# MPS magazine

Society for Mucopolysaccharide Diseases

Spring 2017 • www.mpssociety.org.uk

# Patient Advocate Leader 2017

CONGRATULATIONS TO CHRISTINE

RESEARCH & TREATMENT

A round up of the latest news from the pharma industry

WHAT YOU THINK

The results and responses from the member's survey

# Call for a PAUSE in the HST decision

read about the campaign on page 7

## 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, and can result in death in childhood.**

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

## 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. We aim to:**

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

## Board of Trustees



Registered Address:
**MPS Society, MPS House, Repton Place, White Lion Road, Amersham, Bucks, HP7 9LP, UK**

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## Magazine deadlines

Summer 1 June 2017
Autumn 1 September 2017
Winter 1 December 2017

To submit content email:
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The articles in this magazine do not necessarily reflect the opinions of the MPS Society or its Management Committee.

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



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# Contents



**Mobile phone scam**

We have had reports of a mobile phone scam from a company called Hot Prizes. If you are asked to click on any prizes for MPS Helpline then this is nothing to do with the MPS Society.

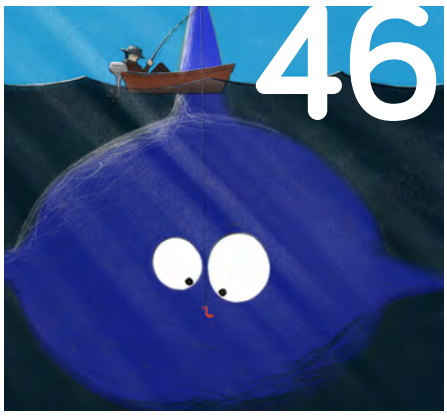
We advise you to ignore any phone messages like this and if you do have any concerns you should contact your mobile provider.


## PAUSE CAMPAIGN

The latest update from the campaign to PAUSE the NICE and NHS England decision on reimbursing rare disease medicine


## THE STRESS IS OVER

One family's fight for access to Vimizim




## THE BIG DRAW

A novel approach to fundraising

## WELCOME

It's our spring issue which means new beginnings, spring cleaning and lots of chocolate! Have a look at page 44 for some Easter inspired activities.

It is also nearly 15 May which means we're gearing up for MPS Awareness Day. You'll see our new look Wear it Blue poster in the fundraising section and the back cover tells you all you need to know about spreading awareness on the big day, as well as a leaflet enclosed for a trip to Gulliver's Land in Milton Keynes the Sunday before awareness day.

We have a lot of research and treatment news in this issue including a round up of the latest from the pharmaceutical industry, categorised by disease type, and the results of our very own member's survey.

You should also find enclosed a nomination form for the our 'Courage over Adversity' awards which will be announced at the conference in July.

# From the Group Chief Executive

*Christine Lavery*
*Group Chief Executive*

I am writing this to you as I return from one of the most vibrant and informative WORLDSymposium on Lysosomal Storage Diseases (LSDs) I have attended in its 13 year history. The programme brought together academia, clinicians, specialist clinicians and nurses, pharma industry and leading patient organisations to gain enormous insight into where we are in the LSD arena in basic research, translational studies and clinical application.

One of the key areas of interest for the MPS Society was the multi-pharma company interest in developing new therapies for the MPS, Fabry and related lysosomal diseases. Whilst it was disappointing to see little progress specifically in ML II and ML III it was encouraging to see the rapidly growing interest in gene therapy across many of the MPS diseases as well as ML D and Fabry disease. We can only continue to hope that developments in gene therapy may in time translate into a treatments for ML, Multiple Sulfatase disease and other particularly challenging LSDs.

> “One of the most vibrant and informative WORLDSymposium on Lysosomal Storage Diseases I have attended in its 13 year history

Another very important area of interest was the pre-clinical and clinical trial outcomes across a range of pharmaceutical companies developing therapies for MPS III. There is too much to report here but if you turn to the research and treatment section of this magazine there is wealth of information on MPS III and other MPS diseases including Fabry disease.

There were so many highs here at the WORLDSymposium but sadly we were recipients of two unrelated pieces of very bad news.

Firstly, the NICE announcement that Kanuma, an extremely effective enzyme replacement therapy for the treatment of infants, children and adults with LAL D, will not be recommended for treatment for both the infant and late onset form of LAL D. If diagnosed as tiny babies, Kanuma has been transformational, with infants on the clinical trials growing up and going to nursery and primary school with normal cognition and virtually no disease burden. The MPS Society issued its strongest press release ever and will not give up the fight for Kanuma. It is also appealing the NICE decision alongside the principal investigator of the clinical study and the company.

Secondly, the announcement by Alexion who is running a Phase I/II intravenous enzyme replacement therapy clinical trial for MPS IIIB will not be taking this clinical trial forward to Phase III/IV. At the time of writing we do not have a clear understanding of how this will impact our members’ children who are currently on this Phase I/II clinical trial but we will be offering support alongside the principal investigator of this clinical study to anyone affected by this news.

Finally in this issue you will see a piece on MPS events and safeguarding. As a result of one complaint viewed as unfounded by the Board of Trustees relating to the Lapland, Finland event some MPS colleagues who give their time freely at weekends to make our family events such a success are now reluctant to put their careers at risk. Families who participated in the Lapland, Finland event have all been written to and I and the Trustees are grateful for their incredibly positive feedback. However I would encourage you to read the article and if you have any feedback relating to MPS events, safeguarding or the value of these activities I would like very much to hear from you at c.lavery@mpssociety.org.uk.

*Christine Lavery MBE*

# News from the Board of Trustees

The Society's Board of Trustees meet regularly. Here is a summary of the main matters discussed and agreed at the Board meeting held on 26 November 2016.

## Governance

Paul Moody set out his experience as Acting Chair over the past three months. Paul said he had worked closely with Bob Stevens, Chair of MPS Commercial and stated that he would be prepared to stand for Chair but would want others to put their hats in the ring if so inclined. Paul gave an insight into the level of work and time involved. Tim Summerton then asked if there are any other nominations for Chair. In the absence of any other nominations Paul Moody was elected Chair.

As Paul Moody was one of the two Vice Chairs it was agreed the Board defer any decision on this role until the next Board meeting.

James Garthwaite resigned from the Board in October 2016 for personal reasons. Trustees were delighted that James could join the Board and MPS employees for their Christmas dinner on the 25 November 2016.

Trustees accepted Judith Evans resignation as Treasurer and discussed the relevance of having a Treasurer when the MPS Society has a Group Finance Officer. It was agreed that Judith has managed extremely well the Society's due diligence on all expenditure and agreed that the financial monitoring role is important but does not require the individual to be a Treasurer.

The Trustee Recruitment process distributed in advance was considered. Paul Moody said that it was an excellent document and we need to bring people onto the Board that strengthen the skills of the Board. The Trustee Recruitment Process was agreed unanimously.

The Trustees Induction pack, distributed in advance was considered. Tim Summerton said that it is a valuable document. The Trustee Induction Pack was agreed unanimously.

Pal Moody invited Trustees to comment on the Staff SWOT analysis and the Trustee SWOT analysis. Due to time constraints it was agreed to defer in-depth discussion until the next Board meeting.

## Personnel

The personnel report was noted. Trustees commented on the calibre of the employees and they being a high achieving team with an energy around being innovative.

## Financial Management

The Board considered the profit and loss report; cash flow; income and expenditure and Group Finance Officer's report and noted that fundraising income is down but also so are fundraising expenses. The Board met in private to consider the employee salary review.

## Risk Management and Policies

A review of the risk register took place. Two policies were deferred until the next Board meeting

## Advocacy Support

Paul Moody outlined the Advocacy Team Manager's proposal for recruiting an additional Advocacy Officers. Previously Sophie Thomas, Advocacy Team Manager had suggested to mitigate risk she really needed two additional Advocacy Officers but only asked for what she thought the MPS Society might afford. It was felt a valid point was raised that with just one extra Advocacy Officer Sophie Thomas had still left herself with a team that can't support the case load. The proposal put forward was to underwrite the funding of the one new Advocacy Officer post from MPS Society funds with a view that funds may be achieved through successful grant applications in the future for a second additional Advocacy Officer. The Trustees agreed to immediate funding for the employment of an additional Advocacy Officer per the proposal.

# WHAT'S ON?

## Regional Clinics

**Great Ormond Street Hospital**

MPS IV – 25th Jul, 10th Oct

MPS I BMT – 27th Jun, 12th Sep, 28th Nov

MPS III – 11th Jul, 26th Sep, 12th Dec

**Birmingham Children's Hospital**

MPS IV – 15th Sep

MPS III – 21st Jul, 18th Aug (afternoon)

Transition clinic – 24th Apr (afternoon), 29th Sep (afternoon)

Fabry – 24th Apr (morning), 19th May, 20th Oct (afternoon)

MPS I – 16th Jun (afternoon)

BMT – 16th Jun (morning)

Mixed clinic – 18th Aug (morning)

MPS II – 17th Nov

MPS VI – 29th Sep (morning)

**Queen Elizabeth Hospital Birmingham**

Adult Fabry – 9th May, 13th Jun, 11th Jul, 8th Aug, 12th Sep, 10th Oct, 14th Nov, 12th Dec

**Manchester Children's Hospital**

Post HSCT clinic (over 6 years) – 28th Apr, 14th Jul, 13th Oct

Post HSCT clinic (under 6 years) – 7th Jul, 6th Oct

## Conferences and Regional Events

**Gulliver's Theme Park, Milton Keynes to celebrate MPS Awareness Day**
Sunday, 14th May 2017

**MPS Awareness Day**
Monday, 15th May 2017

**Fabry International Network Meeting • Athens, Greece**
19th–20th May 2017

**MPS Weekend Conference 2017 • Hilton, Coventry**
7th July–9th July 2017

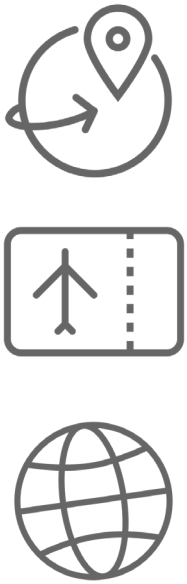
**Weekend for bereaved families • Warner Leisure Hotel • Thoresby** Hall, Nottinghamshire
13th–16th October 2017

**Childhood Wood**
15th October 2017

**MPS I/II Expert and Patient Meeting • Hilton, Northampton**
18th–19th November

**15th Annual International Symposium on MPS and Related Diseases • San Diego, California**
1–4 Aug 2018

# MPS Society events in jeopardy



It was lovely to see the wonderful photos in the Winter MPS Magazine of the families on the MPS Society organised event to Lapland, Finland and we have heard from many of you who had an amazing time!

As a Board of Trustees to the MPS Society, seeking and approving funding for events that may benefit the MPS Society's membership has been a high priority over many years. It is therefore only after much consideration that we have decided to tell you that these activities are now in serious jeopardy.

The Board of Trustees have had to respond to serious but unsubstantiated written safeguarding allegations made against the MPS Society by one parent in respect of the Lapland, Finland event in December 2016. As I am sure you will appreciate 'safeguarding' is taken very seriously in the MPS Society and such allegations cannot go unaddressed.

For clarity, I want to confirm that the visit to Lapland was a public event; shared with other holiday makers and was not exclusive to the MPS Society. The two members of staff on the visit to Lapland were there to facilitate the event from a logistical and operational perspective to ensure the Lapland event went smoothly and in case of an emergency to be on hand to assist appropriately, a role akin to a tour operator. Indeed, within moments of arriving in Lapland the two MPS Society employees assisted with a medical emergency, reallocation of appropriate rooms, as well as during the event, helping with the loan of a personal buggy, as well as offering to get a new one couriered to the airport if required. They also provided additional help to families as needed on an individual basis.

An allegation was made in respect of staff member's families attending the event. Firstly we must make it clear that the partners and children of these two members of staff, did not take spaces away from MPS members, nor were any allocated funds used. The commercial price of being in Lapland by the additional family members was paid to the MPS Society by the two members of staff and the MPS Society is in receipt of funds totalling almost £4,000 in respect of this cost.

In the 10 weeks between mid-October and Christmas 2016 the MPS Society held seven weekend events including Lapland. MPS staff attending these do so unpaid. The Board of Trustees have had an agreement in place for a long time that in exceptional circumstances like the Lapland trip, provided there is no cost to the MPS Society and that family members in attendance do not compromise the safety and quality of the event, this arrangement is in the best interests of the MPS Society and its members.

I am sure you will appreciate this but I need to make it very clear that at no time in the lead up to the Lapland event in any of the paperwork, or during the event, was it suggested, implied or stated that MPS Society staff would be acting in loco parentis. This was an MPS family event with parents in attendance and the parent(s) solely responsible at all times for the health, welfare and safety of their own children in Lapland. The Board of Trustees therefore concluded that at best the allegations were misguided.

Furthermore, in response to an allegation that MPS staff had a drink I confirm that the MPS Society has a policy that staff working in the capacity outlined in this letter must at all times drink modestly even in their own time. We find no evidence to the contrary. For clarification on events including sibling weekends where staff are loco parentis a strict no alcohol policy is in place.

Sadly, the situation the MPS Society now finds itself in is that whilst all staff are totally committed to working beyond the call of duty and optimising all opportunities for our members these unfounded allegations have been severely detrimental to the individual staff concerned and for the MPS Society as a whole. Lapland and our many other events are opportunities for MPS members to build cherished memories and in no way do we want to take away from you that experience. However, in essence due to the unfounded allegations made our rolling and future programme of events in the UK and beyond is potentially in jeopardy from a staffing perspective and we felt you needed to know.

The MPS Society has a robust Safeguarding Policy that can be accessed on the MPS Society website www.mpssociety.org.uk This communication does not require you to do anything, however knowing that hundreds of our members have enjoyed our MPS events annually we felt you should know the situation we face. If you would like to provide your own feedback or thoughts please send any comments care of the Group Chief Executive (c.lavery@mpssociety.org.uk).

*Paul Moody*
*Chairman of Trustees*

# NICE and NHS England's decision puts patient access to rare disease medicine at serious risk

**NICE and NHS England's decision to use a new cost-effectiveness threshold to decide whether rare disease drugs are funded or not has sent shock waves through the rare disease community. MPS Society and another 200 patient organisations have grouped together to call for a PAUSE to this process so it can be reconsidered properly. Read the MPS Society press release below and see how you can help with the campaign.**

Reeling from NICE's recent decision to change the arrangements for evaluating and funding drugs and other healthcare technologies assessed through NICE's highly specialised technology appraisal MPS Society Chairman, Paul Moody said:

"A decision by NICE and NHS England to implement this new policy in just two and half weeks' will affect the most vulnerable in UK Society and confirms that children and young adults with ultra-rare diseases going forward are economic pawns in a failing NHS and cheaper dead than alive. The UK government now needs to act at lightning speed and reverse this initial NICE & NHS England policy not just in the context of patients with ultra-rare diseases but also that of the life sciences industry who will see no incentive to investing in the UK market if their innovative medicines and technologies have no prospect of reaching the patient."

Christine Lavery MBE, Group Chief Executive of the MPS Society who very rarely speaks personally said "Being born with an ultra-rare disease, a disease affecting less than 110 people in England, is not a life style choice; it is no one's fault; it happens albeit very rarely; it happened to my son. At that time, there was no treatment and Simon died aged 7 years. I can only imagine now how it might be to be faced with a child with an ultra-rare disease who could be treated with a highly-specialised medicine but is denied treatment on cost grounds. The pain for the family of seeing their child condemned to death by Andrew Dillon, Chief Executive of NICE; Simon Stevens, Chief Executive of NHS England and the UK Government is unimaginable. Let us also be clear to Members of Parliament, many of the babies and children who will be affected by this catastrophic decision are 'yet to be born or diagnosed' members of your constituencies."

This decision is particularly devastating to the MPS Society who felt they had worked constructively with NICE and NHS England throughout the consultation period to devise a response that offered budgetary solutions whilst ensuring patients likely to show benefit from highly specialised technologies were given the opportunity of treatment.

If we don't stop these changes coming into force after April 1 this year, we will see the introduction of a £300,000 Quality-adjusted life year (QALY) threshold for medicines evaluated via NICE's Highly Specialised Technologies (HST) programme, which assesses treatments for ultra- rare diseases. This threshold will effectively stop the flow of new medicines reaching patients with ultra-rare and complex diseases. Many treatments for ultra-rare conditions that are currently funded by NHS England have costs per QALY of more than £500,000 including the three medicines that have been approved by NICE's HST process to date. It is widely acknowledged that QALY thresholds are not appropriate for evaluating medicines for ultra-rare diseases, due to the small patient populations and often limited data.

**In Parliament on 28 March, Daniel Zeichner, Member for Cambridge spoke at the Rare Diseases Strategy**

We must listen to what charities are saying. They warn that lives will be lost because of this new rationing system. I was particularly struck by the words of the MPS Society, which provides support to people affected by mucopolysaccharide diseases. It said that the decision by NICE and NHS England:

"will affect the most vulnerable in UK Society and confirms that children and young adults with ultra-rare diseases going forward are economic pawns in a failing NHS and cheaper dead than alive."

Those are very strong words. They are not mine, but they show how people feel, and I am sure that the Minister will have noted them.

Read the full Hansard here: https://goo.gl/LFy9PY

**Read the latest from the campaign page at: www.mpssociety.org.uk/mLuhM and find out how you can get involved.**

# Announcements

Nominate someone who has shown courage over adversity by 1 May 2017

## Courage over adversity

May 2017 is the 35th Anniversary of the founding of the Society for Mucopolysaccharide Diseases. To celebrate this milestone the Board of Trustees will at the MPS Gala Dinner at the conference on Saturday 8 July 2017 be recognising family members of the MPS Society who have shown 'Courage over Adversity'.

The recipients of these awards will have demonstrated how they have used their personal experience of MPS, Fabry or a related lysosomal storage disease to not only meet the challenge of MPS but build on their experience to benefit the wider community including the MPS Society.

It is at this point that we are asking all our members to put their thinking caps on and nominate individuals who have shown 'Courage over Adversity' and are members of the MPS Society in the following categories:

- **Adult** (18 years and over) living with MPS, Fabry or a related Lysosomal disease
- **Parent** whose child (of any age) is living with MPS, Fabry or a related Lysosomal disease or has died from MPS, Fabry or a related Lysosomal disease

We are also looking for nominations in the following categories:

- **Grandparent** whose grandchild(ren) (of any age) is living with MPS, Fabry or a related Lysosomal disease or has died from MPS, Fabry or a related Lysosomal disease
- **Young person** (13–17 year) living with MPS, Fabry or a related Lysosomal disease
- **Sibling** (13 years and over) whose brother(s) and or sister(s) have or have lost their life to MPS, Fabry or a related Lysosomal disease

We look forward to receiving your nominations no later than 1 May 2017 on the enclosed nomination form which can be returned by post or scanned in and emailed to b.cotterell@mpssociety.org.uk. The nomination form is also available to download from the MPS website at www.mpssociety.org.uk/TvPFQ

## NEW MEMBERS

The Abdul-Rauf family have recently been in contact with the Society. Their son, Khalil, has recently been diagnosed with MPS III Sanfilippo Disease. The family live in the Midlands.

Barbara and Tony Simpson have recently been in contact with the Society. Their two adult sons, Oliver, 42 years old, and Benjamin, 40 years old, have a diagnosis of MPS III Sanfilippo Disease. Oliver and Ben live in County Durham. Tony and Barbara live in Berwickshire.

Bernadette has recently been in contact with the Society. She has Fabry Disease. The family live in the Glasgow area.

Steve and Lorraine have recently been in contact with the Society. Their son Jon Charles has a diagnosis of MPS III Sanfilippo Disease. The family live in the South East of England.

Welcoming Amanda Minett to the MPS Society fundraising team

I was originally trained as an Architect in the 1980's, however multiple redundancies caused me to diverse into many fields including being the librarian at a private girl's school and running a charity shop.

I joined MPS Society in March 2017 from Leonard Cheshire Disability Chiltern House in Gerrards Cross, where I was the Volunteer Coordinator. At the MPS Society I will be working within the fundraising team, concentrating on Corporate Fundraising.

I have two grown up children in their 20's. When I am not working I enjoy walking my dog and knitting brightly coloured throws.

And Julie Dunster who will also be working with the fundraising team

I joined The MPS Society at end of January as a Grant and Major Donor Fundraiser. I am busy sourcing funding for The MPS Society by writing grant applications to charitable trusts, foundations and other funders and will be writing reports to show the benefit each grant has brought to The MPS Society. I am learning so much about the amazing work of The Society and am thrilled to be working for such a great organisations. I am looking forward to meeting some of the families and children we support at our Conference this July.

I have a background in housing research and writing funding applications and am driven by making a positive difference to people's lives. I have worked for various organisations including the Civil Service, national and local charities and a Hospice.

Outside of work I enjoy walking, cycling, handicrafts and various voluntary roles. I am married to Andrew and we have a rescue cat called Chilli Pepper who is like our child!


**Christine collected the Patient Advocate Leader (PAL) award 2017 at the WORLDSymposium in February. Thank you for all your lovely messages of support for Christine.**

"Many congratulations from us on your PAL award. Very much deserved. Thank you for all you do and have done for many years for our children and our siblings. Your work helps us survive this MPS life we were given." Tony, Lorraine, Gary and Rebecca

"She deserves this. She did Hayleigh proud." Robert Reynolds

"Congratulations Christine you really deserve this, I could never thank you enough for all you and others at MPS Society did for my two and me over the years" Nicola Hall

"Well done Christine, you deserve this as you are always there for every family that needs you. You are an inspiration to all...without you Christine we were lost, you were there all the way with us. Very grateful and thank you again" Ann Fleming Jones

"Well done Christine, you certainly deserve it. We are all very proud of you and the marvellous job you have done" Barbara Pollard

# Advocacy

Our advocacy support service is at the core of everything we do at the MPS Society. We know how isolating and challenging it can be living with MPS or a related disease so we want you to know that you are not alone and we are hear to help. We are always striving to improve the support we offer and to sure we respond to each individual need as best we can.

Our service is flexible and a wide range of support is offered on a needs led basis but here are some of the services we can offer.

### Telephone helpline
We provide an active listening service, information and support by phone, including an out of hours service. You can reach us on 0345 389 9901.

### Disability benefits
We provide help and support in completing in completing claim forms for Personal Independent Payment and, where needed, will take a representative role in appeals and tribunals.

### Housing and equipment
We 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
We help 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.

### MPS careplans
We undertake 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, by producing a careplan.

### Respite care
We work closely with a number of respite providers and can make individual referrals if needed.

### Independent living/transition
We provide 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.

### Befriender service
We link individuals and families affected by MPS and related diseases for mutual benefit and support.

### Bereavement support
We are here whenever you need us, especially at the most difficult times.

### Advocacy Resources
The Advocacy Team have also developed a range of information resources focussing on particular issues which are free to download from the MPS website: www.mpssociety.org.uk

- Life insurance
- Travel insurance
- Hospital travel costs
- Disabled access holidays
- Carers legal rights
- Carers allowance
- Wheelchairs and flights
- Guide to housing and disabilities facilities grant
- Benefits including, Personal Independent Payment, Benefit Cap, Council Tax Benefit and Universal Credit.

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

**Sophie**
Manages the MPS Advocacy Team

**Debbie**
MPS IV Morquio, MPS 1 Hurler BMT, Hurler Scheie, Scheie, MPS VI Maroteaux Lamy, MSD, ML II

**Rebecca**
Fabry, MPS II Hunter, ML III/ML IV, Mannosidosis, Fucosidosis

**Louise**
MPS III Sanfilippo (shared with Steve), LAL D, Gangliosidosis

**Alison**
Supports members living in Ireland

**Steve**
MPS III Sanfilippo (shared with Louise), MLD, AGU, Winchester, Geleo Physic, Dysplasia, SLY, Sialic Acid Disease

Each advocacy officer works to a high level of professionalism. To make sure of this the following skills, knowledge and person qualities are present, applied and reviewed regularly:

- Qualified Social work
- Public/statutory services
- Genetic services

We're pleased to welcome Alison back from maternity leave

## New home

The advocacy team moved to a new ground floor location at MPS House earlier this year and have been careful not to cause disruption to the support they offer while they were relocating. All phone numbers and emails remain the same so hopefully you have received a seamless service.

## Member registration

There have been quite a few new members registering with the charity over the last couple of weeks and we are making sure to process all information and get you access to services as quickly as possible. In the meantime if there is anything we can help with please get in touch.

## PAUSE campaign

We want to offer as much support as we can throughout the Society's PAUSE campaign where we are asking that NICE and NHS England PAUSE to reconsider changes to NICE's highly specialised technologies programme. If you need any help at all, from writing to your MP to finding out how the changes will affect you, then get in touch with the advocacy team. You can also find out more from our campaign page: www.mpssociety.org.uk/campaigns/

## Howletts – The Aspinall Wild Animal Park

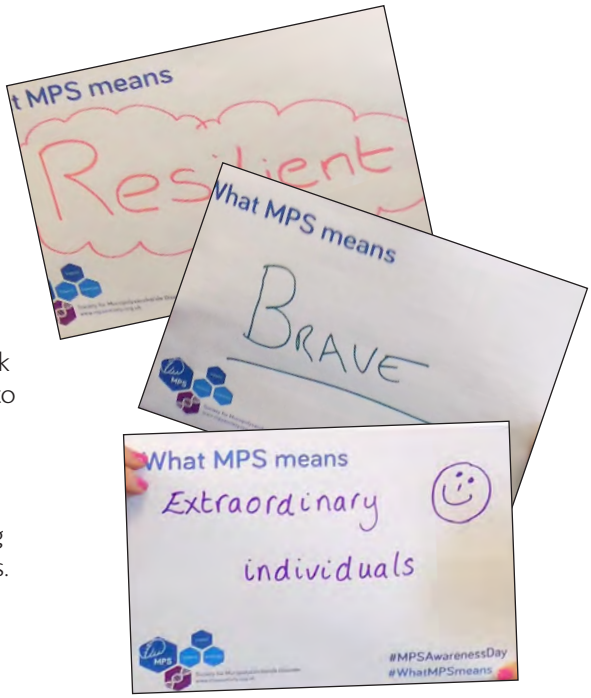
We are looking forward to seeing some of you at Howletts, the wildlife park in Kent. It is set to be a fun filled and exciting day and we are very excited to see the giant anteaters and the largest herd of African elephants in the UK.

## What MPS means

As we count down to MPS Awareness Day on 15 May we have been joining in this year's awareness campaign by thinking about what MPS means to us. Other than a disease, the word MPS sums up the close-knit family that are brought together due to the rarity of the condition. We look forward to seeing what MPS means to you when the social media campaign starts.

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:

advocacy@mpssociety.org.uk
0345 389 9901
facebook.com/mpssociety




# New staff at Queen Elizabeth Hospital

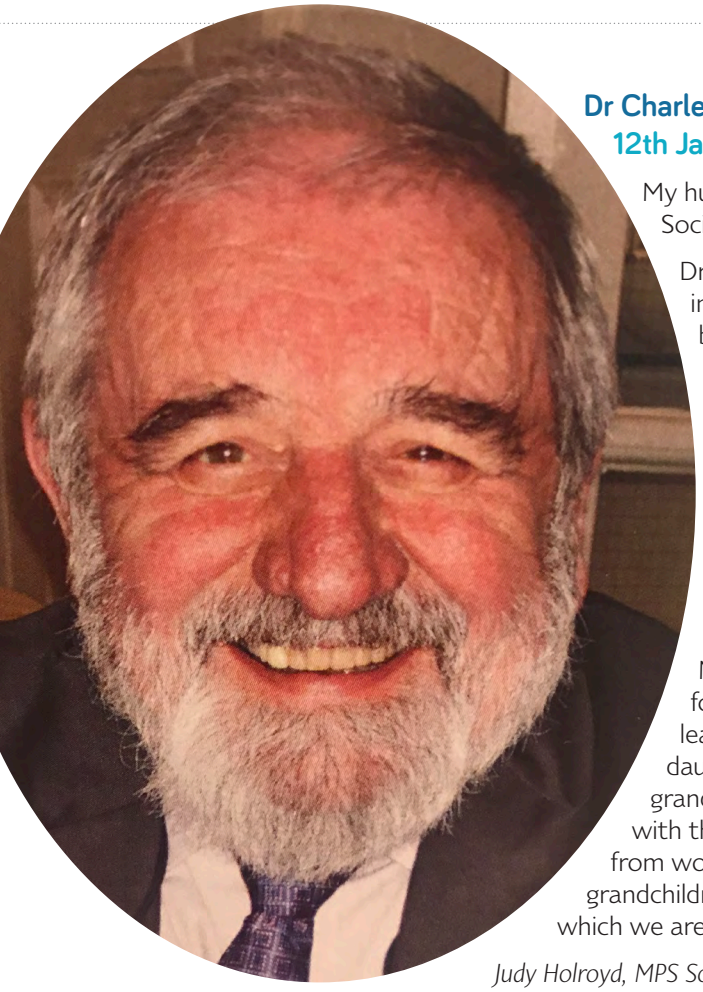
**Sarah Steeds** joined the Queen Elizabeth Hospital trust in 2015, starting as a nurse specialist for rare diseases. Having enjoyed this role, she was excited to join the IMD team as lead clinical nurse specialist in January 2017 and take on more of a key worker and patient centred role.

Sarah has come from a background of predominantly oncology and palliative care, with a keen interest and insight into research. Outside of work, Sarah is busy being a mum of three teenagers, not to mention the guinea pig and the dog!

**Helen Gallagher** recently joined the Queen Elizabeth Hospital trust in January 2017, joining Sarah as a clinical nurse specialist in the IMD team. Helen has also come from a background of oncology and palliative care, and recently completed an MSc specifically in palliative care. The role of CNS within the IMD team appealed as an exciting prospect, as a role where nurses can get to know their patients well and hopefully be a resource for information and support.

Outside of work, Helen is generally busy looking for a new project, from home improvement to creative baking ideas!

# Remembrance



## Dr Charles Anthony Pennock
## 12th January 1938 to 14th January 2017

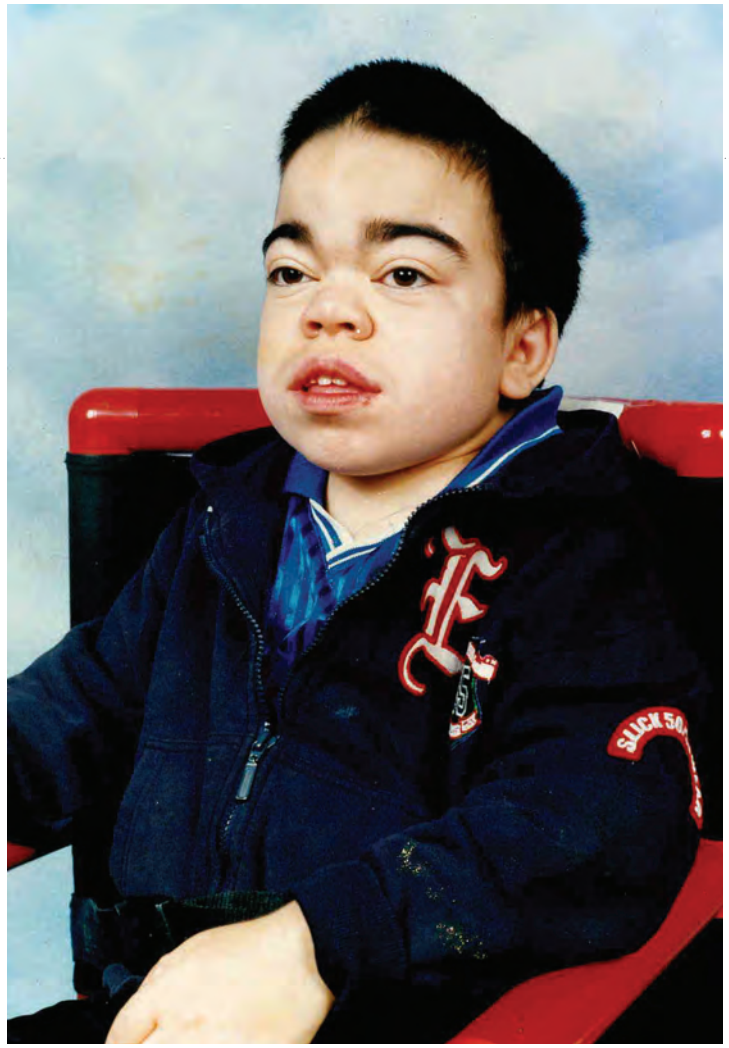
My husband, Chris and I attended the funeral service, on behalf of the MPS Society, to celebrate the life of Dr Charles Pennock on the 30th January.

Dr Pennock qualified as a doctor in 1962 and from 1963 began research into MPS diseases and inborn errors of metabolism. By 1972 he became a Consultant Chemical Pathologist and Senior Lecturer in Child Health at the University of Bristol. His research and expertise was respected worldwide and he was a leading authority on the diagnosis of MPS disorders by enzyme analysis, heading up the Enzyme Laboratory at Bristol for many years. He was conferred an honorary degree of Doctor of Science by the University of the West of England in 2000. He strongly supported the MPS Society and was an outstanding chairperson at several family conferences in the 1980s. He continued supporting students and giving tutorials up to the last few months of his life.

Many people as well as Dr Pennock's large family came together for the service which was held in St John's Church Keynsham. He leaves behind his wife, fondly known as Paddy to friends and family, daughter Sarah and son Christopher, many grandchildren and five great grandchildren. In the addresses he was described as an outstanding leader with the ability to look on the positive side. He had many interests away from work and his strength of character was an inspiration especially to his grandchildren. Donations for the service were collected for the MPS Society for which we are very grateful.

*Judy Holroyd, MPS Society Trustee*

From left: Charles Pennock speaking at the 1988 MPS Society UK conference, outside Downing Street with Christine Lavery and at the 1988 MPS Society UK Conference



### Shaun McCawille
### 23rd December 1990 to 29th August 2016

The 29th August was a sad day for us when our son, Shaun Paul McCawille, passed away. He was only 25 years old but for those years we were privileged to share in his life and we were all the better for it. He was a beautiful boy, always happy and friendly. It would be rare to see him without a smile on his face and a hand outreached for all those he met.

Despite having Hunter Syndrome, Shaun had great communicative skills and could let you know what he wanted despite the fact he had no speech. For such a young man, Shaun was remarkable and enjoyed his life in his own way with a strong interest in television and films with a particular love of comedy sitcoms.

To all of us in the McCawille family, Shaun was our best friend, our brother, our son. His loss will always be felt and he will never be forgotten.

## Bereavements

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

Asma Ullah who had MPS III Sanfilippo

Deborah Thorburn who had MPS III Sanfilippo

Peter Dine who had Fabry and passed away on 29 October 2016

Harold Martin who had Fabry and passed away on 17 November 2016

William Todd who suffered from MPS I Hurler Scheie and passed away on 17 December 2017

David Camfield who had Fabry and passed away on 9 March 2017

Erin Berry who suffered from MPS I Hurler and passed away on 17January 2017

James Stewart passed away on the 13 March 2017, he had MPS II Hunter Disease.

# Clinics


1. Roman

## BCH MPS IV 20 JAN

(1-6)


2. Asif

As far as clinic's go it was a calm clinic, which gave plenty of time to catch up with our children and families. It was lovely to see everyone looking well and hearing about what they have coming up in their lives.

It will be great to hear how Tia got on in her school play Bugsy Malone with not one, but three parts to play, and to see if her wish of splurging everyone with foam came true. It would be fantastic to see some pictures the next time we see her.

Juveria looked fantastic in her pretty dress as usual, and well what can I say about the double trouble duo Roman and Xena.

It was great to see you all and if I don't see you before, I hope to see some of you at our conference in July.

*Louise Cleary*
*Advocacy Support Officer*


3. Mohammad


4. Tia

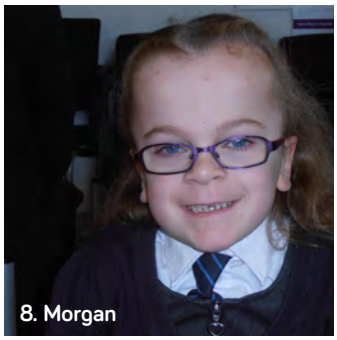

5. Juveria

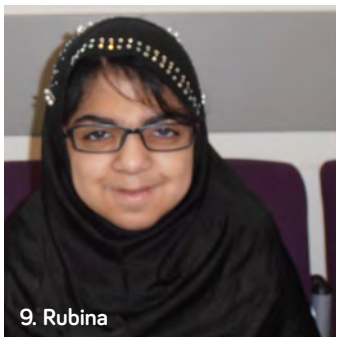

6. Xena


7. Rodina

## BCH MPS III & LAL D 24 FEB

(7)


8. Morgan


9. Rubina

## RMCH Post HSCT 20 JAN

(8–14)


10. Mia


11. Mikko and Harvey


12. Sonny


13. Chantelle


14. Melissa

As always it was lovely to meet with the families and staff at Manchester Children's Hospital in January. Everyone was in good spirits following the Christmas break. The children spent time trying to build the highest tower with the big blocks in the play area and succeeded after several falling down attempts!

*Debbie Cavell*
*Senior Advocacy Support Officer*


15. Sienna

## GOSH MPS I BMT 28 FEB

(15–16)

Lovely to meet all the families at GOSH on a wet and windy day. It's great when members have the chance to meet each other for the first time and it can be particularly beneficial for the parents to share experiences.


16. Seren Rose

# Your stories

*We are all fighters in our own unique, special way*

Barbara Mamatis was eight years old when she discovered her symptoms were caused by MPS I. The lack of understanding she experienced around her condition prompted a move into patient advocacy to pass on the knowledge she has collected including how she expresses herself through art and the benefits of pilates.

We are all fighters in our own unique, special way.

My story begins in 1982, when I was born in Sfax, Tunisia, to Greek parents, themselves born and raised in Cairo, Egypt. A minor dysplasia of my hip joints, a heart murmur and an umbilical hernia were detected by the paediatrician.

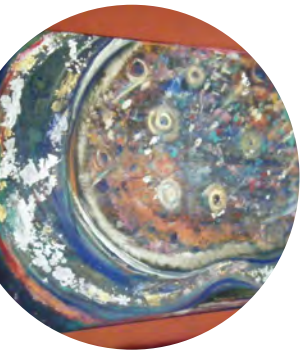
As I got older, there were no obvious signs that would reveal my rare condition. The only signs were my slightly reduced joint flexibility and movement.

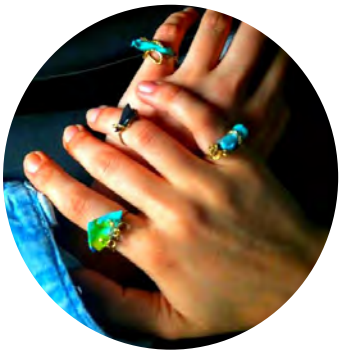
At the age of two we flew to the USA to seek medical advice from the Baylor children's hospital in Houston, Texas. The doctors at that time did not diagnose a syndrome but advised my parents to keep monitoring my heart condition.

At the age of eight, after having permanently settled in Athens, Greece, I started having vision difficulties as a result of a clouded cornea. My ophthalmologist, was familiar with my condition and as a result I was diagnosed with MPS I, Scheie syndrome.

Unfortunately the lack of knowledge, at that time, and understanding of the severity of my disease made me neglect my physical condition. I had muscular, skeletal and vision limitations and slightly deformed fingers but all that seemed normal to me.

I was advised to attend physiotherapy sessions from a very young age but I decided to quit as the physiotherapists were not aware of MPS and would cause me a lot of pain during my sessions.





I would never go beyond my natural limitations and was convinced these would never change.

Until the age of 21, I was lucky enough to lead a relatively pain free life that would not prevent me from finishing high school and studying International and European Affairs at the Panteion University of Athens.

Having an interest in communications I started, while studying, working for Ernst and Young, in the Marketing and Communications department. I was responsible for the Firm's Media Monitoring procedure.

It was around that time when I started experiencing my first intense muscular pain in my neck, back and arms. All those years of limited movement would now be the reason of having reduced muscular endurance, intense headaches, dizzy spells, burning, tingling sensations and muscular spasms in my back and neck.

I had to cope with my condition at work, with my studies and with the passing of my younger sister, who did not suffer from MPS. It was at that time in 2004 when I started my Enzyme Replacement Therapy and experienced my first panic attacks.

Having an extremely rare condition, and facing amongst others, the challenge of adulthood in an environment with no other people to relate to and in a country that provides minimal support, it was inevitable for me to feel lonely and helpless.

After an extensive search on the internet, my father found out, from abroad about a treatment for MPS 1 patients and the existence of The Greek Lysosomal Association in Athens. I was now slowly realizing that other people in Greece and around the world were experiencing the same difficulties that I was. I started to feel part of a group and not utterly alone in this race we call life.

But that was only the beginning. I had to accept my disease and start taking care of my body. ERT and continuous, specialized physiotherapy sessions were now permanently part of my life.

While seeking for a better future, I decided in 2008, to apply for the first traineeship Pilot program for people with a disability which the European Commission launched in Brussels. I was one of the 9 people with a disability to enter this 5 month work scheme program.

During my traineeship, I was working in the General Directorate for Communication and was

*I'm continuously getting to know my disease and trying to coach others to understand that they can move beyond their capabilities*

also part of the Media Monitoring group that would inform the Commissioners' Spokespersons.

Unfortunately my body was, once again, not in a position to handle my first attempt at independent living. I was experiencing great fatigue and muscular pain I could not deal with never the less I was determined to successfully finish my training.

As my body was preventing me from enjoying life and the benefits of being a full grown adult and not having a centre of expertise in Greece, I had to visit the Mainz University Clinic.

I was advised to start training to reduce my pain and try the therapeutic exercise technique called Pilates otherwise the intake of pain killers would be endless. Physiotherapy was not enough. I was determined to make my body and soul stronger. I had to quit my job at Ernst and Young, even though Greece was in the middle of a financial crisis in order to dedicate my energy into taking care of my body.

It was that time when my colourful art creations, paintings and handmade jewellery out of paper became a great way for me to express myself, proving that MPS cannot deprive someone from his talents as long as he has the will to discover them.

My MPS hands even through great difficulty would now create works of art that people could never have thought I was capable of. Being self-taught made them feel even more appreciative of my accomplishments, thus making my art even more unique.

It may take me longer than others to create but it's that great sense of accomplishment that makes any kind of pain and limitation seem insignificant. (Facebook page: Little Miss Cupcake's Art Creations).

Having also experienced major benefits from exercising on a regular basis such as growing taller, improving my posture and my flexibility and getting to know my body, its needs and its reactions, my teacher and mentor advised me to transmit all that knowledge I had obtained from my own experience and try to help others who may be facing similar difficulties.

Through great perseverance, tremendous work and continuous self-motivation though sometimes it seemed impossible to me, I finally managed last December to successfully finish the Pilates Teachers Training program after an intensive 3 year course. I was the only person with special needs within my training group.

My example gives people the strength and the motivation to work on themselves. Become more active and self-confident as I did.

I try, in my own different way, to be a reminder to others that despite a condition, whatever that condition is, someone can make a difference, with his or her own unique and individual way, as we are all special!

I'm still coming into terms with the fact that I was born with a very rare disease that makes my life very different from others, even from my own relatives, who none suffer from MPS.

I still have pain and muscular fatigue but less than I used to have. I feel more empowered as I now have several therapeutic tools and relaxation techniques such as Fascia and Reiki in order to cope with those factors.

I'm continuously getting to know my disease and trying to coach others to understand that they can move beyond their capabilities, as we all have infinite powers of mind over body. The first step is to learn how to love ourselves and accept reality as it is.

It's this kind of knowledge I desire to transmit through patient advocacy and by being a newly elected member on the Board of Directors of the Greek Lysosomal Association, I have the opportunity to do so.

We are all fighters for a better future, for a future with equal rights. With each MPS Society around the world, we get to build a stronger network that will make us never forget that.

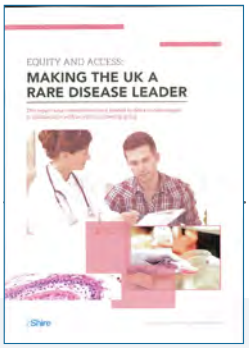
*Barbara Mamatis*

# Fabry patient competing in Miss Pinup UK

From Fabry sufferer to a pinup in the making, Gemma also known as Miss Lily Cheeks, burlesque starlet, is pushing out the boundaries of her condition and not letting pain crisis and stomach flare-ups stop her. Gemma is taking part in the Miss Pinup UK 2017 competition and it's her second year to enter. After reaching the final eight in 2016 on the Pinup Stage at the London Tattoo Convention in Tobacco Dock, Gemma has high hopes for this year's competition!

Miss Pinup UK was created by Rio Wild, Managing Director of Pinup UK who create all-inclusive Pinup competitions including Miss Pinup UK, Miss Pinup Ireland and Miss Pinup International which collaborates with like-minded competitions around the world, even as far as Australia. It's a celebration of all women and a wonderful fun experience for everyone!

Since entering Miss Pinup UK in 2016 Gemma has found a new support network and amazing friends for life with the Pinup UK Family. She may be suffering on the inside but now she doesn't feel like she has to hide from it. Gemma says, 'I may have a genetic condition but I'm not going to let that stop me from having fun!' The semi-finals and finals for the Miss Pinup UK take place in October. More information can be found at www.PinupUK.com or follow Miss Lily Cheeks on social media.




### Equity and Access: Making the UK a Rare Disease Leader

On 16th March just 18 hours after learning of devastating decisions from the NHS England and NICE consultation, a summit was held to mark the launch of a report into orphan medicinal products 'Equity and Access: Making the UK a Rare Disease Leader'. This report focuses on comparing the UK policy environment for patient access to orphan medicines to four of its European counterparts. Using new analysis from the Office of Health Economics and sponsored by Shire, the report puts forward a series of recommendations about making improvements to the access environment in the UK for rare and ultra-rare diseases in order that UK patients receive the world-class care they deserve and need.

At this summit I was invited to speak on the MPS Society's experience of specialised commissioning and the challenges that patients face due to determining value of a new highly specialised medicine. I was able to talk about the innovative Managed Access Agreement (MAA) for MPS IVA and how having an ultra-rare disease doesn't mean you can expect to be treated less favourably compared to someone with a condition seen more frequently.

On my way into the Kings Fund building for this summit I bumped into the Secretary of State for Health, Rt Hon Jeremy Hunt but needless to say he was not coming to this meeting to listen!

*Christine Lavery*
*Group Chief Executive*

‘The bad

# days and stress for Grace are now over'

After two years of campaigning, a girl with an ultra-rare disease has been told she can have the only drug in the world that can treat it.

Morquio has left Grace (9) the size of a toddler and living with pain and fatigue that will not get better without Vimizim.

"The bad days and stress of trying to get access to it for Grace are now over. She will get a chance at a better quality of life," her mother Grainne Cogan said.

The HSE told the family that Grace will be able to receive Vimizim – an enzyme replacement therapy – at Temple Street Children's Hospital.

Ms Cogan said she was shaking when she heard the news that she, her family and friends have worked so hard for.

"To us this means everything. Nothing comes close to this feeling. I feel like a weight has been lifted off me," she said.

The family live near Carrickmacross, a short drive from Northern Ireland where Vimizim is provided by the NHS to other adults, and children, with the condition.

Vimizim costs around €400,000 a year for an adult and the manufacturers, BioMarin, made an application for it to be funded by the HSE.

As part of that process, Vimizim was assessed by the National Centre for Pharmacoeconomics (NCPE), who decided in December last year not to recommend it for reimbursement.

That blow came just weeks after the HSE, in error, approved their application for Grace to receive the drug under the Treatment Abroad Scheme (TAS).

Now the family's stress and heartache is over as the HSE, which has the final decision on the matter, told them they would provide it for Grace.

"Grace is a very special girl who deserves every chance at life and hopefully from now on her symptoms will improve," Ms Cogan said.

She is not giving up her fight for anyone else with Morquio to also get Vimizim.

"It does not mean that the drug is approved in the Republic but because of the HSE's error in approving it through the TAS they have organised for Grace to have access to the drug," she said.

"I was working when I got the call and I was shaking with total shock and excitement all day.

"It is still very important to me, however, that the drug gets approved within the Republic so that any person, child or adult, diagnosed with this progressive condition has access to treatment that would help them."

A HSE statement said that access to a drug therapy is only available where the specific drug has been approved in this jurisdiction.

But it added: "An individual patient was approved access to drug therapy under the Treatment Abroad Scheme (TAS) in error.

"Notwithstanding this error the HSE gave a commitment to honour the TAS approval as our decision had already been communicated to the family and a reasonable expectation of access to the non-approved drug had been raised.

"The HSE is finalising a solution for the patient on an individual exceptional basis in Ireland and we hope to have a service in place shortly to enable treatment to be delivered."

*This article was written by Elaine Keogh and first published 4 March 2017 on independent.ie. Reproduced here with permission.*

> "Knowing that Grace will be getting the best chance at a better quality of life is just a wonderful feeling

### Grainne Cogan, Grace's mother told the MPS Society:

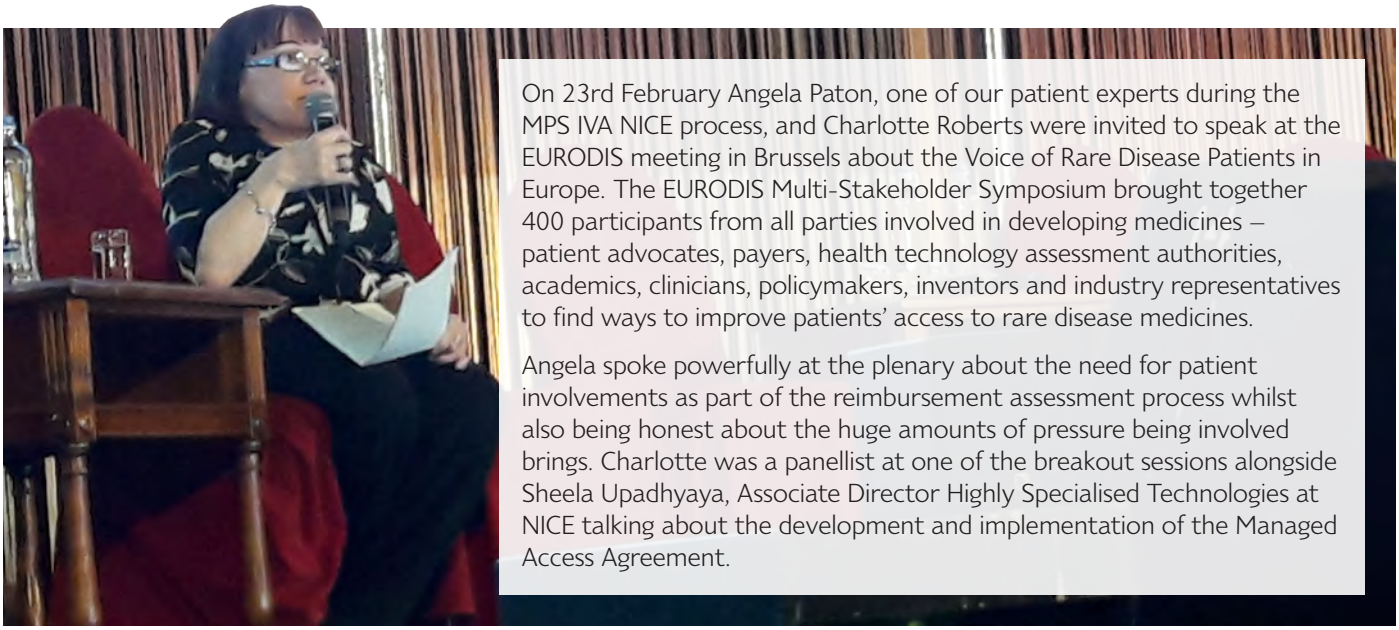
Trying to gain access to Vimizim, the only medication available to help Grace, was, to say the very least, one of the most stressful things I've gone through to date. Obviously Grace's actual diagnosis and all that goes with that was also extremely stressful but at that time nine years ago there was no treatment options for her. Then Vimizim became available and I thought all our prayers had been answered only to realise it was not going to be as simple as it should be to get access to this medication.

I campaigned for over two years in the Republic of Ireland and recently due to an administrative error on behalf of the HSE we have now got approval for Grace to receive Vimizim in Temple Street Hospital, Dublin.

The day I was told was the best day ever! I don't know what it feels like to win the lotto but that's how I felt. Knowing that Grace will be getting the best chance at a better quality of life is just a wonderful feeling. She is such a funny and independent child. Our hope is that her general energy levels and joint pains will improve. She herself just wants to be able to run faster!!

Having a treatment option is so very important and vital with conditions like Morquio.

# International



On 23rd February Angela Paton, one of our patient experts during the MPS IVA NICE process, and Charlotte Roberts were invited to speak at the EURODIS meeting in Brussels about the Voice of Rare Disease Patients in Europe. The EURODIS Multi-Stakeholder Symposium brought together 400 participants from all parties involved in developing medicines – patient advocates, payers, health technology assessment authorities, academics, clinicians, policymakers, inventors and industry representatives to find ways to improve patients' access to rare disease medicines.

Angela spoke powerfully at the plenary about the need for patient involvements as part of the reimbursement assessment process whilst also being honest about the huge amounts of pressure being involved brings. Charlotte was a panellist at one of the breakout sessions alongside Sheela Upadhyaya, Associate Director Highly Specialised Technologies at NICE talking about the development and implementation of the Managed Access Agreement.


## The ISMRD Board of Directors are thrilled to bring the 5th International Conference for Glycoprotein Storage Diseases to Rome!

**Date: 1st–4th November 2017**
**Venue: A. Roma Lifestyle Hotel**
**Registration: via the ISMRD website at www.ismrd.org**

Alessandra d'Azzo has agreed to be the primary investigator for the Scientific Conference. Dr d'Azzo's vision for the conference is:

"In November 2017, the 5th International Conference on Glycoproteinoses will be held in Roma, Italy. We believe that moving this conference from the United States to Europe will raise awareness of these rare lysosomal disorders and increase the global visibility of ISMRD as a family-centric organization that funds scientific research and meetings."

Like earlier ISMRD gatherings, the conference will bring together basic scientists and clinicians from around the world to share with patient families and colleagues their latest discoveries in the areas of glycoproteinoses pathophysiology, investigational and preclinical therapy development, and clinical trials for these rare disorders.

The intent of the conference is 4-fold:

1. to stimulate the exchange of ideas,
2. to form new collaborations among investigators and clinicians with different expertise,
3. to spark interest in these complex diseases among postdoctoral research fellows and graduate students, and
4. to strengthen connections among affected families around the world, so they are better informed and supported.

The ultimate goal of the ISMRD is to foster national and international partnerships to efficiently advance therapies for children and adolescents who suffer from these rare and currently incurable diseases.

## FDA permits marketing of first newborn screening system for detection of four, rare metabolic disorders

The US Food and Drug Administration has permitted marketing of the Seeker System for the screening of four, rare Lysosomal Storage Disorders (LSDs) in newborns. The Seeker system is designed to detect Mucopolysaccharidosis Type I (MPS I), Pompe, Gaucher and Fabry. It is the first newborn screening test permitted to be marketed by the FDA for these disorders.

"The Secretary of HHS recently added Pompe and MPS I to the list of routine recommended newborn screening programs and it is anticipated that additional states will begin requiring use of screening tests to detect these disorders," said Alberto Gutierrez, Ph.D., director of the Office of In Vitro Diagnostics and Radiological Health in the FDA's Center for Devices and Radiological Health. "Accurate screening tests will help with early detection, treatment and control of these rare disorders in newborns, before permanent damage occurs. That's why availability of LSD screening methods that have been assessed for accuracy and reliability by the FDA are so important."

Several states currently mandate LSD screening in all newborns, including Illinois, Kentucky, Michigan, Missouri, New Jersey, New Mexico, New York, Ohio, Pennsylvania and Tennessee. However, until today there were there were no FDA-authorized devices for screening of these disorders. Availability of the Seeker System provides laboratories with a screening tool that has been reviewed by the FDA for clinical and analytical validity.

The Seeker System, consisting of the Seeker LSD Reagent Kit-IDUA|GAA|GBA|GLA and Seeker Instrument, works by measuring the activity level of proteins required for healthy lysosomal storage found in dried blood samples collected from the prick of a newborn's heel 24 to 48 hours after birth. The Seeker Instrument is a device that automates the analysis of dried blood spots. Reduced enzyme activity of proteins associated with any of the four LSDs detected by the kit may indicate presence of a disorder. Results showing reduced enzyme activity must be confirmed using other testing methods, such as biopsies, genetic and other laboratory tests.

The FDA reviewed the data for the Seeker System through the de novo premarket review pathway, a regulatory pathway for devices of a new type with low-to-moderate-risk that are not substantially equivalent to an already legally marketed device and for which special controls can be developed, in addition to general controls, to provide a reasonable assurance of safety and effectiveness of the devices. During this process, the FDA evaluated data from a clinical study of 154,412 newborns in Missouri whose dried blood samples were tested for protein activity associated with MPS I, Pompe, Gaucher and Fabry. Efficacy was determined because the system was able to accurately identify at least one of each of these four LSDs in 73 of the screened newborns.

Risks associated with use of the screening system include false negative findings. As part of this study, the Missouri State Public Health Laboratory conducted active surveillance of four of the state's metabolic clinical centers for new diagnoses of these disorders. The state laboratory's surveillance activities extended 15 months following the study's completion to determine cases of false negatives that had not been identified during the study. No false negative results were identified either through the study or the state's 15-month surveillance program.

The Seeker System was created with funding from the Small Business Innovation Research program in National Institutes of Health's Eunice Kennedy Shriver National Institute of Child Health and Human Development. It is manufactured by Baebies Inc., located in Durham, North Carolina.

The FDA, an agency within the U.S. Department of Health and Human Services, promotes and protects the public health by, among other things, assuring the safety, effectiveness, and security of human and veterinary drugs, vaccines and other biological products for human use, and medical devices. The agency also is responsible for the safety and security of our nation's food supply, cosmetics, dietary supplements, products that give off electronic radiation, and for regulating tobacco products.

Unfortunately new-born screening for MPS diseases is not on the horizon in the UK or Europe.

## Canadian researchers use gene therapy to treat patient with Fabry Disease

A team of Canadian physicians and researchers is believed to be the first in the world to have used gene therapy to treat a patient with Fabry disease.

In the trial, researchers collected a quantity of a Fabry patient's own blood stem cells then used a specially engineered virus to augment those cells with copies of the fully functional gene that is responsible for the enzyme. The altered stem cells were then transplanted back into the patient on January 11, 2017.

"It's too soon to say whether this therapy will ultimately be a long-term treatment for Fabry disease but, based on the success of animal trials, we are hopeful there will be a benefit to patients"

says Dr. Aneal Khan, the Alberta Health Services medical geneticist and member of the Alberta Children's Hospital Research Institute, Cumming School of Medicine, University of Calgary, who is leading the experimental trial in Calgary.

Dr. Jeffrey Medin, a researcher with the Medical College of Wisconsin and the project's principal investigator, says it will be months before the researchers are able to measure whether they've made an impact.

"This experimental trial nevertheless marks a major step forward in treating inherited genetic diseases in adults," says Dr. Medin. "It is very promising that we were able to engineer the complex logistics of such a trial and that the procedure itself seems to have been well-tolerated."

For more than 10 years, Darren Bidulka has had to set aside two hours every two weeks for enzyme replacement therapy, which helps his body break down Gb3 and is currently the only form of treatment.

"It's somewhat disruptive having to arrange your life around a biweekly infusion schedule, but compared to many people with Fabry disease, I've been fortunate," says the 48-year-old Calgary patient who underwent the gene therapy. "There are day-to-day challenges but, other than that, my quality of life is very good. A long-term treatment for Fabry disease would be fantastic, but I don't want to create any false hopes for myself or others at this preliminary stage," Bidulka says. "If my involvement in the research helps shed more light on the disease and brings doctors closer to an ultimate cure, then it will have been worth it."

The treatment, which has been approved by Health Canada for experimental purposes, is also believed to be the first trial in Canada to use a lentivirus in gene therapy. In this case, the specially modified virus was stripped of its disease-causing capability and augmented with a working copy of the gene that's responsible for the missing enzyme.

"We've proven this works in mice but, of course, successful animal trials don't always mean successful human trials,"

says Dr. Medin, who has been working on the project for more than 20 years. Adds Dr. Khan:

"Even some improvement in enzyme levels could brighten the long-term outlook for these patients and lead to a better quality of life."

Source: www.albertahealthservices.ca/news/releases/2017/Page13691.aspx

# Research & treatment

## Supporting diagnosis and understanding of MPS III

It's been over 50 years since Dr Sanfilippo first described MPS III, yet there are still many unanswered questions about the disease. Families often face several years of doctor and hospital visits before MPS III is diagnosed or even suspected.

MPS Commercial will soon be contacting our members to take part in a unique study that will explore MPS III from the family's perspective. They will be looking at how your child's symptoms developed over time and which healthcare professionals were involved both before and after diagnosis. Importantly, they will be identifying both healthcare professionals that are suspecting and diagnosing MPS III and those that may not recognise the signs of an MPS disease.

Through an understanding of both the pattern of early symptoms and the medical professionals who are seeing the children at an early age, the need for information to support both families and healthcare professionals in obtaining a diagnosis can be identified.

The study will also give pharmaceutical companies currently involved in developing therapies key knowledge to support their clinical trial programmes. For more information see the MPS Commercial pages at the back of this issue and look out for your letter of invitation to take part in the coming weeks.

Key facts

- One of the largest MPS III surveys undertaken
- Will survey families from 7 European countries
- First study to track the journey to diagnosis of MPS III
- Sponsored by multiple companies involved in developing therapies
- Collaboration with European MPS Societies to collect information from local families

## Life sciences roundtable briefing

I was in San Diego at the World conference on Lysosomal Storage Diseases when an invitation arrived from Mr John Simmons, Minister Counsellor for Commercial Affairs at the Embassy of the United States of America requesting the pleasure of my company at a life sciences roundtable breakfast briefing on Thursday March 16th 2017.

The day came and this roundtable provided a unique opportunity to discuss the post-Brexit business environment for small US pharmaceutical companies facing uncertainty regarding the future regulatory system for clinical trial arrangements for the licensing of new therapies for rare and ultra-rare diseases in the UK. This meeting came a little over 12 hours after NICE and NHS England announced their devastating response to their public consultation on 'Access to new medicines' that includes the 'highly specialised technologies' for MPS, Fabry and related lysosomal storage diseases.

The discussions at the American Embassy started with how the current operating environment could be developed for all parties, most importantly for patients with ultra-rare diseases, to ensure the UK remains a world leading destination for investment by US based life sciences companies.

The key issues considered were :

- Will the UK, and England in particular continue to be the central lynch pin for clinical trials for ultra-rare diseases in Europe?
- Will the European Medicines Agency (EMA) have a role in appraising new orphan drugs for marketing approval in the UK post Brexit?
- Will the UK continue to be able to attract and recruit highly skilled workers from across the EU to work in the UK without restrictions?

The group of nineteen including representatives from the US Commercial Service, the pharmaceutical industry (BioMarin, PTC, Alexion), patient organisations (MPS Society, Action Duchenne, Tuberous Sclerosis) and Mr Daniel Zeichner, Member of Parliament for Cambridge all however agreed that the first priority was to urgently address the devastating outcome of the NHS England and NICE consultation because if reimbursement of ultra-orphan drugs stops in England, US pharmaceutical companies will no longer invest in clinical trials in the UK and the impact of Brexit will be of no consequence.

# News from the pharmaceutical industry

**A round up of the latest information from the pharmaceutical companies working in MPS and related diseases**

## MPS I

### ArmaGen

ArmaGen are conducting a clinical trial to test the safety and determine a well-tolerated dose of an investigational treatment AGT-181 in people with attenuated MPS I (Scheie and Hurler-Scheie diseases). AGT-181 is an investigational enzyme replacement therapy (ERT) designed to treat both the body-related and central nervous system-related symptoms and complications of MPS I. Currently approved treatments for MPS I are unable to penetrate the blood-brain barrier, a filter that protects the brain from harmful substances like toxins and bacteria but allows vital substances like insulin to cross from the blood into the brain. AGT-181 is designed to cross the blood-brain barrier in the same way insulin does.

A Phase I trial in adults with attenuated MPS I (Hurler-Scheie and Scheie syndromes) to test a new drug in a small group of patients to evaluate the drug's safety, identify potential side effects, and determine a dose of the medication for further testing is underway in the USA.

### Sangamo Biosciences

Sangamo Biosciences through its EMPOWERS clinical research investigational study is trialling SB-318, a gene therapy called genome editing, as a potentially lasting treatment for MPS I. This study which involves a single transfusion of the investigational gene therapy is currently taking place in the USA. Clinical trial participants must be over 18 years. They may be on ERT and can continue receiving ERT whilst on the study. Participants may also have received haematopoietic stem cell transplant (HSCT) in the past.

## GM1 Gangliosidosis

### Lysogene

Lysogene's GM1 program includes the development of an AAV based treatment for GM1 Gangliosidosis. In February 2015, Lysogene entered into a strategic collaboration with the University of Massachusetts Medical School (UMMS) and Auburn University (AU) in the USA. Through this collaboration, Lysogene, UMMS and AU will develop IND-supporting preclinical studies in GM1 Gangliosidosis using AAV gene therapy technology. The collaboration will combine Lysogene's outstanding translational and clinical expertise in gene therapy for CNS disorders with the unique preclinical expertise and infrastructure of UMMS and AU to design and test innovative AAV-based gene therapy approaches to treat GM1-gangliosidosis.

## MLD

### Lentiviral haemopoietic stem-cell gene therapy in early-onset metachromatic leukodystrophy: an ad-hoc analysis of a non-randomised, open-label, phase I/II trial (The Lancet Volume 388, No.10043)

This is an ad-hoc analysis of data from an ongoing, non-randomised, open-label, single-arm phase I/II trial, in which enrolled patients were enrolled with a molecular and biochemical diagnosis of metachromatic leukodystrophy (presymptomatic late-infantile or early-juvenile disease or early-symptomatic early-juvenile disease) at the Paediatric Clinical Research Unit, Ospedale San Raffaele, in Milan. Trial participants received HSC-GT, which consisted of the infusion of autologous HSCs transduced with a lentiviral vector encoding ARSA cDNA, after exposure-targeted busulfan conditioning. The primary endpoints of the trial are safety (toxicity, absence of engraftment failure or delayed haematological reconstitution, and safety of lentiviral vector-tranduced cell infusion) and efficacy (improvement in Gross Motor Function Measure (GMFM) score relative to untreated historical controls, and ARSA activity, 24 months post-treatment) of HSC-GT. For this ad-hoc analysis, the safety and efficacy outcomes were assessed in all patients who had received treatment and been followed up for at least 18 months post-treatment on 1 June 2015.

#### Findings

Between April, 2010, and February, 2013, nine children were enrolled with a diagnosis of early-onset disease (six had late-infantile disease, two had early-juvenile disease, and one had early-onset disease that could not be definitively classified). At the time of analysis all children had survived, with a median follow-up of 36 months (range 18–54). The most commonly reported adverse events were cytopenia (reported in all patients) and mucositis of different grades of severity (in five of nine patients (grade 3 in four of five patients)). No serious adverse events related to the medicinal product were reported. Stable, sustained engraftment of gene-corrected HSCs was observed (a median of 60·4% [range 14·0–95·6] lentiviral vector-positive colony-forming cells across follow-up) and the engraftment level was stable during follow-up; engraftment determinants

included the duration of absolute neutropenia and the vector copy number of the medicinal product. A progressive reconstitution of ARSA activity in circulating haemopoietic cells and in the cerebrospinal fluid was documented in all patients in association with a reduction of the storage material in peripheral nerve samples in six of seven patients. Eight patients, seven of whom received treatment when presymptomatic, had prevention of disease onset or halted disease progression as per clinical and instrumental assessment, compared with historical untreated control patients with early-onset disease. GMFM scores for six patients up to the last follow-up showed that gross motor performance was similar to that of normally developing children. The extent of benefit appeared to be influenced by the interval between HSC-GT and the expected time of disease onset. Treatment resulted in protection from CNS demyelination in eight patients and, in at least three patients, amelioration of peripheral nervous system abnormalities, with signs of remyelination at both sites.

Interpretation

Our ad-hoc findings provide preliminary evidence of safety and therapeutic benefit of HSC-GT in patients with early-onset metachromatic leukodystrophy who received treatment in the presymptomatic or very early-symptomatic stage. The results of this trial will be reported when all 20 patients have achieved 3 years of follow-up.

## MPS III

### Abeona Therapeutics

Abeona Therapeutics announced on 1 February 2017 that the first high-dose subject was enrolled in the ongoing Phase I/II trial for ABO-102 (AAV-SGSH). The first-in-man clinical trial uses a single intravenous injection of AAV gene therapy for patients with MPS IIIA (Sanfilippo syndrome type A).

The ongoing Phase I/II clinical trial, which has received FastTrack designation by the FDA, is designed to evaluate safety and preliminary indications of efficacy of ABO-102 in patients suffering from MPS IIIA. Per the design of the trial, subjects in the low-dose and high-dose cohorts received a single, intravenous injection of ABO-102 to deliver the AAV viral vector systematically throughout the body to introduce a corrective copy of the gene that underlies the cause of the MPS IIIA disease, particularly the CNS. Subjects are evaluated at multiple time points over the initial 6-months post-injection for safety assessments and initial signals of biopotency.

### Lysogene

Lysogene's most advanced product is rAAV vector serotype rh.10 carrying the human N-sulfoglucosamine sulfohydrolase (hSGSH) for the treatment of MPS IIIA. Lysogene's gene therapy is delivered directly to the CNS in one neurosurgical procedure. It is hoped that the delivery of the missing SGSH gene provides a permanent source of functional enzyme in the brain that reverses phenotypic abnormalities of CNS neural cells. The recently completed Phase I/II study in four MPS IIIA children (three patients around 6 years of age and one patient nearly 3 at the time of treatment) demonstrated that the gene therapy and neurosurgical procedure is safe and well tolerated, and exploratory efficacy profiles are encouraging.

Lysogene is currently preparing to initiate a Phase II/III pivotal clinical trial with its next generation gene therapy formulation and has a downloadable 'Family Guide to Understanding Lysogene's Gene Therapy Approach for MPS IIIA' in English on its website www.lysogene.com/pioneering-science/pipeline/

### BioMarin Pharmaceutical

BioMarin is currently recruiting to both its Prospective, Observational Study of Mucopolysaccharidosis Type IIIB (MPS IIIB) and Phase I/II Open-Label Dose-Escalation Study to Evaluate the Safety, Tolerability, Pharmacokinetics and Efficacy of Intracerebroventricular BMN 250 in Patients with MPS IIIB, Sanfilippo Syndrome Type B. Additional information on these two studies can be found on www.clinicaltrials.gov

### Alexion

We understand that Alexion bought out Synageva in an $8.4 million deal a little less than two years ago, the big attraction was Kanuma, a rare disease drug for LAL D, that went on to market approval. However, there was one notable pipeline therapy, SBC-103 for rare cases of Sanfilippo B, that drew some attention for the valuation. In February 2017 Alexion decided to reduce its investment in SBC-103. We understand that patients currently enrolled in the Phase I/II trials will continue to receive SBC-103 but that no additional Alexion studies are planned. Alexion says it will reassess the value of this asset on a go-forward basis.

### Shire

In August 2016 Shire announced termination of its MPSIIIA enzyme replacement therapy clinical trial. This affected five children and their families in the UK and many others globally. Published statements from Shire say 'The Phase IIb study of SHP610 in paediatric patients with early stage Sanfilippo A did not meet its primary endpoint of slowing the cognitive decline in patients'. Shire has also stated that 'it will now terminate all clinical trials of SHP610 and plans to publish the results of the SHP610 program "for the benefit of the Sanfilippo community."

### Sobi

On the 19 October 2016 the European Commission granted SOBI003 orphan designation for the treatment of MPS IIIA. Sobi is in the late stages of preclinical development of SOBI003. Preclinical studies to date with repeated systemic infusions have demonstrated efficacy in reducing substrate levels in the brain and signs of disease modifying effects. Sobi is preparing for clinical studies, which it plans to start in 2018. Clinical studies will focus on exploring the safety and efficacy of SOBI003 in MPS IIIA patients. SOBI003 is a chemically modified variant of a recombinant human sulfamidase product candidate intended as an enzyme replacement therapy to reduce heparan sulfate storage materials in affected cells. SOBI003 is taken up by cells and transported into the lysosomal compartment where heparan sulfate is degraded. The modification of the molecule results in an extended half-life, which may enable transport of SOBI003 over the blood brain barrier and facilitate uptake of SOBI003 in to the brain.

# Interview study to learn more about progression and quality of life in children and caregivers affected by Sanfilippo Syndrome Type B

The impact of the progression of Sanfilippo Syndrome Type B (Mucopolysaccharidosis Type IIIB, MPS IIIB) on the quality of life for children and their families is not well understood. To date there is little evidence on how the disease impacts children and the stress it can cause caregivers. We also know little about the quality of life of children diagnosed with this disease.

The aim of this study was to:

- Understand the relationship between progression milestones of Sanfilippo Type B and the quality of life of the affected children and families.
- To find out what are the best measures of quality of life in children diagnosed with Sanfilippo Type B.

The study was split into two parts. In Part 1, caregivers were asked about their experience of caring for a child diagnosed with Sanfilippo Type B. Caregivers were also asked to complete questionnaires used to measure quality of life of children affected by Sanfilippo Type B, as well as one questionnaire used to measure stress in caregivers.

After the caregivers had completed the questionnaires they were asked to provide feedback on how well they thought the questionnaires captured their experience and their child's experience of Sanfilippo Type B.

In Part 2, we asked caregivers to provide feedback on whether they agreed or disagreed with the results from Part 1.

## How was Part 1 of the study conducted?

Researchers from the UK MPS Society visited caregivers at their homes to talk about their experience and what they thought about the questionnaires. Researchers also talked with some caregivers over the phone.

Researchers asked caregivers to complete the Vineland Adaptive Behaviour Scales to get an understanding of how the affected child was doing at the moment of the study.

The Vineland measures behaviour in terms of communication, daily living skills, socialisation and motor skills. For example, how well a child can communicate or move around.

The caregivers also completed the following quality of life questionnaires. They were asked to write down any comments and feedback they had at the same time as they completed the questionnaires.

- Infant-toddler quality of life scale (ITQoL)
- Child Health Questionnaire Parent Form (CHQ-PF50)
- Sanfilippo Behavioural Rating Scale (SBRS)
- Parenting Stress Index (PSI-4 short form)
- PedsQL Family Impact Module (PedsQL)
- Children's Sleep Habits Questionnaire (CSHQ)

After the questionnaires had been completed, the researchers asked caregivers what they thought about these questionnaires in terms of:

8 caregivers took part in Part 1 of the study, 6 were female, 2 were male

- How easy or difficult it was to understand and answer the questions.
- How well the questions related to their personal situation and experience.
- What they thought about the time period the questions referred to. For example, some questions asked people to report how things were "currently" or "in general". Other questions asked people to report how things were a week, a month, or a year back.
- To point out the questions that were difficult to understand and to answer.
- To let the researchers know if there were issues they thought should be included in the questionnaires.

For example, something that influences their own child's quality of life that was not asked about in the questionnaires.

## What were the results of Part 1?

Eight caregivers took part in this part of the study, six were female and two were male. The average age of the people participating was 39 years (range was 26-51 years). The average age of the children with Sanfilippo Type B was 8 ½ (ranged 5-10 years).

The Vineland questionnaire showed that all children had difficulties, including problems

with: Physical functioning (e.g., caring for self, caring for home), Communication (e.g., talking, listening, writing), and Behaviour (e.g., playing, using large muscles).

The Vineland questionnaire also calculates an adjusted age based on the child's current abilities. This is called the "age equivalent" score.

- The average age equivalent score of the caregivers' children was 16 months ( i.e., 1 year and 4 months, with a range of 10-21 months).
- When comparing this score to the actual average age of the children, which was 8 years and 6 months, we clearly see the severe impact Sanfilippo Type B has on these children's lives.

## Questionnaire Feedback

**ITQoL** (only two caregivers completed this questionnaire)

- Caregivers found most areas relevant to their child's Sanfilippo Type B. These included overall health and satisfaction with development.
- Both caregivers said that amount of worry they experience has the greatest impact on their child's quality of life.

**CHQ-PF50**

- Most areas relevant in terms of child's experience. These included physical functioning, family activities and behaviour.
- Some caregivers felt bodily pain/discomfort was not relevant as it was difficult to judge their child's discomfort (e.g., as they were not able to communicate with their child about how the child feels).

**SBRS**

- All areas of this questionnaire were seen as relevant to their own child's experience, including communication, temper tantrums and behaviours.
- Some caregivers thought there should be a 'Not Applicable' response option included in the questionnaire.

**PSI-4 short form**

- Due to the nature of the questions (e.g. questions about feeling frustrated with the child), most caregivers felt the questions of this questionnaire made them feel bad and that it caused negative emotions when they were asked to answer these – e.g., "Made you feel exposed".
- Caregivers reported that the stress they experienced day to day made them tired, frustrated, and irritable.

**PedsQL Family Impact Module**

- Caregivers said that the areas they personally were most affected by since their child's diagnosis with Sanfilippo Type B were physical and emotional functioning.
- Caregivers said that the areas their family have been most affected by since the diagnosis were daily activities, social functioning and worry.

**CSHQ**

- Most caregivers said that sleep duration had the greatest impact on the quality of life of their affected child.
- The questions on sleep anxiety, parasomnias, and daytime sleepiness areas were not seen as relevant by caregivers as they were not part of their experience with Sanfilippo Type B.

## How was Part 2 of the study conducted?

Researchers from a health research company, ICON, spoke to some caregivers on the phone about whether they agreed or disagreed with the results we found in Part 1 of the study.

The caregivers were given a written summary of the results from Part 1 for them to read before the call. They could also refer to this during the call, if needed.

The average age of the caregiver's children with Sanfilippo Type B was 7 years in Part 2 of the study

During the call, the caregivers were asked about the developmental milestones their child had reached and abilities their child had gained or lost as a result of Sanfilippo Type B. We also asked about care priorities and how these priorities had changed over time.

## What were the results of Part 2?

Three caregivers took part in this part of the study, 2 were female and 1 was male. The average age of the people participating was 37 years (range 26-45 years). The average age of the caregiver's children with Sanfilippo Type B was 7 years (range was 5-10 years).

Caregivers generally agreed with the findings from Part 1:

- Similar to the Part 1 results, the question about the Family's ability to get along with one another question from the ITQoL was not seen as relevant.
- The CHQ-PF50 questions related to Bodily pain/ discomfort were not seen as relevant to their own experience with their child, and the questions related to behaviour were reported to capture the greatest area of impact.
- The caregivers also fed back that the section of the SBRS questionnaire that captured the greatest area of impact on the quality of life of their affected child was communication. They also agreed that the questionnaire should include 'Not Applicable' response options.
- They also agreed that the stress they had experienced personally had made them feel tired, frustrated, and irritable. The negative emotional impact of the questions of the PSI-4 short form questionnaire was also mentioned on the calls.
- In the PedsQL questionnaire, the caregivers agreed with the Part 1 results that Physical

and emotional functioning were the greatest areas of impact on quality of life. Daily activities was again reported as having been affected the most by Sanfilippo Type B in terms of their family as a whole. The caregivers also thought that the questionnaire should capture reactions of, and impact on, siblings of children diagnosed with Sanfilippo Type B, which is currently not included in this questionnaire at all.

- As in Part 1, caregivers did not think that questions from the CSHQ questionnaire that related to sleep anxiety and Parasomnias were relevant. They also thought that the questionnaire should include questions about how medications affect the affected child's sleep habits.

However caregivers disagreed with some findings from Part 1:

- All three caregivers in Part 2 did not think that the ITQoL questions about the Amount of worry caregivers experience had the greatest impact on quality of life.
- All three caregivers on the Part 2 calls did not think that physical functioning, as measured by the CHQPF50, was a problem as their child was "very mobile"
- One caregiver also said that temper tantrums questions from the SBRS were irrelevant. This was mainly because the caregiver did not see their child's challenging behaviour as temper tantrums per se.
- All caregivers said that the stress they experienced did affect the child, although one caregiver reported it was not an issue for them personally (PSI-4 short form)
- In Part 1 some caregivers thought that the questions of the PedsQL were worded too negatively.

However, the three caregivers who participated in Part 2 did not agree. They instead said that the questionnaire would be unlikely to upset responding caregivers.

- Unlike the caregivers in Part 1, the three participants in Part 2 said that the sleep duration questions of the CSHQ did not have the greatest impact on quality of life of their child. They also thought that the daytime sleepiness questions were relevant, although one caregiver reported that it was not relevant to their particular situation.

In terms of milestones the Part 2 caregivers' affected children had reached, all three could walk, eat unaided, and swallow without difficulties. Two children could also communicate using a few words. However, none of the children could use the toilet unaided, or write, or read.

All 3 caregivers reported finding it difficult to remember the exact time point when their child lost certain abilities, for example, because it "does not stop all at once".

One caregiver described how certain abilities come and go; for example, the use of certain words. One caregiver disagreed, and was not sure whether their child was losing or gaining skills, or if "he always had the ability to say that word, he's just chosen not to until that particular point in time".

Caregivers discussed a number of different care priorities, including: safety of their child, their child's health, and meeting their child's physical needs. In addition, caregivers discussed that the impact of Sanfilippo Type B also has a profound impact on siblings which was missing from the questionnaires.

Care priorities, such as feeding and toileting, generally changed over time as the Sanfilippo Type B progressed. One caregiver reported no particular changes in the care priorities, but felt caring had become more difficult as their child grew "stronger and heavier".

## Conclusion

Overall, this study highlighted that whilst the questionnaires we tested were predominantly relevant and useful to capture quality of life of children and caregivers affected by Sanfilippo Type B, there are some missing aspects and some concerns with certain questions. The relevance of particular questions differed between individual families' and some important areas, such as the impact Sanfilippo Type B could have on siblings, were missing all together.

Wording of questionnaires could be adapted to be more considerate of the potential emotional impact on caregivers from being asked sensitive questions. The abilities that affected children gain or lose over the course of the disease also differed between families, and caregivers found it difficult to report the exact time points this happened; something that some questionnaires require. Care needs, such as safety, health and physical care of affected children could vary in priority over time and could also become increasingly difficult to manage as affected children grow up.

Some important areas, such as the impact Sanfilippo Type B could have on siblings, were missing

## Acknowledgements

The study team would like to thank the caregivers who took part in this study. We would also like to thank the MPS Society, who helped to recruit caregivers and conduct interviews and BioMarin who sponsored this study.

# MPS Society awards two new research grants

Assessing the Bioviability of Genistein in Patients with MPSIII, IVA and VI

Dr Gisela Wilcox - Consultant in Metabolic Medicine

The Mark Holland Metabolic Unit, Salford Royal Foundation NHS Trust

Grant awarded £12,000

## Rational for the project

Project to be carried out by Bhawna Sharma an Intercalating medical student in Mres Medical Sciences

Genistein is a naturally occurring compound currently under investigation as a form of substrate reduction therapy use in the treatment of patients with MPS. This naturally occurring compound in the family of plant chemicals (phytochemicals) known as isoflavones, is found in soy and other legumes. Pharmacological doses, allowing significant quantities to cross the blood brain barrier, have shown very promising results in MPS III rodent studies. Human clinical trials are presently underway, but initial studies have yet to translate the promising findings reported in rodents to patients with MPS III.

There is at present very limited data regarding the blood levels of genistein, even in MPS patients given pharmacological doses. Current data suggest that patients with MPS III given genistein achieve lower levels in blood than do mice. This may be due to altered metabolism taking place in the liver, and /or intestine, resulting in lower levels of 'free genistein' (which has a beneficial effect), as opposed to the levels of conjugated genistein (which has little benefit until made free). Also, we do know how levels in patients with MPS compare with those in the general population not suffering from MPS; antibiotic use and altered liver metabolism may result in differences in MPS patients.

Many families around the world, awaiting clinical trial results, are currently using soy extract as a source of genistein. Although the amount of genistein that can be consumed from soy is much lower, soy also contains daidzein (an isoflavone) and many other phytochemicals. Daidzein, especially in combination with other natural phytochemicals, has also been shown to decrease GAG synthesis in vitro (Kloska et al 2011). Preliminary studies with SoyFem® soy extract containing genistein and daidzein, have shown modest decreases in total urinary GAGs,(Malinova et al 2012) when given in doses10 fold higher than seen in traditional isoflavone–rich diets. However blood levels of isoflavones were not reported in this study. There is therefore limited knowledge of isoflavone blood levels from consuming soy extract and/or soyfoods in MPS patients.

While there is at present, controversy and uncertainty about the effects of high dose pure genistein

in changing the course of MPS III in humans, there is much scientific literature, accumulated over the last 30 years, on the metabolism and biological effects of soy isoflavones and other plant compounds in the general population at levels of intake around one tenth of the levels being used in recent human trials in MPS. These include effects on markers of inflammation, cardiovascular disease and bone health all of which may be relevant for MPS patients, especially into adulthood. It is therefore not only possible that isoflavones may modify GAG production but they might also act at other steps in the disease process.

Any beneficial effect of isoflavones in MPS patients could be particularly useful as an inexpensive, safe, and readily available complement to existing therapies in a cost-constrained healthcare environment.

## The aims of this study are to:

1. Find out the blood concentrations of total and free isoflavones (including genistein & daidzein) after consuming 250ml ml soymilk (or soy dessert) twice daily for 4 weeks and see how this compares with control subjects
1. See if this level and duration of intake has any effect on urine GAG excretion.
1. Find out how altered body composition and body size in MPS patients might affect the blood concentrations of isoflavones.

Freeze blood samples for later testing of markers of bone turnover, inflammation and vascular health, when further funding/resources permit (this will be requested in the same Ethics Application).

Grant Awarded £47,650

# An In-Depth Characterization of Fabry Patients with Cardiac Devices to Predict Risk of Malignant Arrhyhemia and Sudden Cardiac Death

Dr Tarekegn Geberhiwot – Consultant in inherited metabolic disorders

University Hospitals Birmingham NHS Trust

## Case study and rational for the project

A 51 year old man with known Fabry disease (FD) on long-term enzyme replacement therapy of 10 years died at home suddenly and unexpectedly. He had significant heart disease related to FD and he had a permanent pacemaker (PPM) implanted for atrio-ventricular delay in 2013. Post-mortem interrogation of his pacemaker revealed that the cause of death was ventricular tachycardia. Following his death I met his wife and three sons to discuss the circumstances of his death. His wife questioned why the pacemaker did not stop his death and I explained that PPM was to prevent slow heart rhythm abnormalities while he was killed by a fast rhythm. This would have required a different type of pacemaker called an intracardiac defibrillator (ICD) but there was no clear evidence before his death to suggest he needed this. If an ICD had been implanted, he would be alive.

In our UK cohort of Fabry patients heart disease is the main manifestation and leading cause of death. Significant numbers of our FD patients have a heart device of the PPM type and fewer have an ICD. Patients with PPMs and ICDs are an invaluable resource to promote understanding of risk, since modern devices have not only a treatment function but are also able to monitor heart rate and rhythm continuously over a long period of time.

Although data from ours and German centre studies suggest that there is a small but important risk of sudden cardiac death (SCD), there are currently no tools for risk stratification to determine appropriate implantation of ICD, which can help to prevent SCD. In other genetic heart disease such as hypertrophic cardiomyopathy there is a well-defined risk stratification tool to predict a risk of SCD and appropriate intervention. There are no similar data to guide practice in FD patients. We have performed a systematic review to identify variables that will have an association with malignant arrhythmia and SCD in FD, including male sex, age>40years, QRS duration>120ms, LVH, left atrial size>40mm and the presence of late enhancement on magnetic resonance imaging (CMR), tachyarrhythmia and Mainz-severity score(1-3).

Pilot data from Birmingham centre indicate that 13 (15%) of our patients have had a PPM or ICD implanted.

| Patients with devices | No of Patients | |
|---|---|---|
| PPM | 5 | 4 males, 1 Female |
| ICD | 8 | 8 males, 0 females |

## Aims

1. Identify the indications that have been used to justify implantation of a loop recorder/PPM/ICD in FD patients in 3 national UK centres;
2. Obtain monitoring and device data from patients who have had a cardiac device implanted to examine arrhythmia burden, device use and discharge rate.
3. Use these to define arrhythmia risk and produce a common protocol.

## Methods

Screen FD patients from 3 or more adult LSD national centres in Birmingham, London and Manchester. Based on internal communication, the expectation is that this will identify around 50 patients who have had a device implanted. The vast majority of FD patients cardiac device are fitted by the cardiology team at the patients local hospitals and the LSD centres have no direct access to the wealth of continuously monitored cardiac data. These device data are a valuable resource and if all data are combined, could yield vital information to predict arrhythmia and SCD.

We will also gather data from diseased Fabry patients who died from cardiac cause in the last 15 years. This can be gathered from both the LSD centres and the English national death register held in University Hospital of Birmingham. We will employ a cardiology research fellow for a year who will liaise between the three LSD centres and individual patient's hospital.

## Direct benefits of this project to Fabry patients

A) Immediate Benefits

1. Identify parameters which will highlight a group of high risk FD patients for SCD;
2. Use these parameters to re-examine the whole cohorts in each centre to identify cases at risk who can then be discussed at multi-disciplinary team with a view to device implantation. (outcomes would need to be further evaluated prospectively)

B) Long Term Benefits

Prospective randomised trial of implantation of implantable loop recorders in patients with and without risk factors to validate risk score for implementation internationally.


# 'What matters to you?' day 6 June 2017

## #WMTY17

'What matters to you? day is being held on 6 June 2017, please save the date and plan now for how you might join in on the day.

The aim this year is not only to encourage and support more meaningful conversations, but also to focus on the action that needs to happen in response to these conversations to deliver the care and support people really need and want.

We know from experience and evidence that the effect of focusing on what really matters to people can lead to improvements in the quality and effectiveness of what we do. Having a better understanding of what is important to people also helps develop relationships that provide the support and help people need to achieve optimal health and wellbeing.

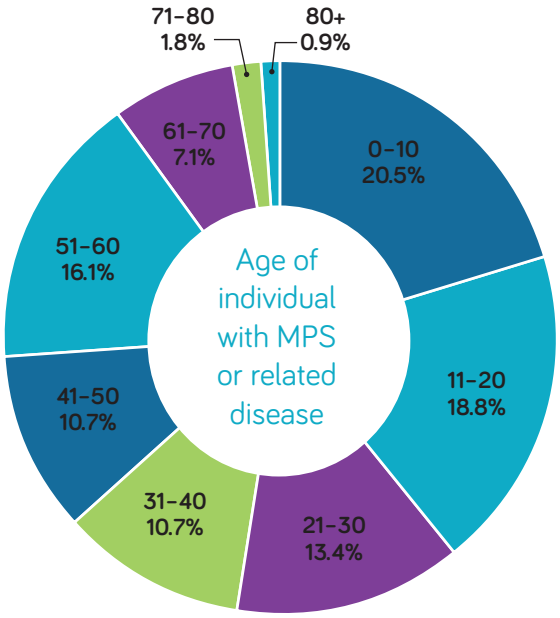
Last year, 'What matters to you?' day had more than 520 health and social care teams across Scotland making a special effort to have more person-centred conversations with the people they work with. In addition to this, more than 100 teams from 13 countries joined our Scottish initiative. You can read more about it in our 'What matters to you?' day 2016 report at: www.whatmatterstoyou.scot

'What matters to you?' day is being supported by the Cabinet Secretary for Health and Wellbeing, the Minister for Public Health and Sport and the Minister for Mental Health and Wellbeing. They will be getting involved on the day as well as sharing messages of encouragement and good practice in the run-up to 6 June.

The Scottish Government and Healthcare Improvement Scotland's person-centred health and care team will also be supporting health and social care organisations practically to participate in the day by providing advice and resources through the website at www.whatmatterstoyou.scot.

To ask questions or find out more, please contact the person-centred health and care team at hcis.personcentredscot@nhs.net

# Member's Survey




Here at the MPS Society, we constantly monitor the services and support we offer to our members to make sure that we are meeting the needs of those who access them. As part of this monitoring process, we regularly ask our members for their input.

In November 2016 we emailed around 600 of our members inviting them to complete an online survey on the services and support that the MPS Society offer.

We often hear that people are 'fed-up' completing surveys, then never hearing anything more about the results or what they have been used for – so here at the MPS Society we want to try to break this cycle.


Which region do you live in?

## And the results are in...

Now, we can't promise anything as dramatic as this years 'Best Picture' announcement at this year's Oscars, but we hope you will find the results interesting - we certainly did!

### Response rate

We were delighted to receive our best response rate to date, with 112 of you sending in your responses, which is approximately 20% of everyone we sent it out to.

### All about you

The first part of the survey gathered data on the age of the responder and where in the country they lived.

### Age of responders

We asked about the age of individual with MPS or related disease. These data are important to help us to improve our services across all age groups. We received 112 responses to this question.

Whist the majority of responses (39.3%) related to babies, children, teenagers or young adults with an MPS or related disease, our oldest responder was an octogenarian!

### Where do you live?

We asked for information on the region you live in, as some of the grants we apply for are region specific. We received 112 responses to this question.

Most responders were from the South West; followed by the South East; and Yorkshire and the Humber. We only had one response from Northern Ireland, so we are hoping for more voices from this neck of the wood in our future surveys so we can present a balanced viewpoint from this region.

## Our Advocacy Support Service

The second section of the survey focussed on the support that our Advocacy Support Service provide to our members.

### Rating the support you have received

We asked our members to rate the support they had received from our Advocacy Support Service as: 'Excellent', 'Good', 'OK',' Poor', 'Really Poor', if they had accessed our service; or 'No experience' if they hadn't.

We are pleased to report that the overwhelming majority of the responders who had been in contact with our Advocacy Support Service felt that they had received an 'Excellent' or 'Good' level of support, across all of the areas listed.

| | Excellent or good (%) |
|---|---|
| Education (School) | 89.7 |
| Social care | 85.2 |
| Housing | 84.2 |
| Advice | 83.3 |
| Information | 82.4 |
| Benefits (PIP, DLA, Housing, etc.) | 82.2 |
| Health | 80.9 |
| Emotional support | 78.7 |
| Equipment and adaptation | 75.0 |
| Education (Further Education) | 70.0 |
| Transition | 65.4 |
| Independent living | 63.6 |
| Employment | 53.3 |

**The advocacy service is committed to improving its service and access for young people and adults and will be developing its resources over the next few years.**

### Frequency of contact

When asked how frequently our members would you like to be contacted by the Advocacy Support Service, the majority of respondents (44.3%) said that they would prefer to contact the MPS Society when they need something rather than at set times (e.g. weekly, monthly, twice a year or yearly).

**44.3%**
of you would prefer to contact us when you need something

## What we do and what could we do?

The third section of the survey concentrated on the services the MPS Society currently offer and what you would like us to offer in the future.

### How can we improve?

In this open-ended question we gave you the chance to tell us areas how we could improve on areas we currently support. We received 24 suggestions, which included:

- Update your advice on travel insurance; the insurance companies currently listed do not support MPS disorders

**Many companies do not recognise MPS conditions and use general medical screening tools when assessing eligibility and risk. This is why insurance varies and why although one individual may get insurance through a listed company another may not.**

### Your wish list

When asked *'What other services would you like the MPS Society to offer?'* we received 21 responses. These included:

- Bereavement support for siblings

**Unfortunately this is an area of support that we cannot currently provide directly; the advocacy team would be happy to support individuals and families to access support locally; or through local hospices.**

- An online forum where information could be shared
- Regional point of contact and meetings

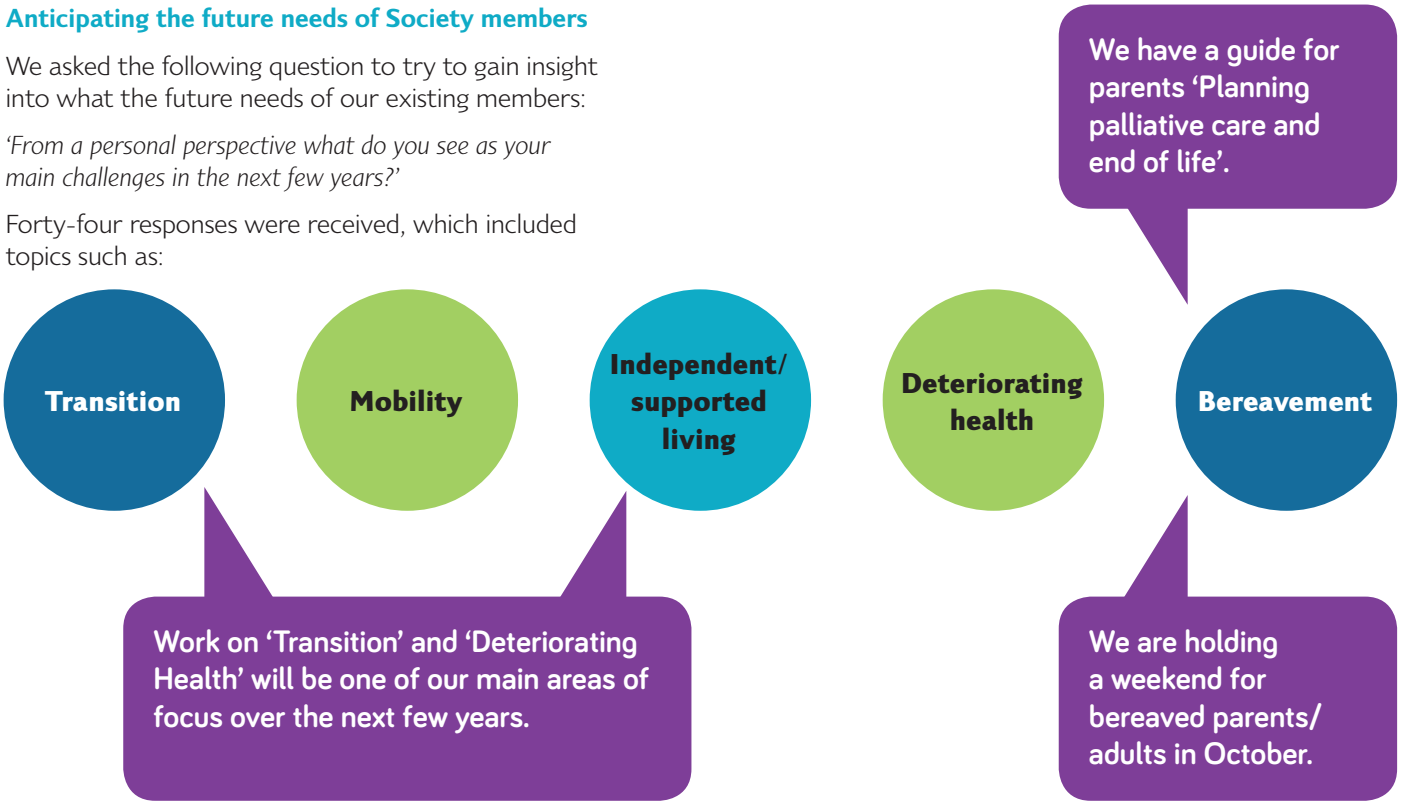
**Regrettably, due to data protection laws, local contacts are not possible. We can, however, link people through our befriending scheme and we hold regional events, subject to funding, to facilitate individuals and families linking together. Check the magazine and e-shots for upcoming events.**

### Anticipating the future needs of Society members

We asked the following question to try to gain insight into what the future needs of our existing members:

*'From a personal perspective what do you see as your main challenges in the next few years?'*

Forty-four responses were received, which included topics such as:




## Our website

In the final part of the survey, we asked questions relating to the MPS Society website, in terms of what was the reason for your visit; how easy you found it to navigate and what you would like to see included.

Most used our website to view our information on specific diseases and to read our news page (25.4% each); to look up contact information (23.9%); and to find out about forthcoming events (19.7%).

In terms of ease of use of our website on their last visit, 88.1% said that they found it either 'Very easy' or 'Fairly easy' to use.

You gave us some great ideas what you would like to see included, which we are currently looking into. These included:

- Related services which can help members
- Local groups for people with similar problems

88.1%
of you think our website is fairly or very easy to use

## How are the data you collected being used?

We promise you that we haven't filed the data to the back of the filing cabinet! Some have already been put to good use by our fundraising team in their grant applications and trying to secure funding for our family events, such as Lapland and Drayton Manor days.

Other data are helping us plan our future resources and expand that range of services we can offer to our members.

Where specific issues have been identified, we are in the process of following up and updating the information as required.

### What can I do to help?

The short answer is – fill in a survey!

For those of you who did not complete the survey this time round, we are planning to send these out regularly (every 6 months or so), so keep an eye out on your inbox. We'd love to hear your opinion.

To ensure you have a chance of completing the next survey, please can you let us know if you have changed you email address.

Remember, just five minutes of your time can really make a difference. If we don't know what you need or would like from us, or how you feel about the MPS Society we can't put plans in place to change it.

5 minutes of your time can really make a difference to how we change MPS Society for the better


If you are planning a sponsored run this vest is perfect for helping you to raise awareness while you do it!

Made from performance polyester wicking material, our vest promises a comfortable run.

A bargain for £5

A black biro complete with our logo and web address! What more do you need? Only £3.50


Our blue awareness ribbon with the MPS web address in white with a pin badge at the back to fasten to clothing

Just 49p


An eco-friendly way to support the MPS Society! 100% cotton long-handled shopping bag for £3.50


This button badge is perfect for fundraising. Buy one for all your helpers or buy lots to sell at an event.

For only 50p each you might just want some to decorate your favourite bag or jacket!


Look good and start a conversation about rare disease with this brand new stylish beanie in navy blue for £5

## All merchandise is available from mpssociety.tictail.com (plus p&p)


A photo/ memo holder, ideal for your desk which displays the MPS hexagon and our website address.

Yours for just £3


Unisex white cotton short sleeve T-shirt for £5


A metal and enamel pin badge. At 25mm x 20mm it's perfect for a lapel or a bag.

Only £1.50

# Fundraising

Oliver, now seven, and Thomas, three, are holding the reindeer dust that their mum, Natalie, made and sold on for charity. The family raised £152 for the MPS Society. Thanks to dad, Adam for sharing the photo and story with us.

## Quiz me

The Hampden Arms held another Boxing Day quiz in aid of the MPS Society and raised a smashing £180. If quizzes are your cup of tea and you wish you could have joined the Hampden Arms then you are in luck as they're holding another for MPS Awareness Day on 15 May!

## It's a dry year

Matt Patrick didn't stop at dry January this year, in fact he's pledged to go dry for the whole of 2017 to raise funds for MPS Society. He has also recruited his brother, Ben, on his alcohol-free endeavours. Good luck Matt and Ben!

## On your soapbox

We will be watching the Micklegate Run Soapbox Challenge this August after Team Anaplan introduced us to this hilarious event last summer.

Lead drivers, Kane Nelson and Ruth Laird, together with pusher-offers, Peter Lester and Oliver Smithson, built a unicorn-shaped soapbox then raced it through the cobbled streets of Micklegate in the hope of surviving at least one run without falling apart.

They raised money for MPS Society as it is a charity "very close to one of our work colleagues whose son has a condition diagnosed as MPS II Hunters."

The team raised £605 on the day but I am not sure the unicorn survived.

## Secondhand but not second rate

The Marina & Friends Fundraisers shop has raised a further £2,771.50 from the sale of secondhand items in their legendary charity shop in Bristol. The total raised by Marina and her supporters is now at the grand total of £170,020.97.

## Dressing down

Northumbrian Water hold a dress down Friday once a month and raised £239.09 in the one held for the MPS Society.

## Sporting fundraisers

UCL Women's Rugby team organised a sports event fundraiser on behalf of MPS Society in December.

Kate Bovey, one of the organisers, wrote in to let us know how successful the event was: "we had over 120 people attending and many sports teams involved. We are also very pleased with the amount of people who had never heard of MPS before or any of the diseases supported and we were able to explain to them the good work that you do and give them more information. For example, our social media posts have reached over 3,702 people!"

As well as what looks like a brilliantly fun evening and the awareness they raised the UCL Women's Rugby Team also raised £260.

A highly successful seasonal concert was held in Haddenham, Buckinghamshire in December, performed by members of the local Witchert Chorale. St. Mary's Church was filled to capacity and all who attended generously donated to the MPS Society raising an amazing £1721. Jenny and Andy Hardy's son, Rebecca's brother, Matthew, was well known in the village during his 13 years. Jenny, who until recently sang with the Chorale, gave a moving account of life with Matthew, who had MPS II Hunter, and the work of the MPS Society. The popular Witchert Chorale is well known for its wide repertoire ranging from early music to modern jazz settings and all their money is raised for charities...some £30,000 during their 12 years of performing.

Mr and Mrs Chalmers bought MPS awareness ribbons to give out as wedding favours when they got married in March this year. Lindsay shared this lovely picture of them on their day wearing the ribbons.

"Everyone thought it was a lovely idea and a great way to involve my brother in law who has Sanfilippo...everyone wore them"

Photo of Adair family: Dinah, Katherine, Eileen, Paul and Damien

Damien and Dinah Adair celebrated their 25th wedding anniversary on 30th December 2016 with a meal for family and friends in Square Bistro, Lisburn.

"Our niece, Roma Drayne, has Morquio IVA and we value the support that MPS have given Roma and her family for over 20 years. We asked people for donations in lieu of presents and are delighted to say we raised £1000 for MPS.

We're looking forward to the next 25 years!"

# Ways to raise

There are so many ways you can help raise funds for the MPS Society – here are some of the organisations where you can nominate the MPS Society as a beneficiary.

## Much Loved

This free tribute service helps you to remember, commemorate, collect and record donations in memory of your loved one in a very special way. MuchLoved Tributes have a number of features including: beautiful designs, adding stories, photos, videos and a personal diary and you can raise funds for the MPS Society as well.

Visit mpssociety.muchloved.org

## Give a car

Giveacar is a non-profit fundraising organisation that raises money for UK charities by scrapping and selling old cars. They provide a FREE nationwide service which:

- Arranges collection of the vehicle from your home
- Depending on its age and condition, recycles it at an Authorised Treatment Facility or sends it to auction.
- Scrap donations raise up to £100 (depending on the price of metal) while auction cars can raise much more.

To arrange collection of a car, visit Giveacar.co.uk or call 020 7736 4242, quoting MPS Society as your preferred charity.



## Easy Fundraising

Whenever you buy anything online – from your weekly shop to your annual holiday – you could be collecting free donations for the MPS Society with easyfundraising

There are over 3,000 shops and sites on board ready to make a donation, including Amazon, John Lewis, Aviva, thetrainline and Sainsbury's – it doesn't cost you a penny extra!

It's as easy as 1, 2, 3...

1. Head to https://www.easyfundraising.org.uk/causes/mpssociety/ and join for free.

2. Every time you shop online, go to easyfundraising first to find the site you want and start shopping.

3. After you've checked out, that retailer will make a donation to your good cause for no extra cost whatsoever!

There are no catches or hidden charges we will be really grateful for your donations.

## MPS Awareness Day and ebay partner up

This MPS Awareness Day shop with a difference as we have been chosen as ebay's Give at the Checkout charity the week leading up to this special day. From Monday 8th May you will see the MPS Society when you shop on ebay promoted by ebay to encourage donations from all shoppers.

So, if you, family or friends are buying on ebay during this week, you will be able to make a donation when completing your purchase. The donations are paid to a PayPal Giving Fund who then forward 100% of the funds to us.

It's a great way to buy something for yourself and to support the MPS Society at the same time.

The MPS Society will be displayed at the checkout throughout the whole week and donors can choose a donation of £1, £2, £5, £10 or £25.

But don't feel you have to save all your shopping to do in May as you can find the MPS Society and donate via ebay anytime. Just visit charity.ebay.co.uk search for 'MPS Society' and save us as a favourite.

## Cash for clutter

Did you have Spring clean this year? If not, now is the perfect time. Do you have unwanted items and don't know what to do with them? Recycling for Good Causes help us raise much needed funds by recycling donated items that are no longer of use.

Here's where YOU can help!

By taking up a recycling project you can clear out your unwanted items and help raise funds for MPS Society. It's free, easy to do and there's no long term commitment. All it takes is a phone call (free of course). You'll be sent a sack to fill with your unwanted items. Then phone again when you're ready for collection. It's that simple!

We can recycle all of the following, even damaged or broken:

- Jewellery (any material, wearable or broken)
- Watches
- Used stamps
- Gadgets (mobile phones, cameras, game consoles, laptops, Sat Navs)
- Unwanted currencies (all those foreign coins and banknotes from your holidays, no matter how old)

You'll be sent everything you need to start your free recycling project.

Contact Recycling for Good Causes at info@recyclingforgoodcauses.org or on 0800 633 5323

## The Weather Lottery

A leading fundraising lottery whereby you have the chance to support the MPS Society and the chance to win £25,000 lottery jackpot in their weekly draw.

When you join, you'll be given a unique six digit Lucky Number to be entered into the weekly Weather Lottery draw.

If you choose more than one entry, you'll be given a separate Lucky Number for each entry.

The Weather Lottery result is based on the last digit of the Fahrenheit temperature from 6 popular European destinations – Corfu, Istanbul, Tenerife, Innsbruck, Edinburgh and Stockholm. Match two temperatures, you win £2, if you match four you will win £20, if you match five you win £250 and if you match all six you will win £25,000.

See www.theweatherlottery.com or call 0844 251 0509 for further information.

# PROCEED WITH CAUTION!

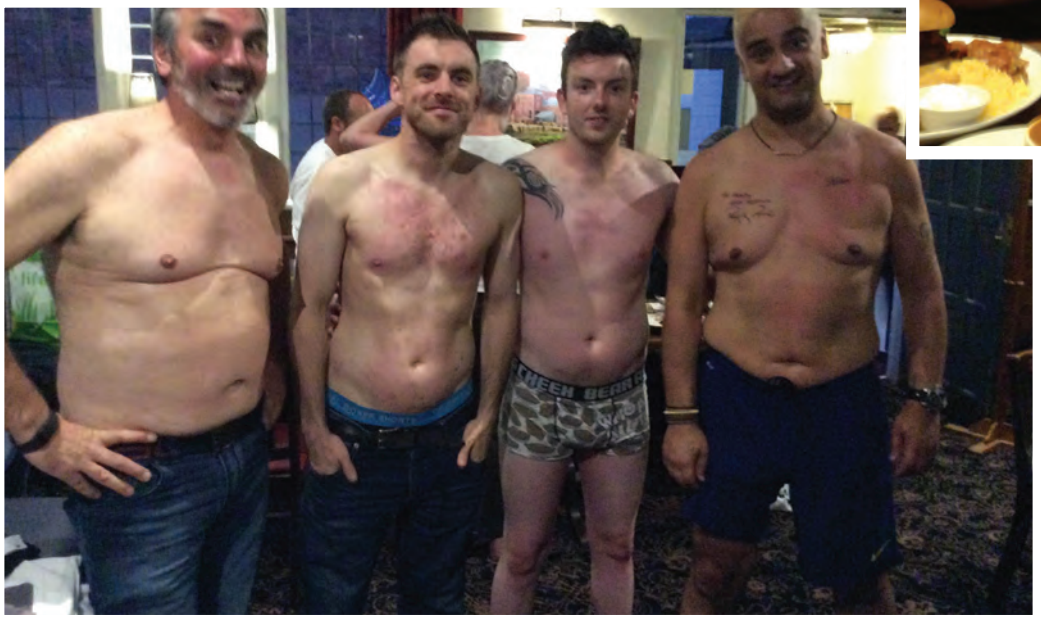
Paul Mahmood first bounced onto the MPS Society radar when he called the office with a long list of fundraising events he was planning to organise after hearing of the diagnosis of a good friend's son, Harley. It turns out Paul is a bit of a serial fundraiser and many charities have benefited from his hard work, organisation and crazy ideas.

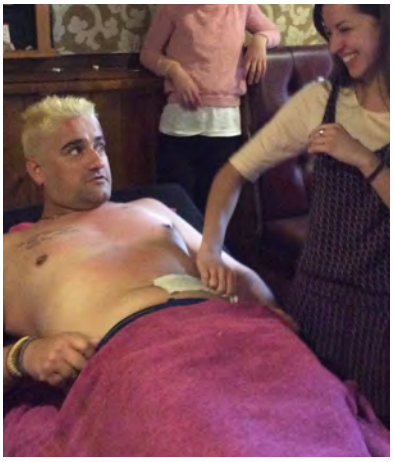
I perhaps should have been alerted to the craziness when I received my first email with the subject line: PROCEED WITH CAUTION! Enclosed was a collection of photos of an organised chest waxing event in Chesterfield.

Then there was the man vs food event where willing volunteers competed to see who could eat a 32oz steak, chips, mushrooms, onion rings and pudding.

Next up Paul is planning a night in a haunted castle this year and I'm sure we'll hear some other whacky ideas soon.

Thank you Paul, and thank you to all of the friends that Paul has roped in to being waxed, force fed and haunted in the name of charity.





# Kiltwalk

**With four events across Scotland and three distances to choose from – anyone can take part and change lives in Scotland.**

**This year you can look forward to Striding, Strolling and Wandering across Glasgow on 30 April, Aberdeen on 4 June, Edinburgh on 17 September and for the first time in three years, it's back in Dundee on 20 August!**

**And now the Kiltwalk is bigger and better than ever with 110% of every penny raised going to your choice of charity.**

**www.thekiltwalk.co.uk**

# New £1 coin but what about my trolley token?

You could possibly see the new £1 coin as early as 28 March which is when banks and shops will start to receive the first 1.5 billion being produced by the Royal Mint. For six months both the old and new coins will be legal tender with the current coin gradually being withdrawn from circulation until 15 October 2017 when it will no longer be legal tender - although you should still be able to pay in the old coin at banks for a little while after this.

But what does this mean for your MPS Society trolley tokens? Fear not, Tesco and other supermarkets have said that the locks on their trolleys have been replaced with coin slots that will accept both the old and the new pound as well as existing trolley tokens.

So there is no reason why you shouldn't have one of these snazzy little numbers on your key ring. Visit our online shop at http://mpssociety.tictail.com to get one for just £1 plus p&p.


And if you've got any paper fivers left make sure you spend those by 5 May.

More information on the new £1 is available here: www.thenewpoundcoin.com

Looking for a running challenge? We have a few charity places available in the Simplyhealth runs during the year, please register your interest at fundraising@mpssociety.org.uk.

Simplyhealth **Great Birmingham 10K** – 30 April

Simplyhealth **Great Manchester 10K** or **Half Marathon** – 28 May

Simplyhealth **Great Womans Run 10K Glasgow** – 4 June

Simplyhealth **Great North Run Half Marathon** – 10 September

## Virgin Sport Westminster British 10K

You might also be interested in the Virgin Sport Westminster 10K which boasts a flat, fast route that will take you through the heart of London and past some of London's most iconic sites and landmarks.

Taking place on 9 July 9 the Virgin Westminster 10K is the perfect event for those new to the long-distance running scene, or indeed for those experienced runners looking to post a fast time. The flat and forgiving route makes hitting a new PB a definite possibility, as long as you're now slowed down by all the incredible architecture and scenery that you'll be racing by!

If you're interested in taking part in the Westminster 10K or you just want some more information about the event then send an email over to fundraising@mpssociety.org.uk or call one of our friendly fundraising team on 0345 389 9901 we have a limited number of spaces available for this event so get in touch today!

# Have fun this Easter

If you're looking for Easter related fundraising ideas then try out our list below. We'd love to hear what you got up to as well so send in your Easter stories and pictures to magazine@mpssociety.org.uk.

- Make a start with a classic Easter egg hunt – you can hide decorated cardboard eggs instead of chocolate ones then swap them for chocolate at the end of the hunt
- Hold an egg painting competition – just provide the paint and decoration and ask everyone to bring their own egg
- Egg dropping competition – Wrap up an egg and drop it from a height. If yours breaks you are out and if it doesn't you stay in until there is a winner
- Raffle off a massive Easter egg or fill a jar with little eggs and get people to guess how many are in there
- Gather together scraps of fabric, ribbons, buttons and tissue paper and set up a "make your own Easter bonnet" stall
- If you can find someone willing to dress up as the Easter bunny you can charge people for a photo
- Make your own Easter cards to sell or donate and send an e-card via www.dontsendmeacard.com/ecard/3Jrw

And finally don't forget to listen to www.mix96.co.uk who are collecting Easter eggs from the public and donating them to local charities. Last year we received over 100 Easter eggs which were shared with children at patient meetings, given to members and auctioned off to raise funds for the Society.

# Wear it Blue

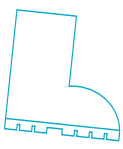
## for MPS and related diseases

Wear it Blue is our most recognisable fundraising campaign. We run it throughout the year but it's always most popular on and around International MPS Awareness Day which is celebrated on 15 May every year.

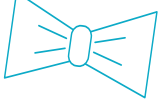
The idea is simple, wear blue and donate £1 to the MPS Society. It couldn't be easier. Get your Wear it Blue fundraising pack from www.mpssociety.org.uk/wearitblue

Combine it with a blue bake sale

Organise a blue walkabout

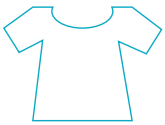


Make it fancy dress with a blue theme

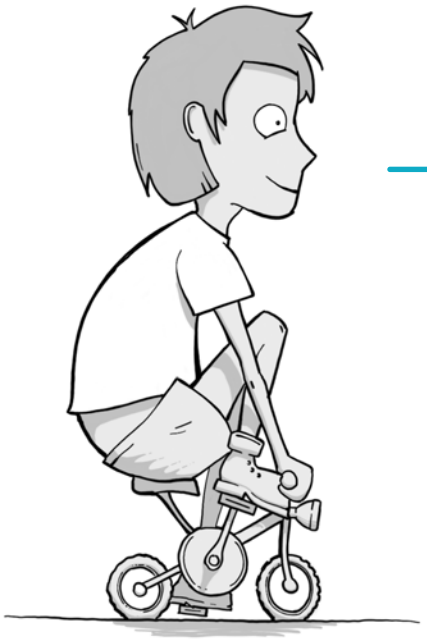


Sell some of our merchandise

Try some blue face paints



Print your own t-shirts



# The Big Draw

In 2016 Pete Norris decided he was going to draw every day to raise money for the MPS Society. His inspiration, a friend's daughter who has MPS III, the idea, that anyone could request a drawing and make a donation. We caught up with Pete to find out what kept him going and if he'll ever draw again!

**How long have you been drawing?**

I used to draw at school and was quite good, I remember being able to draw most things from memory – except animals, and hands, oh and feet (I'm now 44). I've always been a doodler, and my family contains no fewer than three artists. My Gran was a painter and gallery owner in Wales, her son, my uncle Paul, who is a marine and portrait artist on the Isle of Wight, and my cousin Phil, who is one of the best illustrators I have ever seen and a big inspiration. Not forgetting my Dad, who I get what natural ability I have.

I worked for 13 years as a computer network administrator, then five years ago I left to start my own business designing and building websites – needed to scratch that creative itch.

**Why drawing as a fundraiser?**

Because I'm a really terrible runner.

**What motivated you to raise money for MPS?**

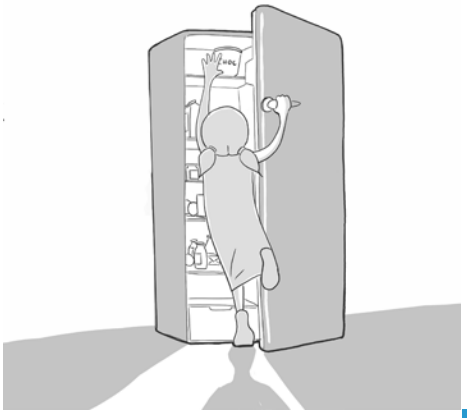
Our really good friend's daughter Sophia was diagnosed with Sanfilippo in 2015. They were understandably devastated. How do you support someone who's been dealt those cards? You want to do something, anything. We started to read more about MPS, and the more we read the more inspired I was to try and help.

**Where did the idea for the big draw come from?**

My cousin Tom is a keen bird watcher (twitcher), I remember saying how I was jealous of him and people with a passion, whatever that passion may be. What's your passion asked Tom? I've always wanted to draw really well, but I've never committed any time to it. I just said it there and then, I'm going to draw every day next year and raise money for Sophia.

**Did you ever have a day you didn't know what to draw?**

Not at first, but after a few months yes, I began to struggle. There are a few interesting still life pictures of



whatever I was doing at the time, a TV, a wine glass. I even drew a chef whilst he was cooking my takeaway down the local Chinese.

**How do you go about composing an idea?**

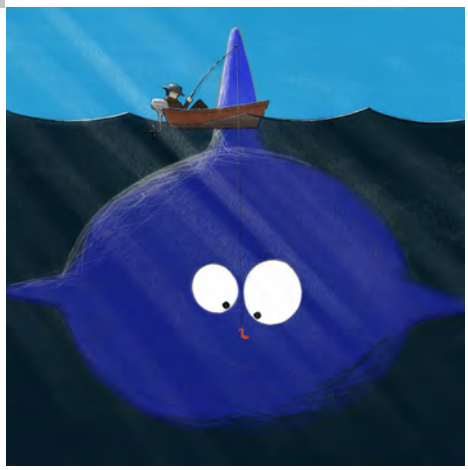
Something will happen during the day, or something topical in the news (never political – this is an escape from all that). I sketch different ideas with a pencil then once I have the composition I get to inking (I love thick black lines) and sometimes adding colour. Getting colour right was a big surprise, I'm still not there yet. It's a science in itself, so I mostly just drew in black and white.

**Do you have a backlog of requests?**

Regrettably I do yes. Most of the requests were to draw people's dogs and I would get a photo. Not only can I not draw animals very well, but I don't know their dog's character either, so I wouldn't have done them justice. One request I had was from an anonymous person asking me to draw a picture of her child being included after they had a bad day at school. It broke my heart and I wanted to do something really good for them. I didn't just want to do a daily draw. So if you're out there, please get in touch and I will do a proper print for you – sorry!

**How do you translate what someone has requested into a drawing?**

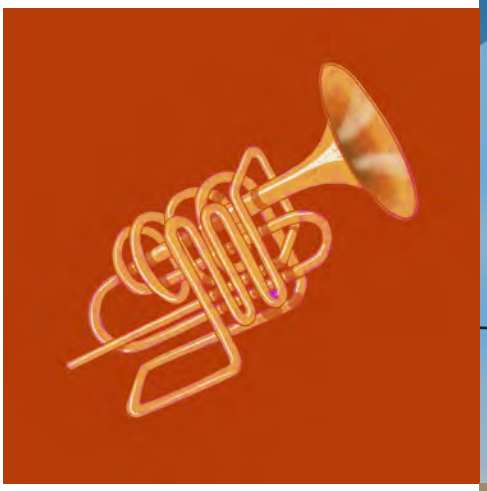
I think having two kids, and watching them shaped a lot of my drawings.

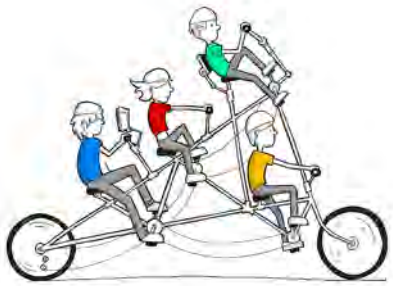


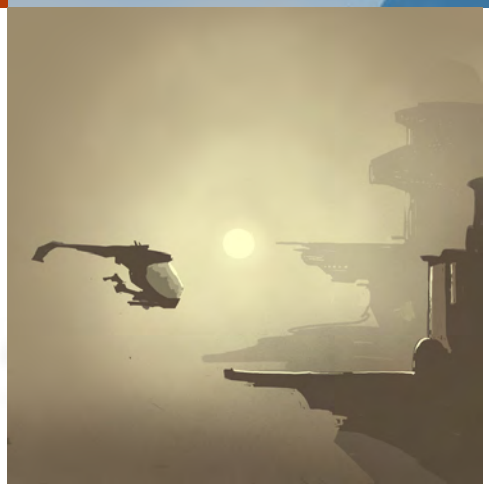
Most requests were translated into playful and mostly cartoony pictures.

Which is your favourite (or is that like asking someone which one is their favourite child!)

Ha ha! No, I'm not precious about any of them. Although some of them I feel work really well – mostly the ones of kids in black and white. And you can probably tell, I have a fondness for spaceships.







There is a picture of Sophia on the website, which I saw each night when I uploaded a drawing. She is the reason I did it and the biggest motivator.

**Where now?**

The first thing I plan to do it is to make a small book of some of the favourites and sell for MPS. It was an idea from Sophia's dad when I first started. I have also had requests for prints, and some requests for new pictures. I also intend to carry on this journey in learning to draw but perhaps without the pressure of doing it every day. I intend to learn to draw hands, feet, and perhaps animals.

**What have you learned from this experience?**

It was hard. Really hard. Some days I loved it and drew for hours, but other days I wished I'd never started. I know there are art students who churn out 10 drawings a day, but my headspace is in my family and running my business, and it was not a natural action to just sit down and draw. It was a huge commitment. My family were very supportive to my 'Sorry, I can't, I've got to go and do my drawing moments'.

The biggest surprise was how difficult it became after time. At the beginning there was a lot of buzz and support from those around me, but after a while I was on my own and having to fit a drawing in would sometimes get in the way of other plans. My wife is a big Facebooker whereas I'm not, so she would post a picture every now and then, the support of which would spur a new bout of inspiration. A camping trip in the summer where each night I had to sit outside a closed pub to borrow their wifi to upload the picture was a particularly negative memory – not aided by the fact the pub was closed.

If anyone out there needs a picture and they are willing to donate money to MPS, then I can be contacted on twitter at @petejnorris or by email at petenorris@gmail.com.


See all of Pete's drawings in The Big Draw at petenorrisdesign.com


# Thank you to all our donors and fundraisers – you inspire us!

Portland Medical Practice in Walsall held a Christmas Jumper Day and raised £66.50.

Elizabeth MacDonald donated £10 being 50% of the proceeds from a work Christmas Jumper Day.

Jacqueline McKennan from NHS Ayrshire & Arran Primary care Trust raffled a Christmas hamper and raised £38. MPS were the nominated charity selected by staff.

Carolyn Rockett, family and friends from Dorset raised £100 at Christmas parties playing bingo, this money is in memory of Carolyn's son Mark who had Hurler (1985–1992).

Carol, Joan and Barbara raised £250 through attending three small craft fairs selling handmade items.

Peter Green Haulage held a Christmas Jumper Day and raised £25.70 for the Society the other recipient was the British Heart Foundation.

Lee Wright from Manchester cycled from Bolton to Scarborough and raised £235 on this virgin money giving page for the Society.

EP Morris and Company raised £500 as they nominated us as their Christmas charity thanks to their employee, Gemma Holyhead.

Abi Whittles raised £2,240 from a half marathon.

Stephen McCawille raised £491.10 from running the Beachy Head Marathon.

Reece Barlow raised £20 from taking part in "My Tital Fight".

Philip Lee and Ashley Monk raised £970 from taking part in the Windmill 10K run.

Joanne Haines ran the Great South Run and raised £243.50.

Deborah and Mark Burniston raised £773.28 from participating in the Great North Run.

Elizabeth Hull raised £557.49 from participating in the Great Birmingham Run Half Marathon.

Beth Chambers raised £10 from a 10K run.

Lee Shepherd raised £1,145 from running in the BUPA Great South Run.

Linda Kirby raised £45.

Katriona Balfour raised £245 from taking part in the Scottish 10K run.

Richard Smedley raised £266.

Amanda Stuart and Nicola Caslin raised £40 in lieu of sending Christmas Cards

Women's Institute of Worfield raised £100 after choosing us as their charity of the year in 2016.

Michael Wheeler sent in £60 raised by the Guildford Gas Golf Society at a golf day held at Sutton Green Golf Club

Stephen McCawille has raised £124.06 running in the Dingle Marathon.

P&H Direct – Sheffield Head Office team raised £38.02 by activities within the Head Office.

t-pow.co.uk raised £60 through organising several festivals – we were nominated by two of their volunteers.

The Haddenham Mummers were once again out and about performing their historic play in the lead up to Christmas. They acted in all the village pubs nearby to Haddenham and for the first time ever on The Chinnor and Princes Risborough Railways steam train. They raised over £3,000, the MPS Society received a £660 share of this along with other local charities.

Matthew Lamb sent a cheque donation of £105 from the Great North Run.

Liverpool Victoria's Ipswich office took part in the Three Peaks Challenge raising £750.

Huby Methodist Church collected £77 at Christmas Eve services.

# Donations

Zoe Warner; Kath Hiller; Jean Davy; Mrs A Baker; Arlene Murray (tins + family donation), Mrs K Crayton, Alan & Monica Bowen; Peter Nelson; Mrs Baker; L & S L Stewart; Eva Stavsoien; Mrs AS Calveley; Mr & Mrs AJ Eaton, Matthew Lamb; Roger Jennings, Paula Waters, Iain Major, Ann Brown, Dave Isherwood; Mr Jennings, Paula Waters, Iain Major, Ann Brown, Dave Isherwood, Dr Janet Stone; Assefash Hagos-Kidan; Malcolm and Blaise Leslie: Jean Mossman; Michael Gayton; I Sanderson; Ann Baker; Gordon & Mary Mellor; Mrs S Brown; Mr & Mrs MacDonald; David Tonge; Rosemary Harding; Margot Carter; Mrs D Brown; Ms C Hallerton; Miss L Hiller; Mr A Dickerson; Mrs M Crespin; Mrs T Brown; Mr A Selwood; Mr M Hughes; Miss D Halleron; A Owens and CJ Owens; Dennis Fiore; Linda Windsor; Associated Manchester Electrical Trades; The Carrington Foundation; Belinda Fitzpatrick; Mrs D M Kent; Liz Rodda; Ann & Rich Coleman; Peter Graves; Andy & Jenny Hardy; Pamela Booty; Sarah Jenkinson; WM. & I. Graham; Danny & Margaret McKeeman; Agnes Palastanga; Kerry Palastanga; Stephen Palastanga; Raymond & Madeleine Lyttle; Elizabeth Kane; Kathleen Taggart; Archie & Anna Graham; Mr & Mrs Alex & Joyce Burnett & Family; Mr & Mrs W.G & M McLaughlin; Desmond & Sylvia Mckeeman; Alistair &Arlene McMullan & Family; Colin & Jane Kane; Syvlia, Robert & Jason Christie; Elaine Hunter; Craigalappin; A. Gaffney; Lynne &Sammy Dobbin; Marney & Jim Watson; Doreen & James Boyd; Mrs Betty McKaig.

### Regular contributions by Standing Order or Give As You Earn

M Kalsi; P Summerton; S Winzar; G Simpson; William Cavanagh; Barbara Harriss; L Brodie; A Sullivan; Evelyn White; C Hume; A Weston; Dorothy Robinson; A Byrne; V Little; S & J Home; K & S Bown; G Stuart; R & K Henshell; Lindsey Stillwell; A Ephraim; R Gregory; R Taylor; E Parkinson; Gordon Ferrier; Gillian Reeves; E Brock; J Dalligan; M Malcolm; E Mee; Elliot Moody; S & D Greening; M Hahner; K Brown; Z Gul; M Fullalove; M Leask; R & N Gregory; V Little; J Casey, J & V Hastings; J Winzar, D Winzar; K & J Hudson, E Lee; Saville Norman; R Dunn; M Tosland; C Cullen; S Bhachu; S Brown; V Lucas; D Forbes; N Cadman; J Wilson; A Tresidder; J Arnold; E Cox; K Osborne; S Robinson; N Thompson; C Garthwaite; J Ellis; I Pearson; Manjit Kalsi; Mrs S Bachu; S Brown; S Cadman; A Cock; E Cox; AP Dickerson; R Dunn; J Ellis: G Forges; Mr & Mrs Gibbs; CL Hume; C Parkinson; I Pearson; M Reeves; S Robinson; N Saville; G Simpson; L Stilwell; P Summerton; R Taylor; A Thomas; M Tosland; J Wilson; A Tresidder;C & M Gibbs; Tim Peach; David Patton; J Garthwaite; Paul Berg; Michael Morris; Abbey Thomas; Elizabeth Merryweather; Nick Miles Amanda Laycock; Peter Rennoldson; P Berg; N Saville.

### Donations via collection boxes, stamps, foreign coins, mobile phones, ink cartridges, jewellery, PayPal Giving, eBay for charities

Sue Lowry; CE and DM Robinson, Wilma Robins

# In memory

Elizabeth Ann McConnell; Gracie Bella Sims; Mark Rockett; Julie Bennett; Dr Charles Pennock; Mrs Caroline A Hill; Pauline Wright


**Thank you also to all those who donated anonymously – we don't know who you are, but we think you're great!**

Thank you to those who donated via the Weather Lottery:

Mrs D Bown
Ms C Halleron
Miss L Hiller
Mr A Dickerson
Mrs M Crespin
Mrs T Brown
Mr A Selwood
Mr M Hughes
Miss D Halleron
Mrs G Plummer
Mrs C Lavery
Mrs J Edwards
Mrs O Megoran
Mrs J Speed

If you want to be in with a chance of scooping the £25,000 weekly jackpot whilst also supporting a good cause go to www.theweatherlottery.com and search for "Society of Mucopolysaccharide Diseases"

# Patient Access to Clinical Trials and Treatment

## Patient Access to Clinical Trials and Treatment (PACT) registered as MPS Commercial

**Registered Address:**

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

mps@mpspact.com
www.mpssociety.org.uk/commercial

T: 0345 389 9901
F: 0345 389 9902

**Board of Directors**

**Chair**
Bob Stevens

**Board**
Derek Beards
Jessica Kafizas
Georgina Smith
David Patterson

**Secretary**
Christine Lavery

MPS Commercial is a Private Limited Company Registered No. 08621283.

MPS Commercial trades as Patient Access to Clinical Trials (MPS PACT), and is a wholly owned, not for profit subsidiary of the Society for Mucopolysaccharide Diseases (the MPS Society), Registered Charity in England and Wales No. 1143472.

MPS Commercial's social objectives are to reinvest any profits for the purposes of education, enhancing needs-led advocacy support, quality of life research and scientific research to the MPS community.

## MPS Commercial Employees

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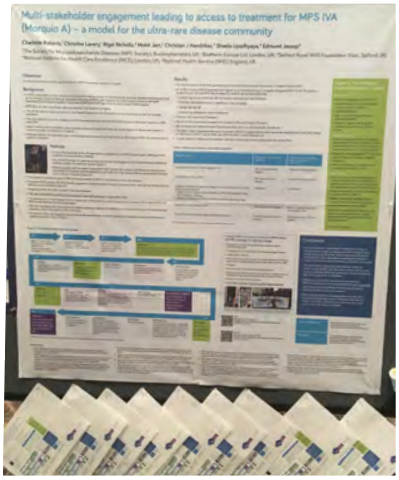
## MPS Commercial at WORLD 2017

Three of the MPS Commercial team attended the WORLD Symposium in San Diego, California, in February.

WORLD, which stands for **W**e **O**rganise **R**esearch on **L**ysosomal **D**iseases, is the annual gathering of scientists, clinical researchers, patient advocacy groups, and doctors, where the latest scientific and clinical trial data on lysosomal diseases are presented.

Charlotte, Alex and Jackie had a busy week of lectures and meetings, which started at 6.30 in the morning and finished at 8.30 in the evening. Meals were accompanied by learning about advances in mucopolysaccharide disorders and Fabry disease in satellite symposia (lectures sponsored by the pharmaceutical industry).

## Meet the team

**Christine** is the Group Chief Executive for the MPS Society and its commercial subsidiary.

**Gina** is the Group Finance Officer for both the MPS Society and MPS Commercial.

**Charlotte** manages the patient access clinical trials team who provide tailored logistical support to patients and their families.

**Jo** is Clinical Trial & Patient Access Officer and supports families participating in clinical trials across the world.

**Benedicta** provides a logistical service for individuals participating in clinical trials.

**Alex** is Clinical Data Lead. She coordinates the collection, management and analysis of data from research surveys and the Vimizim Managed Access Programme.

**Jackie** is Clinical Communications Lead. She coordinates drafting internal and external reports, research surveys and medical/clinical communications.

**Pauline** is the MPS Commercial accounts assistant.

**You can now reach MPS Commercial on their own dedicated number**

**0345 2601087**

We had the opportunity to present our poster about the development of the Vimizim (elosulfase alfa) Managed Access Agreement (MAA), at the poster session; where the poster is on display and the authors stand by it and answer any questions that people may have about it. This went really well and we had a lot of interest in MAA from doctors in other countries.

On our last night, we put on our glad rags and joined Christine and Sophie, from the MPS Society, at a gala banquet, where Christine was presented with the 2017 Patient Advocate Leader award in recognition of her work in the MPS and related diseases field.

## Update on the MPS III European study

Preparations are continuing for this multi-national study that will provide vital information to support both the diagnosis of MPS III and the development of future treatments.

We will be talking to families about their own experiences of MPS III in the UK and also in Austria, Germany, Greece, Serbia, Spain and Switzerland with the help of the local MPS Societies who attended the study training hosted by MPS Commercial in Zug, Switzerland.

We are currently working on the final adjustments to the questionnaire and liaising with our European MPS Society colleagues to ensure that all country specific requirements are accounted for in the study design.

**What will the survey tell us?**

The specifically designed questionnaire covers three main areas. Together these will provide a broad picture of MPS III to further the understanding of the natural history of the disease, inform educational activities to aid diagnosis and help to identify children with MPS III at an early age. Through documenting both the symptomatology and the burden of disease an indication of disease parameters that new therapies could potentially impact can be assessed.

**Path to diagnosis**

- Where knowledge gaps are delaying the diagnosis of MPS III
- The trigger symptoms that first raise the suspicion of an MPS disease
- The evolution of symptoms from birth to diagnosis

**Current symptoms and disease management**

- Presentation of disease by age
- Differences between MPS III subtypes A, B, C and D
- Country differences in disease presentation and management
- Unmet medical needs

**Burden of disease**

- Amount of medical and educational support needed for a child with MPS III
- Effects on parents and families' ability to work
- Need for more suitable housing or adaptations to the home

MPS Commercial host training in Zug


# What does MPS mean to you?


## MPS Awareness Day
## 15 May 2017


## How you can get involved

Shout about #MPSAwarenessDay on your social networks

Download and use the profile image on Twitter, Facebook and Instagram

Print our poster and tell us what MPS means to you

Raise awareness locally using our press kit

Ask us for a Wear it Blue fundraising pack

Donate to the MPS Society text WIBL17 £5 to 70070

**Why are we raising awareness?**

Chances are unless you know someone with MPS you will never hear about it. We need to change that. Better awareness means earlier diagnosis, more research into treatments and a better understanding of the needs of people living with MPS.

**Why so blue?**

The MPS Society Wear it Blue fundraising campaign runs all year round but is an extra big deal on MPS Awareness Day. This year we are asking you who you're wearing blue for so get your fundraising pack and join in.



Your MPS Awareness Day and Wear it Blue info is waiting for you at www.mpssociety.org.uk/wear-it-blue