lysosomal Acid Lipase disease (LAL) and by the end of January the Advocacy Team will be fully equipped to provide a comprehensive advocacy support service to these individuals and their families.

We have held four very special events in the past three months, the Scottish Information Day, visit to Lapland UK, the planting in the Childhood Wood and the family week at the US MPS Disney Conference, Orland, Florida all of which you can read more about later in this MPS Magazine.

Enclosed with this MPS Magazine is the MPS Weekend Conference Programme for 26-28 June 2015 at the Hilton Hotel, Coventry. We hope as many members as possible will participate in this unique learning and networking occasion for individuals and families affected by MPS, healthcare professionals, social worker, teachers and the pharmaceutical industry. Please book early to secure your places.

On behalf of the MPS Board of Trustees and Staff Team we send our best wishes for 2015.

Christine Lavery MBE  
Chief Executive

about your satisfaction – or otherwise! – with the services you have received from the MPS Society. We would like to thank everyone who has responded so far and for the comments and ideas you have provided for us to take into consideration.

We have received a good number of responses but there is still time to complete the survey before the end of January if you haven't already done so – it doesn't take long! On average it takes just 5 minutes or less. Here is the link to log into: <https://www.surveymonkey.com/s/YSWSN9D>.

Thank you for helping us to keep up the high standard of our Advocacy and Support services for members. We will be including some feedback on your comments in our next magazine.

## 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 Meetings on the 4-5 July 2014 and the 5-6 September 2014 at MPS House, Amersham.

### Governance

The Chairman, Sue Peach, spoke to the Annual Review, the content of which had previously been agreed with the accounts for year ending 31 December 2013. It was explained this is a very useful tool for fundraising particularly with charitable grant applications.

The Financial Controls Policy was reviewed and the Board of Trustees agreed that subject to some small amendments related all procedures are being carried out according to the policy

### Treasurer's Report

The Trustees were provided with current figures for both meetings. At the September meeting the Wicked Genes accounts were confirmed as filed. The reserves policy was reviewed and the Trustees unanimously agreed the £11,000 underspend from an unclaimed research grant be put in reserves.

### Risk Management / Health And Safety

At both meetings the Risk Register was reviewed and Trustees acknowledged that despite Senior Managers, Antonia Crofts, leaving and Sophie Thomas's maternity leave the loss of key staff had not disrupted the business of the MPS Society. This was attributed to the excellent forward planning by Sophie and her team and the appointment of a Communications Officer and Business Development Manager.

The Trustees accepted that the Business Continuity Plan (BCP) is work in progress and it is expected the BCP to be ready for approval at the November 2014 Board Meeting.

At both meetings a previously circulated Health and Safety Report was noted and agreed.

### Clinical Management

Trustees were appraised by the Chief Executive of the circumstances of the misdiagnosis (clinical incident) of an MPS child occurring in the Department of Clinical Biochemistry attached to Bristol Children's Hospital, the clinical audit report and the support being given to the family.

### Advocacy Report

The Advocacy Service report previously circulated was considered and approved.

Trustees agreed unanimously that the MPS Society will provide support to individuals affected by Lysosomal Acid Lipase (LaL disease) and their families and the appointment of a new advocacy worker was agreed.

### Policies

The following policies were reviewed and agreed without amendments: No. 15 – Receiving Abusive Calls, No. 16 - Out of Hours, No. 20 - Travel and Subsistence, No. 32 - Security of Storage, No. 33 – Copyright. No. 43 – Eye Care, No. 51 – Board Diversity, No. 53 – Office Key Holder No.57 – Information Systems, No. 64 – Volunteer Fundraising

### MPS Commercial

The MPS Society Operating Agreement, Secondment Agreement, template Secondment Letter and template Loan Agreement were approved by the Board of Trustees and will be considered by the the Board of MPS Commercial at their next meeting.

## What's On

### MPS Regional Clinics 2015

#### MPS I - GOSH:

24th March • 14th July

22nd September • 22nd December

#### MPS III - GOSH:

24th February • 26th May

8th September • 8th December

#### MPS IV - GOSH:

10th March • 9th June

13th October

#### Fabry clinic - BCH:

22nd May • 23rd October

#### MPS clinic - BCH:

6th February • 15th May

12th June • 27th November

#### MPS I Post HSCT (over 6 years) - RMCH:

16th January • 17th April

17th July • 16th October

#### MPS I Post HSCT (under 6 years) - RMCH

23rd January • 24th April

24th July • 23rd October

### Conferences and Regional Events

#### Welsh Information Day, Cardiff

16th January 2015

#### MPSI meeting - Irish MPS Society

17th April 2015

#### Scottish Family Weekend, Ayr

10th April - 13th April 2015

#### UK MPS Conference

Hilton, Coventry

26th – 28th June 2015

## Announcements

### Births



On 14th October 2014 at 12.10pm, the Carter family welcomed Milo James into the world. His brother Jacob (MPSI Hurler) was in hospital at the time undergoing his bone marrow transplant. However, they're all now home together with both boys doing well!



Congratulations to the Sims family who welcomed Gracie's little sister, Nancy Gracie Sims, safely into the world on 13th November 2014..

### New Members:

**Kayla** has recently been in contact with the Society. Her son Thomas has a diagnosis of MPSII Hunter. He is 3 years old. The family live in the Norfolk area.

**Sophie** has recently been in contact with the Society. Sophie and her daughter have Fabry Disease. The family live in the Derbyshire area.

**Sarah** has recently been in contact with the Society. Her son Lewis has a diagnosis of MPSVI Maroteaux Lamy. Lewis is 8 years old. The family live in the North of England.

**Zainab** has recently been in contact with the Society. Her son Adnan has a diagnosis of MLII I Cell. Adnan is 5 months old. The family live in the South East.

**Nadine** has recently been in contact with the Society. Her son Jack has a diagnosis of MPS I Hurler. Jack is 15 months old. The family live in the South of England.

**Heather** has recently been in contact with the Society. She has a diagnosis of MLIII. Heather is 29 years old. She lives in Aberdeen, Scotland and her family live nearby.

## 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](mailto: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](http://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**

MPSIII Sanfilippo type A,B, C and D, MLD AGU, Winchester  
Geleo Physic Dysplasia  
Sly, Gangliosidosis, Sialic Acid Disease



**ALISON**

Supports members living in Ireland



**DEBBIE**

MPSIVA Morquio, MPS I Hurler BMT, Hurler Scheie, Scheie, MPSVI Maroteaux-Lamy, MSD, MLII



**REBECCA**

Fabry  
MPSII Hunter  
MLIII / MLIV  
Mannosidosis, Fucosidosis

### Travel Insurance - please note

It has recently come to our attention that a number of insurance companies on the list of insurers on our website have an issue with their software.

The problem appears to be in relation to Fabry disease. Members have informed us that when they are filling in the online form to get a quote, if they highlight Fabry disease it then asks which type of Gaucher disease they have...

The advice that we have been giving members is if this occurs, not to

complete the online form but to ring and speak to an adviser. Otherwise you may encounter problems if you need medical assistance as the company could refuse to pay because you had clicked on Gaucher disease, even though you had no other option.

When we were informed by members of this problem, we contacted the company concerned and appraised them of the situation. They have all been keen to rectify the problem as they want to encourage the business.

It would be very helpful if you find suitable travel insurance for Fabry, or any other of our MPS diseases, to contact us and let us know which company you have had success with so we can pass this on when we are contacted by other members asking for insurance.

You can download our travel insurance information sheet by visiting the downloads section of our website: <http://www.mpssociety.org.uk/support/downloads-resources/>

### Bereavements:

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

**Abigail Harvey** who suffered from MLII and passed away on 24th November 2014 aged 10 years.

**Reece Culverwell** who suffered from Hunter diseases and passed away on 15th November 2014 aged 18 years.

**Simon Mansfield** who suffered from Sanfilippo disease and passed away on 17th June 2014 aged 40 years.

**Craig McDougall** who suffered from Hunter disease and passed away on 6th December 2014 aged 14 years.

**Sam Hitchin** who suffered from Sanfilippo disease and passed away on 1st September 2014 aged 26 years.

## Manchester BMT Clinic

The Willink Centre - October 2014



Ethan & sister Ellie



Avah

It seems a long time ago now that I last visited the team at the Willink Centre at Manchester Children's hospital in October. As always I received a warm and friendly welcome from Jean, Claire and Pauline and the rest of the team. Dr Alex Broomfield was seeing patients that day. It was a real pleasure to meet with the families to see how they are getting on and to meet some new families too. My favourite part of the advocacy role is when I meet with families, it makes it so much better when you can put a face to a name!

Debbie Cavell  
Advocacy Support Officer



Leighton



Yusuf

## Birmingham Fabry Clinic

24th October 2014

A bright and sunny morning found me getting the train to Birmingham Children's Hospital Fabry Clinic.

Arriving at the Out Patients is usually chaos with lots of noise and people everywhere, but it was the start of half term so it was a lot quieter. Much easier to try and spot everyone.

Megan and her mum were first through the doors; Megan should change her name to Rapunzel as her hair is so long she can sit on it. Megan has asked if she can talk to her form about Fabry disease so her classmates and teachers are aware of the condition: a very brave thing to do. She is also looking at doing fundraising for the MPS Society, so well done Megan!

Charlotte arrived with her mum and dad and was very laid back, feet on the table, very chilled. She did have the coolest shoes.

Charlie went very bashful when I spoke to him but he did allow me to take a photo. One thing that they all like when they go to the hospital is that most of them get some form of a reward from mum and dad after the yearly visit to the hospital.

Jensen swept in with his entourage (mum and gran), then was off to go shopping with the girls - yes, he likes clothes shopping. He will be a dream boyfriend in the future.

Well, it was soon time to take the train back home. Looking forward to seeing you all again soon

Rebecca Brandon  
Advocacy Support Officer



Charlie



Charlotte



Megan



Jensen

## Birmingham MPS Clinic



Beau



Joseph



Muqadas



Riley



Will



Zack & family

As usual with the clinics, we were up bright (well that's stretching it!) and early on Friday 14th November...and arrived at Birmingham Children's Hospital looking like a drowned rat.

We were in the research facility, which is great fun as they have lots of toys and interactive sensory equipment; a lovely bright and cheery space.

Zack arrived with his family and he drew a lovely picture of me! Will was next to arrive and wasn't too impressed with having his picture taken. Riley came in like a little tornado and soon settled down to play with Joe and the rest of the children. Muqadas kept everyone on their toes, who had to make sure he didn't do a runner when the doors opened.

I met several new families and it was lovely to meet the children.

I even managed to persuade two new members of the team to do a write up in the magazine (no one is safe when we have a deadline to meet). I thought Theresa Stokes, the new member of the IMD team, reminded me of someone, then one of the families said that she looked just like Stacey from East Enders. What do you think?

Before we knew it the time had come to go home, leaving a mess of toys behind in our wake.

Thank you to all the staff; it's always a pleasure.

Rebecca Brandon  
Advocacy Support Officer



Rebecca as drawn by Zack!



### New Faces at Birmingham Children's Hospital

Hi, my name is Theresa Stokes and I am a new member to the IMD Team at Birmingham Children's Hospital.

I have joined the team as the IMD Co-ordinator. I am here to help look after our specialist nurses from an all-round administration point of view, which in turn allows them to concentrate on nursing and spending more quality time with patients.

It is likely that you will see me around at clinics doing patient surveys, speaking to the parents and patients and putting faces to names. Getting feedback from them is important to us as a team and for the trust, so we can find out from a patient experience point of view what we are doing well and of course if there is anything they feel we could improve on.

I spend three days a week in IMD and two days a week in Rheumatology.

My role here is for 12 months so I look forward to meeting and supporting you.

Many of you have commented that I look like Stacy from Eastenders, so if you call me Stacy I am happy to answer to that!

Theresa Stokes  
IMD & Rheumatology Co-ordinator

Hi, my name is Alice Stewart and I am the new Inherited Metabolic Disorder Research Coordinator at Birmingham Children's Hospital.

I work closely with the IMD Nurses and Doctor's to organise the set-up, delivery and day to day running of research studies that are taking place within IMD in BCH. You may receive postal information or be contacted by me regarding up and coming research trials and studies, but do not worry- if you would like information before that, I am contactable in the Birmingham Children's Hospital IMD team and happy to chat about what studies we have to offer you.

Research is very important to us, especially in order to gain a better understanding of MPS and also to improve how affected patients are looked after, either looking at the safety of current medications or within the invention of new medications.

I hope to meet you soon

Alice Stewart  
Inherited Metabolic Disorders Research Coordinator

### GOSH's Dr Ashok Vellodi Retires

Dr Ashok Vellodi (FRCPC), Consultant Paediatrician specialising in Lysosomal Storage Diseases at Great Ormond Street Children's Hospital, London, has announced that he will be retiring from his post on 28 February 2015.

Dr Vellodi has been involved in Lysosomal Storage diseases, principally Gaucher, Pompe, Fabry and the Mucopolysaccharide and related diseases since the early 1980s. He was at Westminster Children's Hospital with Prof Jack Hobbs when Bone Marrow Transplants were being pioneered for some MPS diseases. When Westminster Children's Hospital was closed Dr Vellodi moved to the then new Westminster and Chelsea Hospital before joining Great Ormond Street Children's Hospital in the 1990s.

Born in India Dr Vellodi has always continued to commit

time to sharing his knowledge through specialist clinics in Delhi, Mumbai, Bangalore and Chennai. In 2011 and 2013 I was invited by Dr Vellodi to join him at his clinics in Mumbai, Bangalore and Chennai. These were deeply humbling and moving experiences for which personally I shall always be grateful to Dr Vellodi for the opportunities he gave me

Dr Vellodi has been paediatric consultant at Great Ormond Street Children's Hospital to so many of our member families over the last two decades.

On behalf of the MPS Society and its members we wish Dr Vellodi a happy and rewarding retirement.

Christine Lavery  
Chief Executive

## All Ireland Advocacy Support Update

We've had a busy time in Ireland over the last few months. As usual the support provided to our families has lead me to all four corners of Ireland. I've been in schools, council offices, classrooms and clinics educating people about MPS and related conditions. I'm always humbled by the faith you, our members, put in the MPS Society Team. We are delighted that you feel that you can come to us when you're not sure where else to turn.

If you live in Ireland (North or South) and have an unmet support need please do not hesitate to get in touch!

As well as the usual advocacy and support work there have been a few events where the MPS Society has been represented. Have a read to find out what we've been up to!

### Disability Pride

As a director of the Northern Ireland Rare Disease Partnership I was delighted to have the opportunity to attend and take part in Disability Pride on the 20th September.



Disability Pride was born in America and has become an annual tradition. The aim of Disability Pride is to celebrate ability in all its forms and to breakdown misconceptions about disability that can be held by the general public. The parade was held outside of America for the first time a few years ago and this was the first time that the event has been held in the UK. Belfast, as it always does, put on a fantastic party and drew in crowds of more than 1000.

I was truly moved by the event and was delighted to be able to share it with some of our MPS Society members (Roma, Aidan and Jibreel). As well as taking part in the parade, Jibreel had a comedy slot on the main stage!



Thank you to all our MPS Society members who attended Disability Pride 2014. The event was a fantastic success and a true celebration of ability and hope for the future.

### The Northern Ireland Rare Disease Plan

As many of you will be aware, in 2012

the Department of Health launched the UK Rare Disease Strategy and the Health Ministers from each of the four home nations signed up to the commitments outlined in the Strategy and committed to develop their own Regional Implementation Plans.

We were delighted that on the 27th October, Mr Jim Wells (Health Minister) attended an event held in Stormont (Belfast) to launch the consultation period for the Northern Ireland Implementation Plan.



This is a huge step forward and something that has not happened without the involvement of many of the Rare Disease Charities in Northern Ireland.

The Consultation period ends on the 19th January and, along with other charity representatives, I will be responding to the proposed plan to ensure that the needs of those living with MPS and related conditions are addressed.

Alison Wilson  
Advocacy Support Officer

## Your Stories



Hannah Brock

### *A little bit of help goes a long way*

Hi, my name is Hannah Brock and back in September 2014, I started at Brine Leas High School. Because I can't walk long distances, I got a motability scooter. This helps me get up to school and back home.

Since I've started high school, I won an inter-house speaking competition; I spoke about how kids like me should be treated like everyone else. I was in the Nantwich Chronicle two weeks later!

This proves what kids can do with a little bit of help!

*Hannah Brock (MPSVI Maroteaux Lamy)*

### *Tara's Success*

Ivan and I would like to share with you our pride in our youngest daughter Tara (MPSI Hurler) for all that she has overcome and achieved during her school years.



Tara with cheeky grin!

In September 2002, Tara started St Bernadette Primary School dressed in her new uniform and pictured with her renowned cheeky smile. Throughout Tara's school years she received support which enabled her access to her learning. During secondary school Tara underwent major surgery on her neck in 2009 and again double leg surgery in 2012 which resulted in long absences from school. However, due to Tara's own determination and the wonderful dedication of her support worker, Tara continued to work hard and succeeded in achieving 8 GCSE's. We are so proud and especially pleased for Tara to know that she achieved this amazing success despite any difficulties that she was faced with.

To celebrate their final year of school, Tara and her friends attended the School Prom held at the Thistle Hotel in Bristol. Since leaving secondary school, Tara has started a Child Development and Care course at college which runs for two years. She is enjoying the new challenges and is looking forward to starting her placement in the next few weeks.

Kate, Tara's older sister has had a busy time also. During her GAP year she travelled to Australia, Indonesia and Thailand and has now moved to Manchester to study History.

Kate and Tara are now lovely young ladies and looking forward to new and exciting times ahead. We have wonderful memories of their childhood many of which were spent with our extended MPS families and friends and look forward to all the adventures ahead of us.

*Maria and Ivan Murphy*



The School Prom

### *Onwards and Upwards*

In May, the O'Hara family chose to walk, run, skip, hoola hoop and blow raspberries to raise money and awareness for Sam Jamil who is now 11 and has Mucopolidiosis type III.

Their first challenge was a 15 mile walk along a coastal path on the Wirral in Cheshire. The weather was harsh with blustery rain and wind whipping up a sandstorm from the coast and pushing against them. Tired and weather beaten, Lily and Morgan completed the walk in 5 hours.

A week later, Morgan completed the 10k Manchester Bupa run in 28 deg heat in 49 mins.

Morgan and Lily's daughters, Jasmine, 7, and Olivia, 4, asked their mummy why they were fundraising. When Lily explained to them that it was for their cousin Sam, Olivia said she wanted to raise money too. This is when they decided carefully what would be the best and fun way to do this. Jasmine loved to hoola hoop and skip, and Olivia decided her best talent was blowing raspberries on mummy's tummy. So the challenges began, and more money came flooding in when video clips of their challenges were posted on Facebook. In total, £900 plus Gift Aid was raised by their Just Giving page and sponsor forms. £80 of the total was raised by Olivia and Jasmine's challenges. We are so proud of the O'Hara family for what they have done to raise the funds. As a family, we all remain close, and the O'Hara's help out in many ways.

Sam will be 12 in November. He was diagnosed with MLIII in 2009. Although Sam is at the lower end of the disease spectrum, his health has begun to deteriorate over the past couple of years. Sam is noticing more stiffness and pain in his legs and joints in general. The cold weather does affect Sam as it seems to slow him down a bit. Thankfully, we have had a good summer this year, so Sam has been more mobile. This in turn means that he is more tired and achy when he has played out with friends.

In the past couple of years, Sam has had 8 plates inserted into his knees to try and help his legs grow straighter, and



he has had operations on his carpal tunnel nerves to try and increase the sensation and movement in his hands. Sam now has the use of a wheelchair for when we go on long journeys as he tires very easily and can't bear his own weight for long. For trips to the park and at home with friends, Sam uses a regular motorised scooter to get him out and about. This helps him to conserve his energy whilst getting to his destination so he has more energy to play when he gets there. Sam is still mad on all sports and has loved the fact that we have had the Olympics, World Cup and Commonwealth games recently. He hopes to become a Paralympian one day or a performer as he loves singing and dancing in the church Am Dram club. Sam still loves the massage and Bowen Technique moves I use on him regularly to help ease his pain and strengthen his muscles to enable him to move and play.

Sam recently won the Head Teacher's award and trophy as he left Primary school. The reasons for winning were that Sam never moans or complains; he tries everything and does his best to adapt where possible.

Sam really enjoyed going to high school in September. This is a daunting time for any child of this age. The school we chose has been very positive in wanting Sam to be able to access all areas of the school and the curriculum and have allowed extra visits for Sam to get used to the school. We recently gained a Statement for Educational Needs for Sam which will help him with any special requirements he may require. We must thank Rebecca Brandon from the MPS Society for helping and guiding us in the process of attaining the Statement. Rebecca has been very supportive and has given us advice when we needed it. As MLIII is a very rare condition, and not many people understand what it is, Rebecca's advice and knowledge has been very important in terms of helping agencies such as the local educational authority and medical practitioners understand what MLIII is and what its implications are.

New school, new challenges and experiences. Onwards and upwards for Sam.

*Shirley Jamil*

## Moving On

Hi my name is James and I have MPSII Hunter. This year I sat my GCSE exams and had decided that after school I wanted to go to college to study agriculture. I called the MPS advocacy team who helped me with the transition from school to college. Rebecca from the advocacy team liaised with the tutors at the college and together we were able to restructure my care plan. This ensured that whilst at college I would receive the maximum support I would need to enable me to complete the course and reach my goals. I feel with Rebecca's help I will reach my full potential.

At the college I study various areas of agriculture such as; crop production which looks at how crops are grown, anatomy and physiology of farm animals, animal husbandry and estate management.

I am currently learning to drive a tractor which I find very exhilarating and our class was given the job of putting up the new fence at the front of the college.

On a Tuesday morning I go the farm and help out if there are any jobs to do.

On Thursday we went on a trip to Carlcroft, a hill farm which is rented by the collage. Most of the time when I am there we do sheep work.

I get personal satisfaction from doing a good job and when the tutors come to me and tell me I've done well it really boosts my confidence. I am really enjoying the course and going to college and have made great friends. All of the skills I have learned are skills that I never had before college and I feel enormously proud of how far I've come so far.

James Fair



### James's Mum wrote:

I would like to take this opportunity to thank the advocacy team at the MPS Society who helped my son James with his transition from school to college.

We were given great support which enabled us to put our case forward so James will receive full support from the college whilst he is studying. Rebecca's support has meant that James move went without a hitch.

The small things make such a difference to our lives enabling James to achieve great things.

James is proof that anything is possible and that having a life limiting condition shouldn't define you or limit choices that you want to make.

Life is there to be lived and a someone once said "Don't count the days, make each day count"



**Simon Mansfield**  
20th September 1975 - 17th June 2014

### For Simon, 1st July 2014

In a way there were two Simons: one was during the early Dennis The Menace years; the other more recently when he was mainly in his wheelchair. Some of you may only know him from these later days but many will remember him from earlier times when he was completely different.

In those days Simon was incredibly happy and talkative and enthusiastic, with absolutely no inhibitions. He glided untroubled from school to school even though he was expelled from at least one of them. Luckily, at home he had a built-in early warning system for some of his 'spontaneous moments' by commenting on himself as he went along: "He's going in the fridge" or "He's eating all the Easter eggs". Or he would instruct his long-suffering brother (in his parents' presence): "You do naughty things, Jonathan!" Yet there was no malice whatsoever in him; everything was forgotten a second later.

Simon had so many obsessions then, which he would shout out with unconstrained enthusiasm, as some of you may remember: Batman, Thundercats, motorbikes, Kirkby; even, bizarrely, the name of the ringmaster at the Hippodrome Circus, who must have been touched by the thought that he had a single fan out there in the audience...

In the car to Liverpool, Simon had his own unique variation on "Are we there yet?" His was:

Where are we going?  
Me: Where are we going, Simon?  
Simon: Kirkby.  
Me: That's right.

Pause

Simon: Where are we going?  
Me: You tell me, Simon.  
Simon: Kirkby  
Me: That's right.

Longer pause

Simon: Where are we going?

Even then, he was awkward in his walking, but he could also be amazingly quick – often subjecting us to the parents' nightmare of their child disappearing in a shop. Luckily he could always be found at the book shelves. Except once, when he was about eight or ten, when I discovered him in a supermarket with the top off a bottle of gin.

In his own way, Simon was very able. He could finish 100 piece jigsaws by himself without even referring to the picture on the box. He had a fantastic memory for the characters in children's classics, sometimes knowing more than his teachers. He knew the story of Jason and the Golden Fleece, even though his teacher only knew it as Jason and the Golden Fleas! Or having an eye-test and identifying the letter "U" by shouting out "Ulysses!"

Simon had no concept of social conventions or expectations. There's a photo of him running in the Warren School sports day with a great smile on his face even though he is coming in well last. And a visit to your house by Simon was always well remembered. One family friend tells me how her hot water system went mysteriously cold after he'd left. She eventually realized he'd discovered their main heating

switch and turned it off. Or maybe you would find water pouring through your downstairs ceiling light because he'd decided to leave all your upstairs bathroom taps running.

In his later years Simon became increasingly disabled, with frequent stays in hospital. Exactly how much he suffered we can never know because by then he no longer spoke, but his expression sometimes gave a clue. I think of the line by George Herbert: "pain hunts his cruel food through every vein." Around this time he depended a lot upon visiting carers from One-To-One and Prestige who all did an excellent job. As did the staff at the Lowestoft Hub who worked with him. They were all more like his friends than his carers. But as at least two people have said to me recently: "He's back with his mother Claire now," and we must all hope that is the case.

In his silent way Simon gave so much to society. He made us laugh so often; he brought out love and affection in so many people, even strangers. I once sat with him in a supermarket café when a lady came up to me, put something on the table, said, "Here's a pound!" and walked off. (I thought: there's money to be made here!)

I once read that if a Hindu in India gives money to a poor person, the rich man should thank the beggar: because the poor man gives the rich one the opportunity to be generous. So, thank you, Simon, for being with us so long and for giving us the chance to bring out the little that is best in ourselves.

Richard Mansfield

## Events



## Howletts Wild Animal Park

On Saturday 30th August a group of MPS families excitedly gathered at Howletts Zoo in Kent for a day of fun together. We were delighted to be able to organise this event following a grant from the Earmark Trust.

The day proved to be a great hit, with both adults and children alike enjoying seeing the many different animals, having time to socialise and eating plenty of cakes and ice cream when we gathered at the café for afternoon tea!

*Jan Gremo, Mum of Nathan, wrote:*

"Nathan who has Sanfilippo A, Rob his dad and me, his mum, were invited to join a group of MPS families at Howletts Zoo in Canterbury on August 30th. Despite the weather in August being unpredictable it managed to stay fine on that day. We live fairly close by and visit the zoo regularly so it was easy for us to get there. When we arrived we met up with old friends and new which was great. It is a good place for disabled people with all the facilities necessary which does make it an easy day out. We all set off at our usual slow pace as we have to chat to catch up on all the news. Steve (MPS Advocacy Officer) arranged for us to all meet up again but mostly we stayed together. Nathan was his usual loud boisterous self and it wasn't always obvious who was making the most noise Nathan or the animals.

One of our first stops was for coffee which then progressed to lunch and yet more chatting. We did see some animals honestly! After lunch we moved onto the elephants and there was an adorable baby elephant called Marimba which was only 3 months old. It was

being very adventurous and copying its older sister. We had a good chat with a keeper who gave us lots of info about the elephants. We continued wandering through the park and relaxing enjoying the animals.

We all met up for afternoon tea and cakes and you guessed even more chatting. Steve's two little girls were busy taking photos of everything including pieces of wood on the ground.

On the way back to the exit we were extremely lucky to see two keepers in with a tiger playing chase with some food. We thought that it was very brave or plain silly but not sure which, however as you can imagine it was very entertaining.

We all had a brilliant day and it was really good to get together and share stories and experiences and also you guessed just have a good chat!"

*Rosemary Nurse, Mum of Lisa, said:*

"Thank you, it was a very nice day, and the weather was good to us as well. Lisa enjoyed herself. It is so nice to meet up with the other families."

*Donna Bown, Mum of Luke, said:*

"We had a lovely day. It was great to meet up with fellow MPS families without travelling so far. Hope we can do this again."



## Childhood Wood Planting

The Childhood Wood planting took place this year on Sunday 26th October and I had the pleasure of being part of this event for the first time.

In 1993 the MPS Society was granted a license to plant a wood of oak saplings in Sherwood Pines that were cloned from the forest's Great Oak. The first Childhood Wood planting took place in 1993. Since then the MPS Society has held annual planting days which has given bereaved families the opportunity to plant a tree in memory of the child they have lost. Remembrance days are also held where families can visit the Childhood Wood with the support of MPS staff. The Childhood Wood has memory boards which have the names of each individual who is remembered along with a message from each family.

Many families come and visit the wood at different times of the year with their friends and families and Sherwood Pines is a lovely place for a summer picnic or refreshing, crisp walk at the cooler times of the year. The Childhood Wood is also open for the public and we have information boards as well to inform them about Childhood Wood.

On the planting day we met at the Clumber Park Hotel for a lunch before heading to Childhood Wood. Sue Peach opened the proceedings and Councillor Pauline Allan welcomed the party to Sherwood Pines on behalf of Nottinghamshire County Council. Commander Judith Swann has supported the MPS Society for many years and spoke about what the Childhood Wood represents. Wilma Robbins then read the very poignant "Remember" poem by Christina Rossetti.

We released balloons with personal messages attached and watched them float up into the sky. Andy the forest ranger had cleared and prepared an area and the families chose a sapling and a suitable spot to plant their trees. The following children were remembered and celebrated:

*Kayen Ramani, Thea Paterson, Edward Morley, Ben Elliot Richardson, Jackson James Whittaker and Gracie Bella Sims.*

I would like to take this opportunity to thank Commander Judith Swann and Councillor Pauline Allan for taking the time to be part of our event. Also thanks to the forest rangers for their hard work and support.

The Childhood Wood is a beautiful, tranquil place within Sherwood Pines and if you haven't had a chance to visit it makes a lovely day out whatever the time of year. If you would like further details, including directions, please take a look on our website.

*Debbie Cavell  
Advocacy Support Officer*

## Scottish Information Day

The MPS Society held its first MPS and Fabry information day at the Beardmore Hotel in Glasgow on 18th November 2014.

The programme contained talks from a mixture of clinicians from Scotland and England and of course our amazing family speakers.

The two Peters, Dr Peter Galloway and Dr Peter Robinson, chaired the first part of the day which covered topics around the Diagnosis and Patient Management, followed by Clinical Management and family stories.

We had a lovely lunch and that gave the families, clinicians and pharma representatives time to catch up. The afternoon session covered Access to Treatment which fuelled lots of questions and interesting discussion.

The final part of the day was about Supportive Care. This was covered by me talking about the role of the MPS Society, and two of our families told their stories.

I think the stars of the show were our family speakers and our sincere thanks go out to you all. Jacquie Mount recited a beautiful poem which we have printed here and this moved us all. Thank you to all the speakers and Chairs and all the attendees for making this day such a success.

Rebecca Brandon  
Advocacy Support Officer

*"To all the Parents with Special Kids"*  
a poem as read by Jacqui Mount at the Scottish Information Day



*You weren't like other children, and God was well aware,  
you'll need a caring family with love enough to share  
and so he sent you to us and much to our surprise,  
you haven't been a challenge, but a blessing in disguise.*

*Your winning smile and laughter, the pleasures you impart,  
far outweigh your special needs and melt the coldest heart.*

*We're proud that we were chosen, to help you learn and grow,  
the joy that you have brought us is more than you can know.*

*A precious gift from heaven, a treasure from above,  
a child who's taught us many things, but most of all real love.*

## Scottish Family Weekend

Thanks to a generous grant from The Adamson Trust, we were able to provide an opportunity for a weekend respite break on 17th-20th October 2014 for Scottish families affected by MPS related diseases. The holiday was booked at the well-known seaside resort of Sundrum Castle Holiday Park on the Ayrshire coast.

For each family we had booked caravans within the park precincts, all very well equipped and where necessary specially adapted for the disabled, offering double-glazing, heating, ramp access and extra wide doorways, grab rails and raised toilet in bathroom, fold down seat in the shower and spacious corridors. One of the families in particular, who have a severely disabled older son with MPSIII Sanfilippo disease, were able to have the full-time help of two of our specially trained volunteers carers over the weekend, staying in the same 3-bedroom caravan, which made their stay that much more relaxed and enjoyable.

Once the groups had settled into their accommodation on Friday afternoon, we had arranged for everyone to join us for a special Friday evening meal together at the Park's on-site restaurant – The 'Tam O'Shanter' – including the MPS volunteer carers and one of our MPS Society Trustees, Judith Evans and her husband, so that parents, carers and children could all feel welcome and get to know one another. Over the course of the weekend, however, their time was their own and adults and children alike were able to enjoy a wide variety of activities at the holiday park, including an indoor pool with flumes, an adventure playground, soft play area, mini tenpin bowling, kids club and evening entertainment; and of course to enjoy the beach, especially on the Sunday and Monday when temperatures were unseasonably high!

Many thanks to The Adamson Trust for this valuable and much-appreciated weekend break.



*We will be holding a Scottish Family Weekend in April 2015 at Sundrum Castle, Ayr.*

*This is a fantastic opportunity for our Scottish families, so please look out in our magazines and facebook page for more information*



## All Ireland Conference 2014

This year the All Ireland Conference was held in September – a little later than usual due to the big Giro D'Italia cycle race arriving in Northern Ireland on our usual weekend in May. Our MPS Community in Northern Ireland were out in force to support the cyclists (some even joined in!).

As usual, we had a fantastic time at the conference! Families travelled from near and far to hear from a range of experts in the field of MPS and to catch up with each other in the lovely surroundings of the Hilton Golf Resort in Templepatrick. We were delighted to have a sunny weekend that allowed us to spend our breaks basking in the sun and watching the Zebre Rugby team recuperating after their game (the score was Ulster 33 – Zebre 13!).

On arriving on Friday evening we had the opportunity to sit down together for a meal and a chat. This is such an important part of any conference weekend or event. Sitting down with other families and chatting about your experiences can often be just as valuable as hearing from the invited speakers. Many of our families comment that it is in these social parts of the conference that they do the most learning and take away the most valuable hints and tips for daily living.

On Saturday we had a packed programme! While the adults heard from a range of local experts, the kids split into two groups. A few of the siblings who attended went for a fantastic day out at W5 (an educational exhibition in Belfast), while the other kids enjoyed a day of sensory experiences led by Mary Connor, a Play Therapist from a local company called Sensory Kids ([www.sensorykids.co.uk](http://www.sensorykids.co.uk)). I would encourage any of you who have children who enjoy sensory play to seek out local sensory play services – they are an excellent resource and a great idea for birthday parties that

your child can take part in and really enjoy.

Our conference programme was packed with updates about the services that MPS and Fabry families have access to in Northern Ireland. We had sessions on pain management, cardiology, orthopaedics, respite, hospice care and lots of other 'hot topics'. It was a fantastic day of learning for both families and professionals. I want to extend a special thank you to our family speakers Steven Shannon, Aidan Kearney, Debs Boyle and Sandra Bates – it was great to hear from each of you.

On Sunday morning we finished off with a relaxed and interactive session on the sensory dimensions of care. This session was led by a Paediatric Occupational Therapist who has a special interest in sensory care. We were delighted that both the children and adults who attended this session had the opportunity to try out some sensory toys and equipment. I have heard that lots of the families who attended have now included some sensory care into their daily routines. Come along to our National Conference in June 2015 to learn more about the sensory dimensions of care.

All that's left is to say a big thank you to all the families and professionals who made our conference possible. We look forward to welcoming you all back next time!

Alison Wilson  
Advocacy Support Officer

*(pictures, left to right: Alison and Jamie (MPSI Hurler) passing the time while mum catches up with friends; the Hilton, Templepatrick; Naomi, Kathleen and Hannah (MPSIII) enjoying their time at the conference)*

## MPS Society Conference - 26th - 28th June 2015

**The MPS Society are holding a weekend conference, spanning from Friday 26th to Sunday 28th June 2015, that is dedicated to the sharing of information on Mucopolysaccharidoses, Fabry and related diseases.**

**Entitled 'Conference on Paediatric and Adult Perspectives in Management and Treatment of Mucopolysaccharide, Fabry and Related Diseases', this weekend will prove extremely informative for both**

**families and professionals alike.**

**We have included Conference Programme and Booking Form along with this magazine which will provide you with all the relevant information on such things as speakers, child care and costs.**

**The last chance to book your place is Friday 15th May 2015, but booking well in advance is advised.**

# Lapland UK Family Day

It was a chilly start to our Lapland adventure on Wednesday 3rd December 2014, though while we were all happily distracted by the waft of coffee, cakes and sausage rolls, we were even more distracted by the anticipation of meeting Father Christmas! The Elves too gave a warm welcome to everyone, teaching the silly Elven wave and skipping along happily.

We were soon taken through to the Forest of the Elves where we had to recite the magic rhyme so that we could go through the magic door to Lapland. There was also a competition to see which group could make the loudest animal noise either Reindeer or Husky, clearly the huskies won... but have you tried to make a Reindeer noise?!



Once through the magic door we entered a snowy white world filled with pine trees and log cabins, we followed the path to the toy factory. Before we knew it a call had come in from Father Christmas asking for our help as he was running out of toys: all the children of the world had been particularly good this year! Each child made a cuddly reindeer and a wooden xylophone and helped to load them through the hatch and on to the sleigh.



Next we continued to Mother Christmas's kitchen where we learned of Father Christmas's love of Gingerbread and of Mother Christmas's concerns over his weight and ability to fit on the sleigh. All the children enjoyed decorating a gingerbread man and story time with Mother Christmas before heading to the Elven village.

At the village there was an opportunity to grab a bite to eat, spend some "Jingles" (Elven currency) in the gift shops and everyone particularly enjoyed the ice skating. Rudolf came to see us and we also got to meet the huskies. Soon it was time to visit Father Christmas.



We ventured deep into the wood and arrived at a little log cabin and we knew that Father Christmas must be waiting for us inside. As we went in we saw a huge bag of gifts alongside which Father Christmas was sat with "The Good List" in hand. Father Christmas knew about each child and their favourite things but most of all he knew who had been good! Each child sat in anticipation as Father Christmas thumbed his list looking for names; thankfully everyone had been at least good enough!



We would like to thank BBC Children in Need for giving our members the opportunity to enjoy this special day.

Steve Cotterell  
Advocacy Support Officer

"We would all like to say a huge thank you for allowing us to join you all in Lapland UK. From start to finish we all thoroughly enjoyed the magic of the experience we had there. To see Ellie and Ethan's faces light up at every part of the day's journey touched mine and Simon's hearts throughout." - The Greening family



"Thankyou to all at the MPS Society who made yesterday's trip to Lapland UK possible. After a pretty tough year it was lovely to have such a magical family day out. We can't thank you enough" - Louise Etches, via facebook

If any of the families who attended Lapland UK would like any of the photos featured here to be emailed to them, please give us a call or email [magazine@mpssociety.org.uk](mailto:magazine@mpssociety.org.uk)



## 40th Annual National MPS Society Family Conference

I have just returned, quite exhausted, from attending the 28th Annual National MPS Society Family Conference at Disney's Contemporary Resort in Florida with Christine Lavery. Whilst I've always known that Christine works extremely hard, I now know that I had no real idea of her workload, the demands on her time (everyone wants to speak to her!) and her ability to keep all the balls in the air!

Four families came out from the UK for the conference and our first job was to welcome them and make sure they were settled into the hotel after the long and tiring flight, to distribute their Disney passes and deal with any queries.

Prior to the conference beginning, we spent a long and hectic day in Miami carrying out a risk assessment of hotels and meeting Dr. Mislen Bauer at Miami Children's Hospital in preparation for forthcoming clinical trials, which will be overseen by MPS Commercial, and I was very impressed with the attention to detail involved in such assessments. First, on landing at Miami airport we investigated travelling by the metro train line to the hotels, much to the amusement of Dr. Bauer and her assistant Yasameen Kharazmi who told us no-one in Miami travels by train....well, we did and it was very accessible and straightforward. Next, to the hotels where we met with staff, inspected rooms, checked for accessibility, facilities and location to the hospital - no detail was missed. Then the next step was our visit to the hospital for our meeting with Dr. Bauer and I learned how important it is that Christine has face-to-face meetings, rather than communicating via email or phone, to explain how the Society will support the families on the trial and to build relationships with both the clinicians and the families.

In the midst of the hotel assessments, Christine was in constant communication with MPS House because of the publicity around developments in the urgent campaign to secure NHS funding for the newly licensed Vimizim for MPS IV patients and even carried out a press interview via mobile phone from a hotel lobby....see what I mean about keeping all the balls in the air!

Dr. Bauer was so keen to speak to us and to show us the facilities at her hospital, of which she is rightly proud, that it almost resulted in us missing our return flight...10 minutes after we were due to board the plane, we were still in the taxi trying to get to the airport! We must have been quite a sight sprinting through security and skidding up at the gate whilst our names were being called by exasperated ground staff! We'd hardly taken our seats before the plane took off but we were very relieved to be heading back to our hotel.

The next morning we had a breakfast meeting with Scott Riccio from Synageva when Christine was able to update him on current projects and discuss ways in which the Society and Synageva might co-operate in the future. I was very pleased to discover that a breakfast meeting really did include breakfast and wasn't just so-called because of the early hour!

The conference sessions were presented by a mix of clinicians, families and pharma companies and all were interesting and informative. We were welcomed by Steve Holland, President of the US Society and this was followed by a comprehensive overview of MPS diseases and potential new therapies by Dr. Jo Muenzer. The following sessions covered all aspects of MPS including management of diseases, palliative care, research updates, fund raising, a sibling workshop and finally updates from the pharma companies.

The conference concluded with the Awards Banquet and Prom which we all attended and, during which, Christine made presentations on behalf of our Society to Steve Holland who was retiring as President of the US Society, and to Barbara Wedehase who is retiring from her post of Executive Director, in recognition of many years of collaboration between our respective societies. We were told there was to be a special surprise after the dinner and before the disco began - imagine everyone's delight to find Mickey and Minnie Mouse, Donald Duck, Pluto, Cinderella, Chip n Dale and Goofy had come to join in the fun and pose for photos - the children were, of course, thrilled....and so were the adults!

I am so fortunate to have had this amazing experience.

*Judith Evans,  
Trustee*

# International



## News from Austria - Maria's Vimizim Story

*This article was originally written to appear in the Austrian MPS-Newsletter and has been translated for the English Magazine. Maria has MPSIVA Morquio, is 21 years old, has got five siblings and lives in Austria.*

Last year I wrote about our trip to a high rope course during the Austrian MPS Therapy Week, where I was climbing and jumping from tree to tree at a height of about 10 meters. I also told you about different studies I was part of. But today I want to tell you something much more amazing:

Vimizim was licenced in Austria in May 2014. Now, what does this mean for me? I was able to leave the study centre in Mainz where I had received the ERT for 15 months (after having received it in London for 18 months) and changed the hospital to Vienna, where I had my first infusion on June 1st. They had offered to do my treatment for 12 weeks and I was happy for that, because it would give us enough time to arrange ERT in my home town in Upper Austria. Even though Vienna was still a full day's programme, at least I didn't have to stay over-night. Thanks to my doctor I was able to take one of the infusions even to Brazil to the MPS World Congress, where they did a really great job with infusing me!

In the meantime we had applied for reimbursement with my health insurance, and I could hardly believe my eyes when they immediately agreed. My plan would have been only to change the hospital to the one in my home town as a first step. I was looking forward to this relief already as I planned to have my treatment after work. It's quite easy for me to get around in my electrical wheelchair in town and the hospital is not very far from my office.

But, everything happened differently and I am so glad to tell you that after three years of travelling to London, Mainz and Vienna, I am just about to get my very first home treatment while I am typing this article!

It's a little bit weird for me to know that Vimizim is flowing through my veins right now and I am not anywhere else but sitting in front of my laptop in my room. During the past three years I have travelled almost 300,000 miles, and this was only for flying to my infusions. I received the Golden Senator card for miles and more, and a lot of airport staff

knew me personally, even in London, Heathrow. In Linz they even started to check me in when they realised I had entered the airport. Today, almost exactly three years after my very first flight to the Royal Free Hospital in London, I am enjoying my first home infusion!

### How a home treatment day works for me

Today, my nurses Nicole and Franz rang the doorbell around half past eight. Mum had fun receiving them on a red carpet leading into my room. They prepared everything for the infusion; Franz inspected my veins and was happy to find good ones immediately. Everything went well and at 9.10am the infusion was ready to go.

We had to do some paperwork this first time, first assessment papers and signing for my own pump and the bag to carry the infusion around. While managing everything we had delicious apple pie and coffee. I was sitting in our living room and we were chatting about this and that. After a while we decided to walk our dogs – I enjoyed my new freedom during the infusion a lot!

From November on Nicole won't be here anymore, but Franz will stay with me (hopefully forever). We are getting along very well and it's like a friend would be visiting me. The only difference is that he would check my blood pressure and my arms now and again. Isn't that crazy? I don't have to fly, I don't have to stay anywhere overnight. The only thing I have to do is to wake up in time, open the door for Franz, hold my arm towards him, let him put up the IV and then I can even walk my dogs in our little village.

### Thanks

My special thanks go to my wonderful family. Thank you mum for your "total YES" when I was offered the chance to take part in the clinical trial in London. Thank you for considering the "how to do it" only in the second step.

Vimizim is the best that could have ever happened to me. I feel so much better than before, even though I was not really aware how miserable I was. I was used to how it was, but now so much has changed! Thank you for these three years that I was able to win before the drug was licensed just because of your arrangement and your support during the trial. Although travelling was very exhausting and pretty nerve-racking and it was hard for you to keep the family and your MPS work running while you were with me, it was definitely worth it.

Many thanks also to everyone accompanying me from time to time to my weekly infusions, to give my mum a little break to spend time with the rest of our family and attend meetings. Thank you also to my stepfather Martin as well as to my sister Anna and to my Granny for holding the fort at home with my younger siblings.

I am also very thankful for the teams in London (Alan, Alison, Anna, Cecil, Dr. Hughes and of course Keith, who did not even forget me once), in Mainz and in Vienna. And a very big THANK YOU to Biomarin for developing this amazing treatment. Even though we Morquios are only a small part of the big MPS family – I would like to tell you on behalf of all of us: You are awesome!

### The Future

It's been more than a year already that I have got a job in the electric supply company, where I am working in the office. My contract was changed to an unlimited employment last month and I really like being there. It makes me proud to hear that I am doing a good job and I love the feeling to be needed. I couldn't imagine anything better. Who knows – maybe I will receive my treatment while I am working one day?

The next big step in my life is to try making my dream of a driving licence to become true. I already had the chance to drive a Mercedes Sprinter with Joystick at a trade fair. I guess my obvious interest in the cars was the reason for an invitation to a counselling interview with Paravan Germany, the leading company for car adaption. I will be allowed to do a test drive over there and I can't wait. The problem is how to manage the funding of the car adaption. If it works, I will be able to drive on my own and won't be that depended on my family. Let's see...

*Maria Prähofer*

## Have you got an Emergency Patient Information card?

### What are Emergency Patient Information Cards?

The Society has developed, with the agreement of all specialist centres, Emergency Patient Information Cards. The card is the size of a credit card and includes relevant details about the patient, the condition that they have and contact details of their specialist centre. Included also is a link to the MPS Society.

The card has been designed so that it can be carried easily and, if required, could be presented to medical staff in an emergency, giving accurate information about your condition and who to contact for information.

### How can I get a card?

If you are a member of the MPS Society and you would like a card for you and/or your child, please download and complete the form found on our website under the Downloads & Resources section.

You can also request a form by email at [j.goodman@mpssociety.org.uk](mailto:j.goodman@mpssociety.org.uk), or by calling us on 0845 389 9901

Additional cards are available but may incur a small charge; please contact us for more information.

If you are not a member of the MPS Society and are interested in talking to someone about this then please get in touch



## Aidan's Austrian MPS Weekend

My name is Aidan Kearney. I suffer with MPSIV Morquio. I was on an enzyme replacement therapy for Morquio adults as part of a clinical trial, where I flew to London every week for the treatment. I met some amazing people, including Maria Prähofer and her mum Michaela Weigl.

They live in Austria in a town called Wels. They had to take two flights to London to receive ERT. I also met them and their family and when I was in Brazil as an MPS ambassador, and they asked me if I wanted the opportunity to attend an Austrian MPS adult weekend - of course I jumped at the chance to meet other Morquio sufferers around Europe.

So I booked my flight and was on my way doing the same journey Maria and her mum had to do for nineteen months to receive ERT in London: two flights, a bus, train and short car journey. As Maria's mum said, she would travel anywhere, any place to get a better quality of life for Maria, which she says Maria now has thanks to ERT (Vimizim).

Michaela, Maria's mum, runs the Austrian MPS Society from her house. She has two daughters, one with MPS IV, and three sons. Michaela never stops; she makes jams, drinks, table decorations, cards and bags of dried apples all from her own garden. All these are then sold at markets to help fund the Austrian MPS Society. She only has one person working for the organisation in the office below the house.

The adult weekend started on the Friday evening and there was a buffet and drinks, which was a great chance to meet the other attendees, most of which were Morquio sufferers. There were also two brothers, both with MPSII Hunter, and a 60 year old woman who had MPSVI Maroteaux Lamy, and had only been diagnosed this year in October.

The next day there was a day trip planned to Haubis, a famous family run bakery where they make and sell 1.2 million bread rolls per day and that does not include their other breads, ginger bread and cakes. We were taken on a tour of the bakery and factory where we made our own traditional Austrian bread.

The evening had come and it was time for dinner and drinks (also known as networking). Talking to the Austrian MPS sufferers, I was really interested to find out that they did not take anything for pain relief; only one person said that if they knew that they would be doing a lot of walking that day then would take a couple of paracetamol to ease the pain. In the winter they did struggle with stiffness but on the whole they were very happy and enjoyed life as much as possible. I could tell this as we had a laugh and drink that night and I really enjoyed myself.

All too soon it was time to say goodbye. I had a brilliant time and met so many amazing people and learned a lot from this experience.

I learned that one person can make a difference from Maria's mum, Michaela; she is a mum who juggles family, a daughter with MPS, and runs the Austrian MPS Society all from her house with only one employed worker. It reminds me of how the MPS Society in the UK started and where it is now, how one person can make the difference and to never give up when the going gets tough.

*Aidan Kearney*



## ASTERIX Patient Think Tank

26 - 27 October 2014  
Utrecht, Netherlands

An estimated 30 million European patients suffer from about 6000 known rare diseases, many of them chronic, potentially very disabling and typically affecting children. Since the year 2000, more than 850 orphan drug designations have been granted by the European Commission, but less than 100 orphan drugs have reached the market. There is a substantial unmet need for drug treatment for rare diseases and if this is to be met there are going to be many more clinical trials in small populations like the MPS diseases.

ASTERIX is a EU funded research project focusing on the development of more efficient and effective research designs to study new drugs and treatments for rare diseases. The overall aim is to stimulate the search for treatments for these devastating and largely ignored diseases. ASTERIX is specifically designed to optimise methodology for clinical trials in small populations to achieve more reliable and cost efficient clinical development of treatments for rare diseases. Inclusion of patient level information and perspectives in design and decision making throughout the clinical trial process are important objectives of the ASTERIX project.

I am very honoured to have been invited onto the ASTERIX Patient Think Tank which will be active throughout the entire project, and will be consulted with regard to knowledge translation and dissemination of the methodologies that will be developed during the ASTERIX project. The ASTERIX Patient Think Tank will provide advice to the work streams ensuring that the patient perspective is embedded into the project's deliverables.



one session was on the patient perspective on adaptive design methods in clinical trials that might better serve the rare disease patient population. Another session involved exploring how to involve patients in the weighing of outcome measures. The ASTERIX team presented a template, the POWER MODEL, as a flow chart of how patient organisations might consult with patients or their caregivers, prioritise outcome measures and reach a consensus. There was much discussion and other ideas came to the surface that the ASTERIX Patient Think Tank asked the ASTERIX team to consider.

This is an important initiative that allows MPS to share both its considerable clinical trial experience and learn from new thinking on clinical trial design and weighting of outcome measures.

*Christine Lavery  
Chief Executive*



## Yann Le Cam of EURORDIS becomes Special Advisor to the BioPontis Alliance for Rare Diseases

December 17th, 2014

Raleigh North Carolina, USA and Brussels, Belgium

The Board of Directors at the BioPontis Alliance for Rare Diseases is pleased to announce the appointment of Yann Le Cam, Chief Executive Officer of EURORDIS, as Special Adviser. The appointment of Yann Le Cam as a volunteer senior advisor to BioPontis is part of a broader collaboration between EURORDIS (The European Organisation for Rare Diseases) and BioPontis Alliance, to help build bridges with patient groups and other stakeholders who share a mission to advance more therapy development for rare diseases.

Erik Tambuyzer Ph.D., Board Chair for BioPontis Alliance, states “We are very honored that Yann, as one of the world’s most respected leaders and advocates for patients with rare diseases, will support our initiative. As a founder and past Chair of the European industry Task Force on Rare Diseases and Orphan Medicines, I worked together with Yann and with Eurordis towards the approval and implementation of the EU orphan medicinal products regulation. Yann brings high level strategic guidance to the launch and mission of BioPontis. We look forward to a strong collaboration with EURORDIS and its research-driven member patients’ organizations.

Partnerships between patients’ organizations, their philanthropic support, and the biopharmaceutical industry are crucial for bringing treatments to the 95% of rare disease patients currently without treatment options.”

Yann Le Cam served as Vice Chairman of the Committee for Orphan Medicinal Products (COMP) at the European Drug Agency (EMA). As one of its first patient representative members, and working with the European Commission, he was instrumental in helping to design and in advocating many of the strategic initiatives related to rare diseases in Europe and internationally. Yann also served on the Management Board and Executive Committee of the French Health Technology Assessment agency and on the DIA Advisory Committee Europe. He was Vice Chairman of the EU Committee of Experts on Rare Diseases (EUCERD), and is nominated to the current European Commission Experts Group on Rare Diseases. Yann Le Cam is elected Chair of the Therapies Scientific Committee of the IRDiRC – the International Rare Diseases Research Consortium ([www.irdirc.org](http://www.irdirc.org)).

“I have had the pleasure of working with Yann Le Cam over many years in a common effort to globalize orphan drug regulations. His outstanding leadership in the community of rare disease patients, coupled with his

involvement in supporting rare disease research and international regulatory systems, make him an ideal advisor to our board and management” echoed Marlene Haffner M.D., MPH. and Vice Chair of the Board. Dr. Haffner was Director of the Office of Orphan Drug Product Development at FDA for 22 years.

Yann Le Cam stated “The unique approach taken by BioPontis Alliance is a welcome opportunity for patients’ organizations. They can partner in advancing research toward possible treatments with a fellow not-for-profit organization – yet one that brings the critical expertise to develop therapies and in regulatory planning, and in line with the standards needed by of the biopharmaceutical industry.” He further commented, “The benefit of collaboration between the nonprofit BioPontis Alliance for Rare Diseases with patients’ organizations is that along with BioPontis Alliance’s competencies and abilities to manage drug and intellectual property development, BioPontis is a not- for- profit with a structure to harness financial resources from both philanthropy and the for profit private sector, adding to the strengths of the patients’ organizations and their academic partners. This will form a novel and powerful development machine, hitherto non-existent in the field. ”



### About the BioPontis Alliance for Rare Diseases:

BioPontis Alliance for Rare Diseases, based in North Carolina, USA and being founded in Europe, is an international nonprofit organization whose mission is to advance promising science into treatment candidates, by partnering with patients’ organizations, academic researchers and industry - for the millions of rare disease patients who await a first attempt at treatment. More information on BioPontis Alliance for Rare Diseases is available at [www.biopontisalliance.org](http://www.biopontisalliance.org).

### About EURORDIS

EURORDIS is a leading International Non-Governmental Organization (INGO) and is recognized as the largest European Patients’ Organization in the field of rare diseases.

EURORDIS represents more than 600 rare disease organizations in 60 different countries, covering more than 4,000 distinct rare diseases and is the voice of an estimated 30 million patients affected by rare diseases throughout Europe. More information on EURORDIS is available at [www.eurordis.org](http://www.eurordis.org).

### Contacts:

*Barbara Handelin, PhD, CEO  
BioPontis Alliance for Rare Diseases  
[Barbara.Handelin@biopontisalliance.org](mailto:Barbara.Handelin@biopontisalliance.org)  
(+1) 610 636 2332*

*In Europe:  
Erik Tambuyzer, PhD, Chairperson  
BioPontis Alliance for Rare Diseases  
[Erik.Tambuyzer@biopontisalliance.org](mailto:Erik.Tambuyzer@biopontisalliance.org)  
+32 475 61 57 11*

## Fabry Disease

### Amicus and EMA Conduct Successful Pre-Submission Meeting

We are pleased to inform you that earlier this week Amicus conducted a successful pre-submission meeting with the European Medicines Agency (EMA) to discuss the registration of migalastat monotherapy for the treatment of Fabry disease in individuals with amenable mutations. This positive exchange keeps us on track for submission of a marketing authorization application (MAA) to the EMA in the middle of 2015.

*Jayne Gershkowitz  
Vice President  
Patient & Professional Advocacy  
and Public Policy  
Amicus Therapeutics*

## Abeona Therapeutics to Develop Therapies for Children Suffering from Sanfilippo Disease

On 1 December 2014 Biotechnology start-up Abeona Therapeutics announced it closed \$3.6 million to complete preclinical development of therapies for children with MPSIII, Sanfilippo Disease.

This funding will help support Abeona’s global development of gene therapies for treating patients with Sanfilippo disease type A and B working in collaboration with Nationwide Children’s Hospital in Columbia, Ohio.

Time is a critical factor for children with Sanfilippo disease as 70% of affected children will not reach their 18th birthday. “We are poised to initiate clinical trials in early to mid 2015 for these much needed therapies,” said Kevin Flanigan, Principal Investigator at the Centre for Gene Therapy at Nationwide Children’s Hospital.



## The Fight for Vimizim



*It has been really difficult to write an update for the Vimizim Campaign as things have been changing on a daily basis, so no doubt by the time you read this things will have progressed (hopefully). The best way to keep up to date is through our website, Twitter and Facebook pages, and for those directly affected, we will update you directly. It has been a busy few months pushing for funding approval of Vimizim.*

As many of you know, following five years of clinical trials, which involved 43 of our members with MPS IVA Morquio from across the UK, the treatment Elosulfase Alfa, known as Vimizim, received marketing approval from the European Medical Agency in April 2014.

However, despite this approval and the UK having the largest percentage of participants involved in the clinical trial we are behind other European countries in having funding for the treatment approved. Other countries which have already had the drug approved for reimbursement include France, Germany and Turkey to name just three of 22 countries across the world.

BioMarin are continuing to fund the treatment compassionately for the clinical trial patients, but there is no certainty how many more months this will continue. NICE are assessing the drug next year but we are not expecting a decision from them until December 2015 and there is no guarantee that BioMarin will continue to keep funding the drug compassionately until this date.

It is extremely frustrating that this drawn-out decision making process is preventing individuals from starting treatment at home. Then there are other affected individuals to consider: the children diagnosed with MPSIVA since the clinical trial closed and the young adults who did not get on to the clinical trial, who have not had a whiff of Elosulfase Alfa.

This fight is political; NHS England had a flawed process for making funding decisions for ultra-rare diseases. We set up a petition 'NHS ENGLAND'S SCORECARD DENIES ACCESS TO

TREATMENT FOR ULTRA RARE DISEASES' on 38 degrees and have attracted over 8000 signatures. If you haven't signed it please find it here and share it with anyone you know: <https://you.38degrees.org.uk/petitions/nhs-england-scorecard-discriminates-against-treating-ultra-rare-diseases>

The MPS Society supported one of our members to engage with our lawyers Leigh Day & Co. A letter of intent to go to judicial review was sent to Jeremy Hunt, Secretary of State for Health, giving him seven days to respond. NHS England's lawyers came back to us at the last minute acknowledging the process was unlawful and postponing their CPAG decision making meeting. This was both a positive and negative outcome. It means that Vimizim will now be assessed fairly, once NHS England have worked out how to assess treatment,s but it is equally hugely disappointing to those not on compassionate treatment who will have to wait longer until a decision is made unless we can maintain the intensity of the campaign to get an urgent interim decision.

Individuals should not be permanently disadvantaged due to individuals at NHS England not being able to develop a fair decision making process for their pot of money.

More momentum has been gathered thanks to Katy and Simon Brown's meeting with their MP Greg Mulholland. Greg has now raised two questions in the House of Commons both to George Freeman, Minister for Life Sciences at the Department of Health and the Department of Business, Innovation and Skills (BIS) and Prime Minister David Cameron, which led to our meeting with George Freeman on the 9th December.

The meeting on the 9th December at Portcullis House, opposite the House of Parliament was attended by the Brown's, Suzanne Mallah whose son Kamal headed the legal challenge as well as a family whose son has Duchenne Muscular Dystrophy. The Duchenne drug, Translana, is the other Ultra Orphan drug to be caught up in this delay. Together we hope we could add more weight to the argument. The outcome of this meeting is that George Freeman will meet with Greg Mulholland, the pharmaceutical companies for both treatments as well as representatives

from the two Societies next week.

Turning to Social Media, the Brown family have also started the Twitter and Facebook campaign 'Keep Sam Smiling', which now has well over 1400 likes and features much of the media coverage.

Gracie Mellalieu has been the star of the show in the North West of North Wales - turning on the lights in her home town Mold; she has also been front-page news and met with her MP Mark Tami. Coronation Street stars Cherylee Houston and Ian Puleston-Davie have filmed a little film clip which you can see on YouTube: #KeepGracieSmiling

Due to the rarity of MPS diseases it is imperative that we shout loudly in order to raise the public profile about the treatment and also the debacle of the decision making process taking so long, and the threat of disadvantaging those not on treatment or facing the loss of treatment in the future if there is no reimbursement in England. As part of the decision making process we have submitted patient testimonies to NHS England and thank you to everyone who wrote to us.

Through your hard work over 40 Members of Parliament have been written to by members and their friends and families about Vimizim and the situation, but we need all MPs to know what is going on and what this means to you. If you haven't met with or heard from your MP please chase them. There is a General Election in May and they need your votes. There has also been an incredible push for press coverage and raising awareness in the media but we need more national press. Sam and his family have appeared on most TV and radio stations in the Yorkshire area and been joined by our Chief Executive, Christine Lavery.

At the MPS Society we have attended meetings with NHS

**Greg Mulholland (Leeds North West) (LD):** Following the threat of a legal challenge, NHS England has scrapped its processes for approving drugs for rare conditions, which is affecting 200 children in the country, including six-year-old Sam, in my constituency, who has Morquio. Will the Prime Minister today instruct the Health Secretary

**3 Dec 2014 : Column 301**

to re-establish the highly specialised commissioning service so that we can approve these drugs and ensure that children get the drugs they need immediately, before a new process is put in place?

**The Prime Minister:** I am happy to discuss that issue with the Health Secretary. As the hon. Gentleman knows, we have a procedure for licensing drugs and, for cancer drugs, we have the additional benefits of the cancer drugs fund, but I am happy to discuss his point with the Health Secretary.

**Greg Mulholland (Leeds North West) (LD):** My constituent, six-year-old Sam Brown, is one of 100 people with the rare disease Morquio. His family live in a state of anxiety because they do not know whether the drug Vimizim will be approved for further use on 15 December. Will a Minister please meet me and Katy and Simon, Sam's parents, to give Sam the Christmas present he needs and to keep Sam smiling?

**The Parliamentary Under-Secretary of State for Health (George Freeman):** I would be delighted to meet my hon. Friend and his constituents to review that very important issue.

*Extracts from Parliamentary sessions concerning Vimizim*



Left to right: Christine Lavery, Katy Brown, Simon Brown, Suzanne Mallah

England and NICE, breakfast meetings with MPs and are staying up to date with the changing political situation. NICE Topic Selection – Stakeholder Information Event

Due to devolved health care Scotland, Wales and Northern Ireland make reimbursement decisions separately. Aiden Kearney and Alison Wilson have been meeting with Iain Paisley Junior and are pushing media coverage for an early decision in the Province.

We know that more journalists have covered the story not recorded here, but we just wanted to say thank you for all the hard work up to now. However, we need to make much more noise to the MPs and government. Despite so much media coverage only a few families are making a noise and we need to demonstrate to NHS England that the treatment is vital for our whole MPSIVA membership.

There is so much uncertainty, we cannot rest on our laurels and assume that the drug will be reimbursed. There is compassionate use for now for some, but we honestly don't know how long this will go on for. It is not a solution for those who want access to the treatment long term. For every week that goes by, individuals not on treatment are deteriorating. It has been eight months since the drug was licensed and the waiting began.

At the MPS Society we can help provide information for the press and are happy to speak to journalists. However, often journalists like a local connection in order to cover the story and that is why we need you to help contact your local newspaper or radio station.

We need to do all we can to raise awareness of the issue so that we know we have done all we can to push for an interim policy and early reimbursement for Vimizim.

Thank you again to everyone who has written letters, shared the petition, contacted the press and utilised contacts. If you would like treatment and have not been in contact with the MPS Society please contact Charlotte on 0845 389 9901 or email [c.roberts@mpsociety.org.uk](mailto:c.roberts@mpsociety.org.uk) and I look forward to hopefully sharing some positive news with you soon.

Charlotte Roberts  
Communication Officer



Left to right: Gracie and Cherylee Houston; Gracie, Cherylee & Ian; Gracie and Sarah Harding

## Gracie's Adventures in Media City

### The Fight for Vimizim

Thursday 27th November 2014 is a day that Gracie and I will never forget! We are blessed to have friends in the media, who star in Coronation Street, so called upon them to help us with raising awareness of the ERT drug, Vimizim, that has been licensed in the UK for Morquio Disease but not yet funded. Recently, Gracie's story has been in the local press, where we have been urging people to write to their MPs about the drug and why it needs funding. We thought we needed to step the campaign up a bit, so when we contacted Ian Puleston-Davies (Owen the builder), and Cherylee Houston (Izzy Armstrong), and they agreed to do all they could, we were thrilled.

The plan was to take some photographs to send to the national press, which we did.... but the whole day completely snowballed! Gracie's Grandad Chris is a keen photographer who works near the Corrie set in Media City. He and Ian quickly decided that maybe a funny video would have more impact, so we were soon running around Media City, having a great time together, and compiling a short film! We even had real paparazzi there from the national press, who just happened to be in the same place as us, at the right time! Gracie thoroughly enjoyed whizzing around in her power chair, alongside

Cherylee, who also uses one. It was the only time Gracie has been able to 'run around' with someone, and it must have been a lovely feeling for her.

After our filming, we made our way for a very quick tour of the new Corrie set, followed by lunch in the ITV café, where Gracie couldn't believe her luck: Sarah Harding from Girls Aloud (Gracie's favourite band!) was in there enjoying a snack! She was lovely, and allowed us to take some photos of her with Gracie. She even sang a bit of 'Jump' to Gracie, as Gracie said that was her fave song!

It truly was an amazing day, where we felt like we had really made a difference to the Vimizim campaign - in fact, Gracie and I were buzzing all the way back home to North Wales! The ITV peeps have said that when the video is ready, they will tweet it with the petition, and Cherylee sent Sarah Harding a Tweet after we met her - which Sarah has then retweeted! We really hope that this will make a big difference, to Keep Gracie Smiling!

Yvette Mellalieu

Check out Gracie's video on YouTube (#KeepGracieSmiling Vimizim)

Here are some ways that you can keep up to date with the latest news on Vimizim:

- Take a look at our New & Events section of our website - <http://www.mpssociety.org.uk/news-events/>
- Like our facebook page - [www.facebook.com/mpssociety](http://www.facebook.com/mpssociety)
- Follow us on twitter - [www.twitter.com/MPSSocietyUK](http://www.twitter.com/MPSSocietyUK)
- Like and follow Katy Brown (#KeepSamSmiling) and Yvette Mellalieu (#KeepGracieSmiling) - and why not start your own page to raise awareness and get more signatures on that petition and more people talking to their MPs

TOGETHER WE CAN MAKE A DIFFERENCE



Healthcare at Home

Healthcare at Home  
Patient Organisation Workshop



News from Macedonia

Healthcare at Home held a patient organisation workshop at their headquarters in Burton-on-Trent to discuss the changes they have implemented and the progress they have made in communication and drug distribution this year. MPS, Gaucher, HIV, Ankylosing Spondylitis and Pulmonary Fibrosis representatives attended the meeting with Natalie Douglas the CEO of Healthcare at Home along with other members of their team.

Healthcare at Home acknowledged the difficulties they have faced this year in terms of delivering drugs on time and explained that this was due to a catalogue of events which included a shortage of a drug, new logistics being implemented and the merging of their warehouse facilities.

Natalie Douglas joined Healthcare at Home in June 2014 and has set up a taskforce recovery team and Healthcare at Home made immediate plans to improve patient experience and service. The aim of the meeting was to discuss patient issues and how from a patient's perspective solutions could be achieved.

Healthcare at Home have already made changes which they have seen the benefit of in terms of providing service to patients. They have re-organised their warehouse and dispensary to improve efficiency, including a computerised system to track orders. This is extended to the delivery service as well. There is now a rescheduling team to deal with any changes to

delivery and drivers have received training.

They have set up a new Communications Team so that information can be disseminated and we offered suggestions as to how information could reach our members easily. Healthcare at Home recognise that they treat many conditions and they are working on gathering information from patient surveys and support organisations to understand each condition more thoroughly. Furthermore the call centre hours have been extended from 8am to 8pm weekdays and the centre is also open at weekends.

Richard Greatorex, Marketing Director at Healthcare at Home said:

"I would like to thank the five patient group representatives for attending the meeting. The insights we gained are going to ensure Healthcare at Home is able to develop an engagement programme that truly resonates with our patients. We will be working much more closely with all patient groups as we move forward."

Healthcare at Home would like to hold further meetings to share information with patient organisations and are also considering having a patient council to represent patient views. If you have any comments regarding this please contact at [d.cavell@mpssociety.org.uk](mailto:d.cavell@mpssociety.org.uk).

Debbie Cavell  
Advocacy Support Officer

Macedonia's Association of Citizens for Rare Disease, 'Life with Challenges', has recently informed us that their Prime Minister has announced that the government voted on a new regulation for financing of the program for rare diseases.

Chairman of Life with Challenges, Vesna Aleksovska, reported the following:

"Next year for every packet of cigarettes, 1 MKD (Macedonian currency) is going to the rare disease program, which at the end is around 4 million EUR. At the moment the program has 3 patients and 1 million EUR, so with this additional financing we estimate that around 100 patients will get the much needed treatment (as for HAE, Pulmonary Hypertension, Gaucher disease, MPS IV, Wilson Disease, and others that have existent medicine available in the world but not in Macedonia until now).

This is like a Christmas present for all patients that were waiting for treatment.

The Minister of Health called us and said that we will have an additional meeting to agree on administration for treatment, to see if it is possible for patient representatives to be part of the commission for rare diseases and to negotiate further about adopting a national strategy for rare diseases that we suggested this year (as a draft program)"

Vesna maintains that although they still have a long way to go before they can celebrate, "the start is good and we are going step by step".



Husband and wife fundraisers, Jeff and Suzy Elliott relaxing after finishing the Great South Run. The couple raised an incredible £6,520.02

- Read more on page 42



## Bramley to Paris Cycle

Louis Garthwaite (MPSII Hunter), David Wolff, Piers Mosselmans, Rob Platt and Ben Philipp took on this challenge to raise money for the MPS Society.

Read on for Louis's account of their adventures...

It was a tasteful 20°C and sunny on the morning of August 9th when six strapping members of Team RGS assembled on the Downs link by the disused Bramley Station. Their mission: cycle to Paris to raise money for MPS Society, the only UK charity supporting individuals and families affected by progressive, life-limiting genetic conditions.

Each was buzzing about the forthcoming adventure, and even two punctures within the first hour couldn't deflate us! Thankfully, we didn't continue puncturing at this rate and eventually arrived at the coast following a 50-mile slog through forest and rocky track. A particular highlight was the scenic cycle across the craggy cliffs of Mother England, interrupted by a delightful pause at a seaside eatery (where Piers Mosselmans had an issue with his mucus).

Day Two. Wet. Windy. Nippy. Conditions that might have provoked the weak spirited to surrender, but not my brethren of RGS. Waking early, we persevered through these heinous conditions, reaching our ferry punctually (without punctures), timing our crossing to perfection. As Poseidon raged outside, whipping up a frenzy on the Channel, Ben Philipp was keen to show off his sea legs and enacted scenes from 'Titanic' on deck, with Piers filling in for Ms. Winslet ("I'm flying!"). On the other hand, Rob Platt and David Wolff, aware of the need to conserve energy, prepared for the remaining journey with plenty of tea, coffee and countless rounds of the card-classic, 'Cheat.' Fortunately, when we reached Dieppe, the hormonal weather had cleared to welcome us with blue skies. The journey through France had a few challenges. They included a couple of falls, one of which was caused Ben by trying to tweet 'on the go'. We narrowly avoided a multiple pile-up, but he justified his actions by claiming he was trying to drum-up extra support for the group! What a trooper!

Another challenge came on Monday when, three days into the ride, we ended up riding into the dusk with only one headlight between us because we hadn't anticipated quite how stiff and sore our joints would be. Although hills were least of our worries to begin with, by the time we reached Paris, Ms. Kendell was not surprised to find that we'd scaled over 1,600m in aggregate. The total distance that we covered was slightly over 300kms. Rob calculated that, between us, we'd burned an estimated 55,278kcal – or three weeks worth of food in four days!

When we arrived at the Eiffel tower on that tranquil Tuesday afternoon, I really felt that I had achieved the impossible. I had certainly gone to my own limit. This is something not many people without an MPS disability would think of doing lightly. The sense of satisfaction at our achievement was heightened when we discovered that we'd raised more than £8,000 in total for the MPS Society - a really significant sum that will make a material difference and help make life a little easier for some of those whose lives are so affected by these diseases. I am one of the very lucky ones – the Society's work enabled the development of the enzyme replacement therapy that I receive (one of the most expensive drugs in the world). I can honestly say that without the MPS Society, I would not be here today.

On behalf of Team RGS and the MPS Society, I would like to thank everyone for supporting our charity cycle to Paris. Particular thanks are due to Ms. Kendell, without whom the trip would not have come to fruition, and the MPS Society, whose help over the years has enabled me to give something back.

Louis Garthwaite

## Fundraising

Welcome to the fundraising section of our Winter 2014 MPS Magazine. As per usual, we have not been in short supply of fantastic fundraising articles - in fact, if we included a piece for each fundraiser that was held for the MPS Society, our magazine would more resemble a copy of *War and Peace*! As it is, we can just include a snapshot of your fundraising activities, and I hope that you find this section both enjoyable and inspirational.

Thank you to everyone who bought and sold our National Draw tickets; in total we sold an incredible 3060! Flora Croucher, who won 1st prize, wrote in to say "I was absolutely surprised and delighted to have won. Never having won anything like that before, it came as a lovely surprise". Congratulations to all our winners. You can look at the full list of winners on our website.

Our supporters have held a real variety of different events over the winter period - with an unusual amount opting for undertaking head shaves! From ultra-marathons to school discos, our supporters have once again shown their resourcefulness and dedication to our cause, and, as always, we would like to thank everyone who organised, participated in or donated to one of these fundraising events.

If you would like help with your fundraising, or if you would like to submit an article and photos for a future magazine, please drop us an email at [fundraising@mpssociety.org.uk](mailto:fundraising@mpssociety.org.uk) - we would love to hear from you!

Elkie Riches  
Fundraising & Communications Officer



### The Meadows Primary School Raffle and Disco

Emily Bradshaw (MPSI Hurler) and The Meadows Primary School in Staffordshire, raised a total of £76 for the MPS Society from voluntary donations and a raffle. Emily's mum donated some tickets to see Kylie to encourage people to part with their change. The school raised the same amount again, which will be used in school for pupils with additional needs.

Class 1 and Class 2 enjoyed a joint disco outside in the sunshine, and everyone paid £1 to dress up in their best dancing clothes. All the children enjoyed dancing and some of the girls loved doing one of Emily's favourite, the 'Cha-Cha Slide'! To top the fundraiser off, Mrs R. Morris was ecstatic as she won the tickets to see Kylie!

### Bridgnorth Wicked Walkabout

Emma Hiller, mum to Joe (MPSII Hunter), held a fantastically successful Wicked Walkabout on 26th May, and here is what she wrote about her experience:

"Myself and nine other friends went on the 22 mile Bridgnorth Walk around Shropshire, which included a climb up the Brown Clee Hill. Fortunately it was a lovely sunny day and we had great fun taking part with around a thousand other people walking for many different charities. The first of our group made it back in under 5 hours, but the rest of us had too much fun singing and dawdling and took over 7 hours to get back! The people who took part were - Emma Hiller, Marj and Alan Lautman, Louise Hiller, Karen and Steve Haynes, Ann Rochelle, Rialine Benting, Sam Burrows and Brigid Burbridge."

Emma and her friends raised an amazing total of £2066.75. Thank you so much to all involved.

If you would like to hold a Wicked Walkabout, please drop us email at [fundraising@mpssociety.org.uk](mailto:fundraising@mpssociety.org.uk) for more info.

Emma's local football club (AFC Bridgnorth) put on a childrens' football morning in support of Joe, which was followed by food and the infamous "Disco Daddy" for entertainment. Everyone gave up their time for free and the football club supplied the food and venue free of charge. Joe had previously taken part in various sessions at the club and they wanted to do something to help him as they knew he loves football. £205 was raised with over 30 children taking part..

### Charity Fishing Match

My uncle Robert Brogden organised a charity fishing match through his business Orpington Tackle and Baits on Sunday 14th September 2014.

He organised this because my daughter Lily Brooker has MPSIIIA, Sanfilippo syndrome.

We attended the match yesterday which had 15 participants, and Lily enjoyed watching the fish getting weighed to find out who had won the competition.

1st place was taken by Danny Easson with a combined fish weight of 71 pounds, 2nd place was my uncle Robert with 49 pounds and 3rd place was taken by Ray Gage with 43 pound. Lily had pictures taken with the winners and with all that took part.

A wonderful amount of £375 was raised for the MPS Society.

We would just like to say a huge thank you to all that took part in a raising this amount for the charity.

Michelle Brooker

### Sue's MPS Society Tattoo

The 5 year old boy we foster was diagnosed with MPSVI and secondary kyphosis when he was just 6 months old, and from that moment on our lives changed once again with weekly visits to Manchester. I'm please to say that things have now got much easier with weekly infusions which are done in school, but we do still have to go to Manchester as and when.

He is just the most amazing little lad we have ever fostered; never complains or cries even when he had a halo fitted, and at the old age of 2 he was doing head stands in the garden, which I might add gave Manchester a dicky fit, especially when I showed them a photo of him doing just that!

I just wanted to put a little something back, but didn't know what as I can't run, I'm not too good at long walks and I'm scared of heights, so doing a skydive was out of the question. So what about a tattoo, I thought; it isn't gone in one run, it will stay with me just the same as the MPS will stay with my lovely boy. I can't give his name as he is a fostered child, but we have been told he can stay with us for as long as he needs to.

The tattoo I had is of the MPS logo in a nice blue and I am very proud of it, even though I have never liked or agreed with them, this is very special to me.

Sue Payne

Sue raised an incredible £800 from her sponsored tattoo!

## Mind Over Matter

It would seem that our supporters are very much up for a physical challenge - and they certainly do not do things by half! The next four pages highlight some events, from the tough to the extreme, that have been taken on to raise money and awareness for MPS.

It goes without saying that we are seriously impressed...

### My Ironman Journey

I think it's safe to say 2014 has definitely been my most gruelling in terms of physical training and challenges. I have been a fairly regular competitor in triathlons for the past few years and am a keen road cyclist, but nothing has been as demanding as signing up for the Ironman UK this year.

The reason I decided to take the plunge was due to the fact my wife's cousin, Sammy Watts, had been diagnosed with MPSIII Sanfilippo a couple of years ago and having all been completely unaware of the disease, we were devastated to hear of how it takes effect and that currently there is no cure.

We are in awe of how Laurie & Hannah (Sammy's parents) have coped since the diagnosis and done everything possible to ensure his quality of life and enjoyment is maximised as much as possible. We have two small children and can only begin to understand how they must feel and the dedication it must take to move forwards each day with every issue and hurdle that appears.

I have never before taken part in any race for charity as I always felt I was doing them for pleasure and didn't want to ask as it seems the nations is competing in some race or another every weekend! This felt different - I decided if I could do something that could help raise awareness and potential funds for the amazing work the MPS Society do, then it had to be for the Ironman race.

The race includes a 2.4 mile open water swim, 112 mile cycle finished off with a 26.2 mile run - needless to say; I had to up my training regime! Having signed up early Jan and starting to ask for sponsorship, I realised to complete the race in my desired time I needed to do things properly - I hired a nutritionist to devise me a full 3 month plan taking in all the necessary quantities of protein, carbs and nutrients. I also went to see a physiotherapist each week to ensure any aches and pain were ironed out and I was stretching my body effectively to appease future issues, I also had a full training regime mapped out listed exactly what I had to complete each day and at what time.

My biggest investment was time - in all honesty I relied heavily on my wife to look after the kids in the 3 month build



up as most days I was up pre 5am to either run or cycle to work, or swim nearby. Every Sunday I was out on the bike for around 5hrs starting pre 7am and returning home for around 2 minutes before heading out for a 10k run.

I dropped over a stone in weight from beginning of training to post race and became somewhat obsessive about my training stats, distances, etc - much to the annoyance of my wife!

When the alarm went off as 3am the morning of the race, it's safe to say I was nervous, but above all excited. By this point I had done everything I could in terms of preparation and now just needed to focus on completing the race in the best time possible.

I can safely say, the experience was one of the best of my life - I completed the race in 11hrs and 44 minutes and although had a few moments struggling during the run, I knew I could complete it and felt elated when I did.

The thing I am most proud of and so touched by is the amount of money I managed to raise - over £2000 achieved alongside incredible messages of support from all my friends and family. If competing in this race has managed to help even in a small way towards research and ultimately a cure for this cruel disease, then it was beyond worth it.

Anyone toying with the idea of doing something similar, I would say don't even think about it - sign up now!! Needless to say I've now signed up to do it all again next year, now just need to break it to my wife....

Steve Houghton

### The Thames Path Challenge



Elliot Moody (brother to Ollie Moody, who suffers from MPSVI Maroteaux Lamy) recently completed the Thames Path Challenge in September 2014. This 'Ultra-Challenge' covers a distance of 50km - to put that in perspective, it is the equivalent of two and a half Great North Runs or one and a half London Marathons.

The run took place on Saturday 13th September 2014 and began in Runnymede, which is just outside Old Windsor. The 50km route followed the Thames Path round to Putney Bridge at the finish beside the river at Henley-On-Thames. Despite getting lost at one point and taking a ten minute detour(!), throwing down my routine black coffee and banana at the 4x five minute rest-stops, and running in a slightly odd fashion with calf cramps for the last four miles(!), I somehow managed to be the fifth person to cross the finish line, and the first from the group I set off with. I set off from the start at 8.45am, and finally crossed the line just after 4pm - with a finish time of just over 7 hours 20 minutes.

Elliot Moody



### Tough Mudder

On Saturday 26th April this year I took part in my first Tough Mudder to raise money for The MPS Society and The Hotcourses Foundation jointly. Tough Mudder is a 12 mile run combined with 20 obstacles and it took my team and I around 3 and a half hours to finish.

Some of the challenges we faced included crawling through narrow muddy tunnels, climbing over walls, jumping 12ft into a pool of water and 'Electroshock Therapy', which used 10,000 volts of electricity to shock us as we crossed the finish line.

It was a huge challenge for me as I had never run further than 3 miles in my life, but when either my teammates or I felt like we were struggling we reminded ourselves of how much we had managed to raise for our respective charities. I was delighted to raise £3,380 in total and hope to do another Tough Mudder in the future to continue to support this incredible organisation.

Natasha Fairweather

Our thanks also to Norman Chambers who at 78 ran the Bristol Half Marathon in a time of 2hrs and 20 mins.

He raised just over £100.00 for the Genstein Trial.





### Suzy & Jeff's Great South Run

On 26th October 2012 my husband Jeff and I were saying our vows and getting married. Skip forward two years and we were pacing our way round the 10mile course of the Great South Run, something I could have never imagined doing.

Over these last 9 months, 11 members of my family have been diagnosed with Fabry disease, including my mum, both my sisters and my nephew; and the debilitating condition has left my cousin, who has been most affected, struggling to perform even basic daily tasks. Although obviously relieved when my results came back negative, I felt that I had to do something to support the rest of my family who had not had such good news. So I signed up to do the Great South Run on behalf of the MPS Society.

I have never done any endurance sport, and I barely covered two miles on my first training run. My second, third, fourth and fifth runs were much the same. It felt like I was getting nowhere. But slowly, I was able to increase the distances I was running and come rain or shine I would be out pounding the streets of Bicester. I can't say that I enjoyed my training sessions, in fact I disliked them so much that the biggest mental barrier I had was actually getting myself out of the house!

We set our first fundraising target at £1000. Ambitious, but we thought we should be able to get there. Within a week the target had been smashed. We were blown away by the generosity of friends, family, and people we hadn't even met. And the money just kept rolling in. In response to the donations I felt I had to give my absolute best for the race and so I decided to go teetotal and started eating an athlete's diet.

On race day I was incredibly nervous as we gathered at the start along with 25,000 other people. But as the race began I found myself able to relax, and actually enjoy running! The atmosphere was incredible, with people lining the streets the entire route and it was made all the more special as my family made the trip down south to support us.

I have been able to hang up my running shoes knowing that I never have to go through that pain again, but I am aware that for sufferers of MPS and related diseases there is no escape. I am so proud of my family and the resilience they have shown in the face of their health issues and so I dedicate our total raised (£6520.02) to you: Mutti, Lexi, Pip, Jacob, Glyn, Matt, Jan, Cathy, Lainey, Sophie-Rose and Mikki. I hope this can go some way to making your lives easier.

Suzy Elliott

### Barry Quant's Run for Genistein

Barry's friend, Andrew, came up with the idea to run for his own cause and encouraged Barry to run as well. Barry decided to fundraise for the MPS Society as our son Lewis has Sanfilippo. As neither of them had done any running since leaving school (many years ago!), apart from running after cattle, as they are both farmers, they thought they had better start training.

After a lot of hard work and aching legs in the build-up to the run, they both finished on the day in good time, with Barry coming in at 2 hours 24 minutes – 30 minutes earlier than he had expected!

Jenny Quant – Barry's wife

Barry raised an incredible total of £1717.22 for the Genistein Trial.

### Great North Run

I set myself a MASSIVE challenge by entering the Great North Run in September 2014 - the furthest I had ever run before was 10km and that was over 3 years ago! After finding out earlier this year that my gorgeous niece Thea Paterson had been diagnosed with MPSI, I felt that I just wanted to do something productive - feeling helpless as Richard & Sam (Thea's parents) know all too well is one of the worst feelings in the world especially when it's your own special child.

Sadly, Thea lost her battle with MPS in June at only 9 months old due to many complications. She will be missed by all who knew her during her short life. The run itself was absolutely amazing, the atmosphere was out of this world and the support was overwhelming! It was quite a hot day so my goal for completion in 2 hours 30 minutes wasn't quite realised but I was only 10 minutes over!!"

Aileen Cowler

If you would like to challenge yourself in 2015, you could register now for one of our places for the following events:



- Great Manchester Run - 10th May 2015
- Great Manchester Swim - 4th July 2015
- Great North Run - 13th Sept 2015
- Great South Run - 25th Oct 2015



We only have a very limited number of places for each event, so please let us know that you are interested by emailing [fundraising@mpssociety.org.uk](mailto:fundraising@mpssociety.org.uk) for more information.

Alternatively, you can always enter the event of your choice under your own name and still fundraise for the MPS Society - just don't forget to let us know!

## 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.

## Thea's Trust Ball



All photos courtesy of Nicola Denby Photography

On Friday 28th November we were privileged to attend the inaugural Thea's Ball in Harrogate to receive a cheque of £5000 from Thea's Trust on behalf of the MPS Society.

The Ball was a sports star-studded event at the palatial Crown Hotel with over 200 people attending what was a very well organised, memorable and emotional evening which raised a considerable amount of money through a raffle and auction.

The Trust was set up by Richard and Samantha Paterson following the birth of their second daughter Thea. At aged 6 months she was diagnosed with MPSI Hurler Syndrome. After her diagnosis Thea underwent vital ERT (enzyme replacement therapy), chemotherapy and a bone marrow transplant at Manchester Royal Children's hospital. Unexpectedly during her transplant process Thea suffered multiple complications' including contracting Influenza A and developing Graft Vs Host disease which resulted in her passing away in May 2014.

The purpose of the Trust is to raise awareness of MPS diseases, to provide financial support to families with children diagnosed with MPS across the UK and to donate funds directly to the MPS Society to help with advocacy support and future clinical research projects.

For me personally it was a privilege and honour to receive the cheque from Richard and Samantha. I was also able to speak to the guests about the MPS Society as well as our experiences of having a child with Hurlers.

James Hope-Gill

Our thanks to Thea's Trust for this incredible donation, and to James for representing us.

If you would like to read more about Thea's Trust, please take a look at [www.theatrust.co.uk](http://www.theatrust.co.uk)



### Elizabeth Heath shaves off her curls for MPS

In September 2014, Elizabeth Heath continued her run of fundraising (see our Autumn 2014 magazine) by shaving her curls for the MPS Society.

Sadly, Elizabeth's son Jack lost his brave fight to MPSII Hunter in 2013, aged just 13 years old.

On her JustGiving page, Elizabeth writes "when Jack was diagnosed I asked if organ donation was an option, alas it wasn't - I would have given anything to save my sons life - by shaving my head this is a small sacrifice I can make to raise awareness of these diseases and the Society that assists the families and sufferers, and funds research to try and find a cure."

So far Elizabeth has raised over £3,000 from all her fundraising.



### Charity shave at The Still

Four lads from our local pub decided they would not shave or have a haircut for 3 months and then on Christmas Day they all had it shaved off. The lads involved were Adam Ward, Dave Murphy (Spud), Francis (Lurch) and Sam. They managed to raise a brilliant £765.

This took place in our local pub called The Still in Lincoln, Lincolnshire, where one of the lads involved, Dave Murphy, is the landlord.

Lisa Patrick

### SPAR's Halloween Fundraiser

SPAR Central Avenue, Grangemouth held a very successful fundraising event to help raise funds for local 2 year old girl Chiara Peterson who suffers from MPSI. The fundraiser featured a Halloween theme and was held inside the SPAR store on the 1st of November.

The fundraiser made £350 on the day and another £350 was given from CJ Lang & Sons Ltd from the funds raised from the sale of carrier bags.

The entire SPAR Central Avenue staff got behind the cause giving up their free time to help make the fundraiser a success, employees dressed up in fancy dress on both Friday and Saturday and got involved selling raffle tickets for a very special



Halloween cake made by one of the staff members. The store also held a tombola, where all items were donated. CJ Lang and son's Ltd donated 100 goody bags filled with SPAR branded products to the tombola.

The pumpkin carving contest, guess the bear's birthday and a children's

colouring in competition proved to be very popular with the younger members of the community.

The fundraiser was fun filled day and SPAR Central Avenue were delighted to raise money for such worthwhile cause. Unfortunately Chiara was unable to attend the event actually in hospital on the day of our fundraiser which made my staff even more determined to make a success of the day. Chiara's mum Michelle told staff of the struggle Chiara will face in the years to come. Chiara's bubbly personality and big smile gave inspiration to everyone to get behind SPAR Central Avenues first venture into fundraising as a store.

The staff at Spar, Grangemouth



## Thank you to all our donors including...

Lucy McKay took the Ice Bucket Challenge and donated £10.

Philip Fairclough took the Ice Bucket Challenge and donated £20.

Michala Mawdsley, Gillian Ward and Alex Vasey ran the London 10k for Tillie Mae (MPS III) and raised a spectacular £1556.43 on Michala's JustGiving page.

Jane and Emily Wallis completed the Macmillan Cyclettea in Bedfordshire (featured in Autumn 2014 magazine) and raised £1027.15 on their JustGiving page.

Jacob Singleton completed a coast to coast cycle challenge (featured in Summer 2014 magazine) and raised a total of £941.90.

Kate Collins took the Ice Bucket Challenge and donated £15.

Megan Micklewright raised £52 in sponsorship money for climbing Snowdon.

Jenna and Paul Cole trekked up Ben Nevis and raised £300 on their JustGiving page.

Jason Smith (featured in Autumn 2014 magazine) raised a grand total of £1023.37 for running seven marathons in seven days, and choosing the MPS Society as one of the many charities he supported.

Jessica Bumbay ran the Jane Tomlinson Leeds 10k for the MPS Society and raised £321.25 on her Virgin Money Giving page.

Matt Carter shaved his head to support his son Jacob (MPS I) when he lost his hair due to chemotherapy, and raised an incredible £1636.02 on his wife, Jen's JustGiving page.

Tina Brodgen took the Ice Bucket Challenge and raised £100 on her JustGiving page.

Ken Pierce completed the Prudential London to Surrey ride for the MPS Society and raised £115 so far on his JustGiving page.

The team at Karmarama – a London-based advertising agency – completed the Tough Mudder and raised £1316.34 (including gift aid) on their Virgin Money Giving page.

Sue Standish-Murphy, grandmother to Avah (MPS I), as well as some family and friends made a spectacular 'Frozen' float (featured in our autumn magazine), which she took along to several of the Isle of Wight's carnivals

where she and her team did a great job of raising awareness and funds. They managed to raise £247.30.

Mr and Mrs Mellor celebrated their Golden Wedding Anniversary this year and asked for donations from their guests in lieu of presents. In total they collected a wonderful £190 for the MPS Society.

Iver Heath Bowls Club collected for the MPS Society during a recent Ladies Captain Day, and raised £100.

Martina Wallis sold tea and cakes at Mr and Mrs Wallis's Garden opening and raised £100. Martina's cousin Isaac Turner suffers from MPS I.

Mr and Mrs Jowett donated £50 to Louis Garthwaite's Guildford to Paris bike ride.

J.F.B. and D.F. Hunter donated £50 to Louis Garthwaite's Guildford to Paris bike ride.

Mrs C Metaxa donated £50 to Louis Garthwaite's Guildford to Paris bike ride.

Tina Fanneran completed the Great North Run for the MPS Society and raised £347.50 (including gift aid) on her JustGiving page.

Connor O'Farrell pledged to get head, legs, arms and chest waxed depending on how much was donated: friends and family stepped up and Connor raised £470 in total on his JustGiving page. Ouch!

Jessica O'Neill completed the Yorkshire Spartan Race, a fearsome obstacle course and sprint, and raised £85.

Simon Farrow ran the Great North Run and raised £614.25 in total for the MPS Society.

Sarah Elizabeth Day collected a total of £343.75 on behalf of her friends Tony and Paula who did a skydive for MPS!

Jessica Mullard took part in a 3-legged race and raised £85 on her JustGiving page.

Emma Stuart did a tandem skydive for the MPS Society and raised £936.25 in total on her doitforcharity page.

Elizabeth Gill donated £121 to the Genistein Trial.

Hannah Russell ran the British London 10k and raised a total of £1493.75. Hannah chose to support the MPS Society as her brother suffered with an MPS disease and it would have been his 30th birthday on the day of the run.

Ian Evans and the team at Arriva The Shires, collected for the MPS Society during RAF Halton's village fete and

Cheersley village fete, and raised a total of £270.15.

Mrs P von Spreckelsen donated £50 in lieu of a present for Vicky Fairweather's 50th birthday.

Dr Peter Gordon lent out his marquee to a friend and received a cheque for £50 for the charity of his choice, which was the MPS Society!

The MPS Society recently appeared in Asda's 'Chosen By You, Given By Us' green token scheme at the Skelmersdale CDC Depot, and as a result of the vote, the Asda Foundation donated £200.

Xanthe Whittaker and her family donated £150 for Louis Garthwaite's Guildford to Paris cycle ride.

Sarah Robson from the Sunderland branch of RBS sent in a donation of £33.47 for the Genistein Trial, which were the proceeds from their collection box which was displayed in a local shop.

Mark and Debbie Burniston, Michael Riley and Michael Maule ran the Great North Run and raised £223.75 on Deborah's JustGiving page. The team completed the run in memory of Jake Corcoran (MPS I).

Hayley and Helen Lever ran the Great North Run for the MPS Society and raised £256.43 from online donations on their JustGiving page.

Tracy Wright ran the Great North Run and raised £335 for the MPS Society. Tracy's best friend has a nephew with an MPS disease, and so she wanted to help make a difference.

Vinnie Hutchinson completed the London to Brighton cycle in memory of Gracie Sims, and raised £115 on his online page, with more yet to come.

Matt Baggot also completed the London to Brighton cycle and raised £87.50 for the MPS Society.

Marina has donated another £5,500.67 from the sale of second-hand goods in her shop, 'Marina & Friends', in Bristol. So far the shop has raised a grand total of £130,155.38 for research into Sanfilippo disease.

The Provincial Grand Lodge of Bedfordshire kindly donated £700 to the MPS Society during a cheque presentation evening, in which representatives from the Society were able to speak about the MPS and the work we do.

Linda Beaumont and Shirley Bown held a collection at their local school and raised £60.15 for research into MPS.

## Donations

Ian Hardcastle; The Sir Jules Thorn Charitable Trust; Dawn Smith; Margaret Brock; Judith Swann; Michelle Brooker; M.J. Newell; Mrs A. Baker; Rick & Ann Coleman; St Mary the Virgin Church & churches in the Kinwarton area; Kelly Reynolds; AJ Gunary; Sandra Silcock; Dawn Norris; Mr R.J.L. Byrom; Margaret Lyon; Richard Gregory; Trevor Brown; Norman Saville; Mrs M.A. Jones; Mrs D.P. McDonald; Mr David Caukill; Margaret; Mrs K. Chivers; Mrs L. Sutton; Caroline Bell; D. Haskell; Dr A. Bansal; Carol Copsey; Mrs Y. Puddy; Mrs E.M. Mee; Susan Clarke; A. Palmer; Yvonne Pearson; Jilly Holroyd; Sue Peach; Edna Morrison; Susan; Mrs B. Lunt; Janet Croft; Elizabeth Powell; Janet Hillier; Darrin Minett; Mr T. Lavelle; Mrs M. Loveday; Joan Marsh; John Michael Brown; Anna Eaton; Andrew Seeber; Maria Murphy; Sue Hollidge; Alan Muers; Silvia Marsella; Yuki Nicholas; G. Plummer; Valerie Zaldua; Michael Skidmore Estate Agents; Barbara D. Penny; Ruth; Ruth Hall; Andrew Culley; Jennifer Johnston; Stephen Sefton; T.I. Page; Sarah Read; Mrs L.

Healthcare at Home ran an employee survey and as an incentive they pledged to donate £1 for every completed survey. As a result they raised £156 for the MPS Society.

Ann Matthews made loom bands and sold them to friends and colleagues and managed to raise £41.

The Buckinghamshire Trefoil Guild raised £165 for the MPS Society at their Annual Summer Gathering.

Priory Preparatory School donated £1,000 which were the proceeds from various fundraising activities over the year, including cake sales and cross-country competitions.

Terrie Brown completed the Great South Run in memory of Annabelle for the MPS Society and raised a total of £226.25.

Xanthe Whittaker took up the challenge to Go Sober for October and raised a total of £622.22 for the MPS Society.

Samantha Burrows completed the Great Birmingham Run and raised a total of £287.50 on her JustGiving page. Samantha is friends with Joe Hiller's (MPS II) mum.

Sara Staveley ran the Plusnet Yorkshire Marathon in support of Blake Knaggs, who suffers from ML II, and raised a total of £397.50.

Jack Vicary completed the Wirral Half Marathon and raised £62.50.

Nichola Boynton completed the gruelling obstacle course, the Night Reaper, in support of Blake Knaggs and raised £222.50 for the MPS Society.

Harriet Butler and her colleagues at Fayers Plumbing and Building Supplies Wore it Wicked and held a cake sale on Halloween, raising £237.50. Harriet's nephew, Jacob Carter, suffers from MPS I and has just arrived home after having a bone marrow transplant.

Ruth Wilson donated £10 to commemorate Morvan Pritchard's 40th birthday.

Amicus Therapeutics Wore it Blue for MPS and raised £27.80.

Sandra Morgan took the Ice Bucket Challenge and donated £10 to the MPS Society

The Royal Grammar School in Guildford held a school multi day and managed to raise £817.38 for the MPS Society

Chambers; Christine Hume; Michael Edwards & Louise Bee; Holly Nowell; Mrs W.J. McGinn; Mrs L. Chambers; T. McGinn; William Todd; Joan Ingram; Howard Matthews; Jenny Quant; Amanda Browne; Ethel Shannon; Mrs P.R. Darper; David Howe; Marjorie Banks; Kathryn Wallis; Christine Halleron; Margaret; Jane Inkson; Margaret Leask; Mr D. Ehrmann; P.Headland; Louise Lambe; David Tonge; John Sanderson; Bob Stevens; Alison Wilson; Tim Summerton; Christine Lavery; Thea's Trust; Claire Snowdon; Sally Richards; Baron Davenport's Charity; The Shauna Gosling Trust; Margaret Smith; J.Piomalli; Fer Pidden; Nailsea Lions; Anne Hooper; Amanda Goodall; Julie Solomon; Sarah Winzar

## In Memory

Roy Parkinson; Gethin Robins; Mr Gilbert Sharpe; Frederick Roy Chambers; Mr John Frederick Corder; Gracie Bella Sims; Mr Samuel Joseph Wallace and Mrs Edna Wallace; Mrs Valerie Howard Hollings; Gareth Evans; Rajesh Ravji Varsani; Denis Rowan; Abigail Harvey

Margaret Richardson and Blair Nicholson donated £25 in lieu of a present for Morven Pritchard's birthday

Jane, Martin and Ellen Bate held a coffee morning at the Witney High Street Methodist Church, and raised £72.

Kelly Mitchell completed the Great South Run for the MPS Society and raised £240.71 in sponsorship.

Marc Pearson ran the Plusnet Yorkshire Marathon and raised a total of £162.50 on his JustGiving page.

Andy Heazelton completed the Great South Run for the MPS Society and raised £525

Ian George donated £100 from a charity raffle held at his 50th birthday.

Vicky Fairweather recently held a birthday party and asked her guests to donate to the MPS Society in lieu of presents. Vicky sent in £350, which had been kindly donated by friends and family. Vicky is friends with Claire Garthwaite, who has two sons who suffer from MPS II.

Iris Hitter held a quiz night to remember her daughter Jacqueline (MPS III) and in support of MPS, and managed to raise £230.

Lauren and Mat Sims sent in a cheque for £570, which was raised by their local golf club who held a charity day in honour of Gracie Sims. Many thanks to all involved.

Macy Ann Bechemin and her friends from Bel Royal School made loom bands and sold them, raising a total of £60 for the MPS Society. Macy wanted to help as her brother suffers from Fabry disease.

Staff at Great Sankey High School donated £145 as sponsorship for one of their maintenance staff who completed the London Triathlon, who wanted to support one of their former students, Lauren Bell (fucosidosis).

The Meadows Primary School collected donations, held a raffle and a school disco and managed to raise £152.

Carol Westland and her friend Joan, recently held a table top sale at Didcot Civic Centre, and raised £183.

The Medics Society at Dr Challoner's Grammar School donated £50 in lieu of a fee. Christine Lavery gave a speech to the young medic students and asked that her fee be paid to the MPS Society.

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

J Casey; EM Lee; J & V Hastings; R & K Dunn; S Lttledyke; Norman Saville; Marcia Tosland; S Bhachu; C Cullen; S Brown; I & A Hedgecock; G Simpson; P & R Shrimpton; W Cavanagh; B Harris; L Brodie; B Weston; M J Peach; Raymond Arnold; J Ellis; I & V Pearson; D & A Gunary; D & S Peach; N & S Cadman; J & F York; J Wilson; A Tresidder; J & M Wood; E Cox; M Rigby; K Robinson; Mr Thompson; K Osborne; C & M Gibbs; Andrew Cock; Alan Dickerson; P Summerton; E White; C L Hume; A Weston; E M Lee; D Palmer; S & D Greening; Z Gul; M Reeves; V Little; S & J Home; K & S Bown; A Ephraim; C Pierce; R Gregory; R Taylor; M Leask; G Ferrier; E Parkinson; M & F Fullalove; E Brock; K Brown; A Sabin; M Hahner; E Moody; E Mee; M Malcolm; J Dalligan; A Sullivan; A Byrne; N & S Cadman; R & J Richards; J Heritage; Neil Upton; V M Lucas; D Forbes; A Wells; Mrs K M Wallis; B M Giles; M Newell;

*Collection boxes, stamps, foreign coins, mobile phones, ink cartridges, jewellery*

Warren Farm Tearooms; Ian Evans; Miss L Ricketts; Mrs E. Prangnell-White; Ruth Hall; Karen and Andrew Weedall; Peter Swayne; Luke Edwards; Ethel Shannon; Mrs M. Davison

Staff and patients at the Portland Medical Centre recently held a fancy-dress fundraiser and raised a fantastic £173.84. The event was organised by Ranjit Sagoo, whose daughter has recently received a diagnosis of MPS I.

Anne Cotton and Heather Gordon held an auction and raffle, and received some very generous donations, and made an incredible total of £3,821.98 for the MPS Society. Anne and Heather were originally going to hold a charity golf club, but unfortunately the day was rained off.

Staff of the School of Language, Literature and International Studies at the University of Central Lancashire held a Garden Party and raised £120. Delia Jackson, one of those involved, wanted to support the MPS Society, as her son-in-law had passed away as a result of an MPS associated illness, and wanted to do something in his memory.

Steve Brooks, Brian D'Souza and their friends held a spectacular fundraising event in the village of Frimley Green, which involved a pub-to-pub three-legged race! The fundraising culminated in a lively MPS Ball. Altogether the Frimley Green team made an amazing £5,454. Thank you and well done to all involved.

Lucy King at GE Money Home Lending organised a spinning marathon and raised £181, which was matched by the GE Foundation.

Hayley and Helen Lever ran the Great North Run this year and raised a grand total of £1666.67. With thanks to BT for donating as part of a matched giving scheme.

Louise Hiller nominated the MPS Society for Lloyd's Market Charity Awards, and we were delighted to be awarded £2000

Robert Kenton and Jenny Kilham provided the music for an Christmas Carol programme for the Broadwater Strollers in Worthing and raised £63.23.

