THE MPS MAGAZINE

Society for Mucopolysaccharide Diseases

Support Research Awareness

Summer 2016

www.mpssociety.org.uk

Mucolipidosis Conference

Our first conference focussing on Mucolipidosis in April was a great success.

For highlights from the weekend turn to page 18.

Gulliver's Land family day

We had a fantastic day out at Gulliver's Land Theme Park in Milton Keynes for MPS Awareness Day.

Photos of day are on pages 22–23.

Welfare change in Northern Ireland

Benefits are changing but this article by our advocacy officer for Ireland will tell you what you need to know.

Pages 32–33.

Front cover photo: MPS Awareness Day at Gulliver's Land, Milton Keynes
Icons on p.27 made by Freepik from www.flaticon.com

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Magazine Deadlines

Autumn	1 September 2016
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To submit content email:
magazine@mpssociety.org.uk

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

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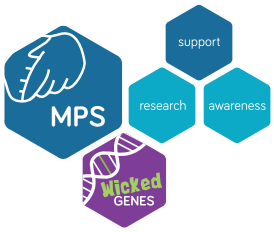
Time for a clean?

If you've had a spring clean or are planning a clear out over summer think of the MPS Society and the environment and recycle for charity. We want your:

- mobile phones
- print cartridges
- used stamps
- unwanted jewellery
- foreign currency
- old cars



The MPS Society

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

Our Aims

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

Our Vision

A world that is free from MPS and related diseases.

Our Mission

All children and adults affected by these diseases:

- Can be treated;
- Have equitable access to state-of-the-art clinical management and therapy
- Are active members of society reaching their full potential
- See pioneering gene therapy research offer a cure for these devastating diseases

MPS and Related Diseases

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

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

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twitter.com/MPSSocietyUK

Welcome

Welcome to the summer 2016 edition of our MPS Magazine.

We have some fantastic pictures of your Wear it Blue fundraising. You have done a brilliant job again this year and the photos show it. Our cover photo is from the MPS Awareness Day family trip to Gulliver's Land where you'll see everyone wore it blue in support of the Society.

There has been a number of events since the last issue. The All Ireland Conference on 20th–22nd May was a great success and our first ever conference focussing on Mucolipidosis was very well received.

We have also included an update on how the Budget 2016 affects you (p31) and what you need to know about welfare change in Northern Ireland (p32–33) which includes a very useful step-by-step diagram about making a claim to PIP.

Best wishes
The MPS Society

PARALLEL LONDON

JOIN THE WORLD'S 1ST FULLY INCLUSIVE FUN PUSH/RUN & FREE FAMILY FESTIVAL!

4th September 2016
Queen Elizabeth Olympic Park
10km, 5km, 1km, 100m

All ages. All abilities.
No cut-off times. No excuses!

www.parallellondon.com

Group Chief Executive's Report

The highs and lows of life are never closer to the surface than amongst the members of the MPS Society and their families. Whether you have just received a devastating diagnosis of MPS, Fabry or a related disease, living with one of these diagnoses or living with the loss of a family loved one however long ago. I know time seems to fly by and stand still all at the same time when it comes to bereavement. Our 7 year old son died from MPSII, Hunter disease 34 years ago on the 17 May. Was I at the crematorium to reflect and celebrate Simon's life? Well no, I was on a flight to Tokyo, Japan to host the Fabry International Network. That is where Simon would have wanted me to be, in a city that for more than half his short life he called home.

This personal reflection came to mind as over many years, but perhaps these past months in particular, we at the MPS Society have supported so many families through too many lows and highs that are tragically out of reach for so many of our members and their families.

For four decades gene therapy has been an aspiration and suddenly today for MPSIIIA and MPSIIIB it may be a reality. With Abeno, Lysogene and Orchard Therapeutics about to, or very close to launching their clinical trial gene therapy programmes we can celebrate for the very few who will be accepted onto these clinical trials and empathise with the many families whose children live on the wrong continent or 'were born too soon'. What we can take heart from is that if these clinical trials show benefit there is real hope for the as yet undiagnosed generation of babies with Sanfilippo disease. Coming up fast behind is gene therapy for MPSI and MPSII and success in these diseases should open the door for clinical trials for other lysosomal diseases in the future. In the meantime we continue to work with the scientists, clinicians, the pharmaceutical industry and regulatory bodies to support new therapies for our diseases.

As I stated in the last MPS Magazine Patient Reported Outcomes (PRO) provide incredibly important data for the pharmaceutical industry developing new therapies in ultra-rare diseases and agencies like NICE, Scottish Medicines Agency (SMC) and NHS England. Over the coming weeks we will be inviting the families of those who have lost their lives to or living with MPSIIIA, MPSIIIB, MPSVI and Fabry disease to participate in two PRO surveys to help us create important anonymised data to support disease knowledge. Clinicians have clinical data but only those of you living with one of these diseases knows the real impact so I trust you will open your hearts when it comes to these surveys.

It was amazing that over 100 members and their families were able to join us at Gulliver's Land near Northampton on 15 May for MPS Awareness Day and thank you to everyone across the four nations of the United Kingdom who supported our MPS Awareness 'Wear it Blue' Campaign. Awareness is so important for these diseases as the earlier the diagnosis the more options there are for managing many of these diseases. It is never more heart breaking for a child to be diagnosed late and missed out on a clinical trial or in the case of MPSI, a Haematopoietic stem cell transplant (HSCT). Quite often we at the MPS Society are talking to the converted but with over 1300 member families spread across the whole of the UK meeting junior doctors and consultants outside of the expert centres as well as health visitors, community nurses, OTs, physios, speech and language therapists and teachers, classroom assistants and SENCOs you can spread MPS awareness every day and drive home the importance of early diagnosis. We have to ask you to do the Government's work as the New Born Screening Committee (NBS) have just refused new born screening for MPSI and won't even look at other MPS diseases or Fabry where there is an approved therapy.

> "For four decades gene therapy has been an aspiration and suddenly today for MPSIIIA and MPSIIIB it may be a reality.

Looking for highs, I and the MPS advocacy team will be travelling to Bonn, Germany on 12 July for the International MPS Symposium. We look forward to taking MPS families and young adults with us and in the Autumn MPS Magazine we will report on what is new in the MPS world. In the meantime we hope the sun shines upon you this summer.

Christine Lavery
Group Chief Executive

News From the Board of Trustees

The Society's Trustees meet regularly. Here is a summary of the main matters discussed and agreed at the Trustee Board of Trustees Meeting held on 19 – 20 February 2016

Governance

One Trustee raised trustee training. All Trustees confirmed their commitment to taking up training opportunities. It was recognised that the current MPS Board is highly skilled and it was agreed trustees would benefit from agreeing areas of training need as they arise.

Financial Management

The Treasurer's Report and Financial Report were distributed in advance and accepted. Trustees discussed how the Gift donation from MPS Commercial might be best expended in the interests of the MPS Society and its members. It was agreed unanimously that these funds support MPS Families to participate in the MPS International Symposium in Bonn in July 2016 and a MPS Family Trip to Lapland in December 2016. The MPS Society Reserves Policy was approved subject to the Trustees amendments and agreed unanimously.

Trustees were asked to approve the MPS Consolidated Year end Accounts for 31 December 2015 that had been circulated to all Trustees for comment in advance. Subject to minor corrections not affecting the figures and points to be clarified with the auditors the accounts were approved unanimously and agreement given for the Chair to sign the final copies when received from the auditors. The Group Finance Officer was thanked for her efforts.

Risk Management

The Group Chief Executive took the Trustees through the Risk Management register and amendments to risk levels were agreed for Loss of key staff (Senior Leadership Team), Generating Income, Disaster Recovery and planning and Compliance.

Policy Strategy

Trustees considered the impact of Trustees not fulfilling their governance obligations and agreed that the Trustees' Conduct policy needs to be strengthened. Two Trustees agreed to co-ordinate a first draft.

A Trustee presented the findings of the Trustees understanding of the Financial Controls Policy. It was agreed amendments to be made to the policy and circulated in advance of the April Board of Trustees meeting for approval.

Subject to one amendment the Anti-bribery and corruption policy was approved. The zero-tolerance statement was approved for the website subject to two minor changes.

Amendments to the Gift and Hospitality Policy were considered. After a full discussion it was agreed there would be no lower figure below which gifts and hospitality do not need to be registered. The policy was approved subject to minor changes.

Personnel

Trustees ratified that Toni Ellerton is now the part time Fabry International Nertwork Co-ordinator as part of her PA role to the Group Chief Executive

Students for Rare Diseases

Trustees were appraised of the sources of funding for the Students for Rare Diseases (S4RD) co-ordinator post. The Trustees agreed unanimously to £2,000 in kind to support this post by accommodating the S4RD co-ordinator at MPS House for 12 months.

Advocacy Support

The Advocacy Support Service Report was circulated in advance to Trustees. The Trustees noted the MPS Society Press Release on LaL Disease.

What's On

MPS Regional Clinics 2016

MPSI - GOSH
26th July

MPSIII - GOSH
13th September

MPSIV - GOSH
8th November

MPSII/MPSVI - BCH
14th October

MPSIII - BCH
13th October (afternoon)

MPSIV - BCH
16th September

MPS Transition Clinic - BCH
29th September

MPSI Post HSCT (over 6 years) - RMCH
7th October

MPSI Post HSCT (under 6 years) - RMCH
14th October

Fabry - BCH
25th November

Adult Fabry - QE, Birmingham
9th August • 13th September
11th October • 8th November
13th December

Conferences and Regional Events

14th International Symposium on MPS & Related Diseases - Bonn, Germany
14th - 17th July 2016

Childhood Wood Planting - Sherwood Pines
16th October 2016

Lapland Family Trip - Finland
2nd-5th December 2016

SAVE THE DATE
MPS Weekend Conference 2017 - Hilton, Coventry
7th July - 9th July 2017

Announcements

Goodbye to Elkie

We say a sad goodbye but a happy good luck to Elkie who has worked at the MPS Society for 2 years as a Fundraising & Information Officer. Elkie has moved on to new pastures at Bucks County Council and we are sure she will do really well there. You can direct any fundraising queries to Helen at the same email address: fundraising@mpssociety.org.uk

Baby news

Baby Cora May Wilson was born in the afternoon of 23 June weighing a healthy 9lb 5oz.

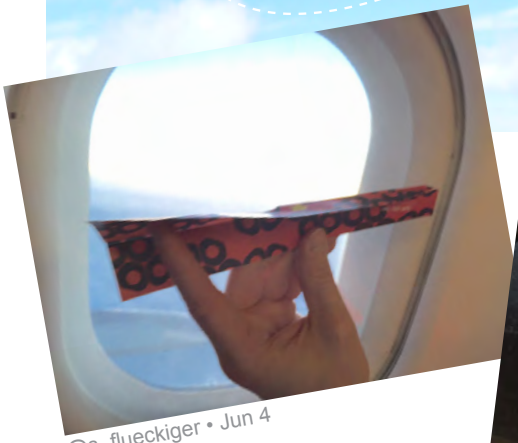
Congratulations to one of our trustees, Jessica Reid, who tied the knot on Saturday 23 April

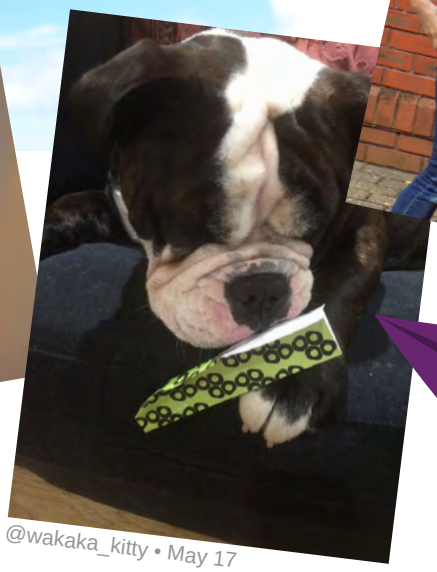
> “Saturday was fabulous – the sun shone, it was amazing and more!

We hope you had a great honeymoon in Costa Rica Jessica!

Fly for MPS

The Fly for MPS campaign for MPS Awareness Day closed on 15 June and made more than 6 million air miles and nearly reached the target of 8000 connections. We hope everyone who joined in and made a plane had fun. It has been great to see the support on social media and we want to say a great big thank you to everyone who showed their support by flying a plane.


@s_flueckiger • Jun 4


@wakaka_kitty • May 17


@MPSSocietyUK • May 15

New Members:

Mrs Bendahou has recently been in contact with the Society. Her daughter Ikram has a diagnosis of LAL D. She is 11 years old and lives in the West Midlands.

Natalie has recently been in contact with the Society. She and her two sons have Fabry disease. The family live in the Manchester area.

Kirsten has recently been in contact with the Society. Kirsten and her two daughters, Amber and Lily, have Fabry Disease. The family live in the Bedfordshire area.

Lisa has recently been in contact with the Society. She and members of her family have Fabry Disease. The family live in the South West.

Sarah has recently been in contact with the Society. Herself and her three children, aged 13, 10 and 6, have Fabry Disease. The family live in the Derbyshire area.

Beverley has recently been in contact with the Society. She has a diagnosis of Fabry Disease and said "I, my two adult children and all five grandchildren have been diagnosed with Fabry." The family live in the Derby area.

Ms Zoya Salam has recently been in contact with the Society. Zoya has a diagnosis of MPS I Disease. Zoya is 26 years old and lives in the North West.

Mami has recently been in contact with the Society. Her sons (4 years and 10 months) have a diagnosis of Hunter Disease. The family live in the London area.

Advocacy

The MPS Advocacy Support Service has been established since the Society was founded in 1982. At this time there were only 40 known families throughout the UK. The support provided was on a voluntary basis and depended heavily on individuals and parents to provide support to individuals diagnosed within their immediate and surrounding areas.

However in 1991, the Society opened its first office and with this the advocacy service we know today was born.

The MPS Society provides, through a team of skilled staff, an individual advocacy support service to its members. The service is flexible and a wide range of support is offered on a needs led basis.

The rarity of these conditions means that in many cases, accurate assessments, support and advice are not given due to the vast majority of social care and health professionals knowing very little if anything about the diseases.

Support provided by the team

• Telephone Helpline

0345 389 9901– the Society provides an active listening service, information and support. This includes an out of hours service

• Disability Benefits –

In understanding the complexities and difficulties individuals and families have in completing claim forms for Personal Independent Payment, the Society continues to provide help and support in completing these forms and, where needed, will take a representative role in appeals and tribunals

• Housing and equipment –

The Society continues to take a major role in supporting and advocating appropriate housing and home adaptations to enable the needs of an individual with an MPS or related disease to be met. Where requested, we can provide comprehensive and detailed housing reports based on individual need

• Education –

The Society helps members to access appropriate education and adequate provision for its implementation. This is achieved through providing educational reports used to help inform and educate professionals, and in many instances, to inform Statements of Special Educational Need. Where requested, we also provide information days/ talks to schools and relevant professionals

• Respite Care –

The Society continues to work closely with a number of respite providers and, where appropriate can make individual referrals

• Independent Living/ transition –

The Society provides advice, information and support through the transition from child to adult services. This could include access to independent living, learning to drive, further education and employment

• MPS Careplans –

The Society undertakes a comprehensive assessment of the issues which need to be addressed when caring and providing support to a specific individual diagnosed with an MPS or related disease, as well as other family members through the writing of a careplan

• Befriender Service –

The Society links individuals and families affected by MPS and related diseases for mutual benefit and support

• Bereavement support -

For more information on any of the above or if there is anything else that you would like to chat with the advocacy team about please contact us:

Email: advocacy@mpssociety.org.uk
Telephone: 0345 389 9901

Advocacy Resources

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

- Life Insurance
- Travel Insurance
- Hospital Travel Costs
- Disabled Access Holidays • Carers Legal Rights
- Carers Allowance
- Wheelchairs and Flights • Guide to Housing and Disabilities Facilities Grant
- Benefits including, Personal Independent Payment, Benefit Cap, Council Tax Benefit and Universal Credit

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

Our Advocacy Support Team

The rarity of MPS and related conditions means that in many cases, accurate assessments, support and advice are not given and individual need is neglected or undermined by policies and practices which, do not address the multi- systemic nature of these diseases.

The MPS Society provides, through a team of skilled staff, an individual advocacy support service to its members. The service is flexible and a wide range of support is offered on a needs led basis. This is achieved through direct contact with our members either in person by telephone, email or letter.

The Society currently has approximately 1300 registered members approximately 750 of these are living with an MPS or related disease. A review of current caseloads over a 12 month period (Jan – Dec 2015) concluded active cases averaging at approximately 150 a month.

The team provide professional advocacy that encompasses support, information, representation with the aim of empowering and promoting access to services and enable our members to express needs and choices.

The advocacy service provides support across the UK and although its main base is situated in Amersham Buckinghamshire we do have a regional service which is delivered in Northern Ireland.

In order to meet the aims and core principles of the service, each advocacy officer must work to a high level of professionalism. In order to do this the following skills, knowledge and person qualities must be present, applied and reviewed regularly

Current staff team professional specifications
- Qualified Social work
- Public / statutory services
- Genetic services

SOPHIE

Manages the MPS Advocacy Team

DEBBIE

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

REBECCA

Fabry
MPSII Hunter
MLIII / MLIV
Mannosidosis, Fucosidosis

LOUISE

MPSIII Sanfilippo type A, B, C and D
LAL D
Gangliosidosis

ALISON

Supports members living in Ireland

STEVE

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

All Ireland Advocacy Support Update

The last few months have been a whirlwind of organisation ahead of my maternity leave – by the time you are reading this article I will already have left my desk (I can hardly believe how fast the time has gone!). In the last few months I have spoken to most of our members across the Island of Ireland and lots of hours have been spent supporting families to complete forms, write careplans and meet with all sorts of different agencies to communicate needs. It has been a hectic few months but I've enjoyed every minute of it and feel that I know you all even better than I did before!

You will all have received a letter encouraging you to make contact with the Advocacy Support Team in MPS House if you need support in my absence. Please do get in touch!

MPS House Contact details:
Phone: 0345 389 9901
Email: advocacy@mpssociety.org.uk

By far the busiest few days of the last quarter were in May. On the 20th May we had our Summer MPS clinic in the Royal Belfast Hospital for Sick Children. Followed by a jam-packed conference weekend at the Hilton Hotel in Templepatrick!

Northern Ireland Paediatric Clinic

As usual our clinic was exceptionally busy. Together with a multidisciplinary team of medical professionals we reviewed 12 of our MPS Society members. Everyone got a top-to-toe check-up and had the opportunity to ask questions and address any unmet Advocacy Support Needs. As usual your feedback is very important and I would encourage you to send back completed feedback forms so that we can ensure the clinic is meeting your needs.

Advance notice: In 2017 our Northern Ireland clinics will be changing! Rather than the usual two clinics per year we are increasing our capacity to three clinics. This will ensure that all of our families have adequate time in clinic to address their medical needs, this should also give us additional capacity for you to see other professionals on the day.

Save the date

Children's Clinics
MPS I Clinic – 7th October 2016
MPS II, III and IV Clinic – 10th March 2017
Other related conditions – 19th May 2017

Adult Clinics
Antrim Clinic – 2nd December 2016
Antrim Clinic – 21st April 2017
Altnagelvin Clinic – 20th September 2017

All Ireland Conference – Hilton Hotel Templepatrick

Our All Ireland Conference was held on the 20th-22nd May in the Hilton Hotel in Templepatrick. We were delighted to welcome a range of local professionals (and some from a little further afield).

In the morning we heard from our core MPS and Fabry Teams (Drs O'Sullivan, Stewart and McKeown as well as specialist nurses Nicky Cluskey and Joanne McOsker) who updated us on the progress of Northern Ireland MPS and Fabry Clinics. It was great to hear how the services have developed over the last few years and also how the clinics will change in the months to come. It's so important to keep reviewing and changing how we do things to meet the needs of those affected by MPS and Fabry – please do let the team know if you have any suggestions for how we can develop the clinic in the future.

Next we heard from Drs Robinson, O'Donoghue and Humphreys as well as specialist nurse Barbara Maxwell who spoke about the Orthopaedic, respiratory and Dental implications of MPS. These disciplines are key parts of the MPS medical team and are present at each of our specialist MPS Clinics.

After a welcome lunch break we heard from Declan Hannah and Katie Burns who are psychologists involved in the care of adult and paediatric patients respectively. These were two exceptionally beneficial talks that 'demystified' how we view phycologists and the work that they do. Declan spoke about the management of long term chronic pain and emphasised

The All Ireland conference was supported by Sanofi Genzyme, Biomarin Pharmaceutical and Shire Pharmaceuticals

the importance of addressing the psychological aspects of pain alongside the medical aspects. While Katie spoke about how important psychology input is for children as they learn to cope with their condition and all the associated medical intervention. Katie also spoke about how she spends time with parents as they adjust to their child's diagnosis.

A look at the sensory aspects of MPS and Fabry management was followed by a Q&A session with an MPS mum, Maureen Walker, who spoke about how sensory processing problems affect her son's everyday life.

In our last session we were delighted to hear Dermott Devlin and Catherine McGrattan's accounts of living with MPS – these talks are always the most thought provoking parts of any conference. There's nothing quite as powerful as hearing real life stories from those who REALLY understand what it's like to live with MPS.

The conference finished with a look at Access to Medicines with Charlotte Roberts and Dr Fiona Stewart. This shone some light on the confusing world of drug development, clinical trials, drug approval and local commissioning of new medicines. We finished off with the take-home message that everyone needs to engage and get involved in the fight to access new medicines. Patient voices are key in campaigning and all those attending were encouraged to make their voices heard!

Thank you to all who were involved in the day and made it such a success.

Thank you!

I want to take this opportunity to thank all of our members and contacts across the Island of Ireland for all your well wishes over the last few months. I hope to return to work in the spring of 2017.

Clinics

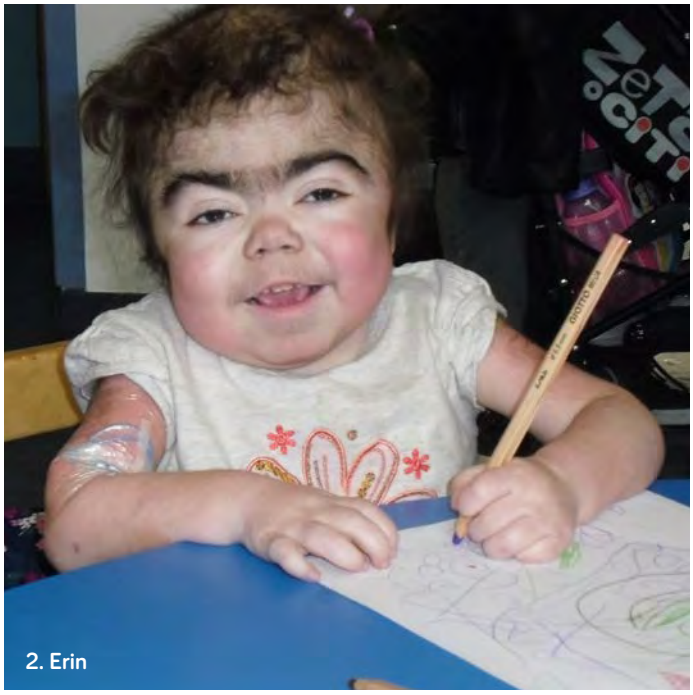
GOSH MPSIV 22 MARCH

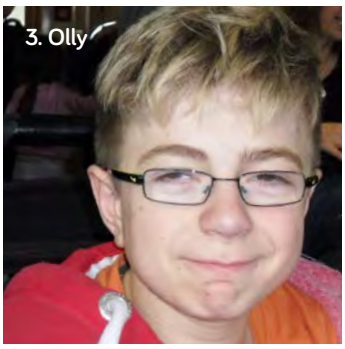
(1)

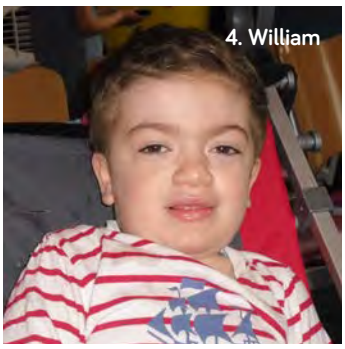

1. Kamal

GOSH BMT CLINIC 12 APRIL

(2-4)


2. Erin


3. Olly

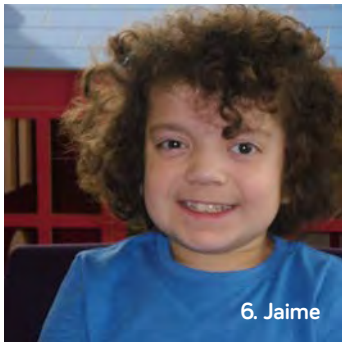

4. William

BCH BMT 6 MAY

(5-10)

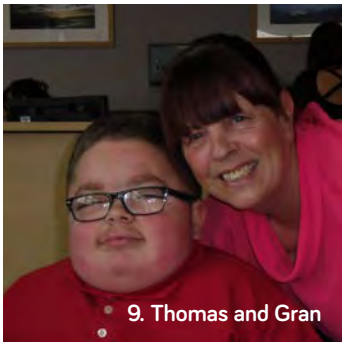

5. Cody


10. Charlie


6. Jaime


7. Luke


8. Melissa and Rachel


9. Thomas and Gran


12. Cameron, Connor & Alison


11. Charlotte


13. Freddie & Elsie

BCH FABRY 27 MAY

(11-14)


14. Kitty, Xanthe & Saskia

Time does seem to fly and before you turn around it's another clinic at Birmingham.

As always Birmingham is a very busy clinic in the outpatients and there is lots of activity and its normally a case of hunt the families!.

Luckily for me I do tend to know the members but it gets more difficult when we have new families who we have not met.

With it being the start of half term, most, not all, were happy to be breaking up early even if it meant coming to hospital.

It wasn't long before Vicki arrived with all four of her children and Kitty, Xanthe and Saskia were happy to oblige with a photo, older sister, Adriana decided against it.

Cheeky Freddie and Elsie are always a good bet for a photo.

Connor and Cameron came with mum and dad and mum, Alison, who was doing the London to Brighton walk the next day with her brother to raise money for MPS. 100k! Well done!

There were a few new patients and it was nice to meet them.

We had a quick break for lunch and then the afternoon clinic was in my favourite part of the hospital which is the research centre, with the interactive sensory flooring, great fun...

Charlotte arrived fresh from school with her mum and dad.

There were a few more members and then it was time to go home. Thank you to the team for looking after me and to the children who allowed me to take their photo and for making the clinic enjoyable.

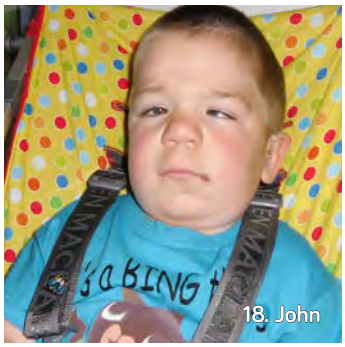
See you all next time.

Rebecca

BCH MPS 10 JUNE

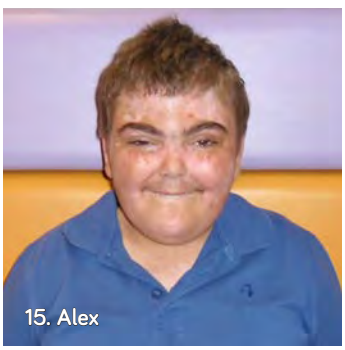
(15-20)


17. Hashim


18. John


19. Michael


16. Frankie


15. Alex

This was my first MPS clinic in Birmingham, it was a bright and early start to get there and the sun was shining. It was good to have Rebecca with me to show me the ropes, to ensure I found my way to and from the hospital, and introduce me to the professionals and families that I had not yet met.

It was a busy clinic and the organisation of the day impressed me. What impressed me more was the friendliness of the staff and brilliant attitudes of the children and parents as this was a long day for everyone, but especially the children.

Rebecca and my photography skills were put to the test, but we managed to get some lovely photos of most of the children. There was one particular child that was initially not willing to have his photograph taken, as his friend was not there, but he changed his mind, as he got used to us. I am glad he changed his mind, as Alex has such a cheeky smile, as I am sure you will agree.

As the day went on, Rachel took me to visit some of the families on the wards and I met some new members as well, which we look forward to supporting.

Louise


20. Gurkiran & brother

Birmingham Children's Hospital Metabolic Team

The Metabolic Team at Birmingham Children's Hospital cares for the whole spectrum of Inherited Metabolic Disorders. The IMD team consists of Consultants, trainee Doctors, Clinical Nurse Specialists, Dietitians, Biochemists and administrative staff. Birmingham Children's Hospital is one of the designated Paediatric Centres for children with Lysosomal Storage Disorders. For patients with a Lysosomal Storage Disorder, the team has designated Psychologists, Physiotherapist, Speech and Language Therapists. There is also a close working relationship with Neurology, Cardiology, Orthopaedics, Neurosurgery, Opthalmology, Respiratory, Dental & Stem Cell Transplant teams for patients with an LSD and these specialities participate in the MDT clinics and patient assessments, as required. As a result, these specialities have a wealth of experience and expertise to assist with the management of our patients.

At BCH we are fortunate to be able to offer multidisciplinary team (MDT) clinics. These enable patients to be seen in clinics on a single day by a number of specialists. The appointments are coordinated by the IMD Coordinator and helps to reduce multiple hospital visits. There are currently 12 designated LSD clinics scheduled throughout the year, to which the Patient Support Societies are invited. Currently BCH are building the first Paediatric Rare Disease Centre which will improve on the facilities offered to families travelling to BCH for MDT clinics. The Rare Disease Centre has been purposely designed to meet the needs of all patients accessing the facility including full accessibility for wheelchair users, a family kitchen, multi sensory rooms, youth room, 10 outpatient clinic rooms to allow full MDT clinics and an open waiting area that will allow families to network and meet with support groups.

The metabolic team offers 24/7 advice and support for their LSD patients, as required and the Clinical Nurse Specialists are available to provide support and advice during working hours. The Team meet weekly to discuss patients as a whole team, to ensure a multidisciplinary approach is taken. All Consultants are actively involved with all patients across the entire range of LSDs and patients and families get to know all the metabolic consultants over the course of their child's care.

The Metabolic Team have a good palliative care service and work well with Community teams to support patients requiring palliative support. The team also have a well established Transition service and link with University Hospital Birmingham and Addenbrookes Hospital to offer joint Transition Clinics for patients with an LSD. Young people are seen in a transition clinic at BCH between the age of 14 and 16 years and then are supported at an adult clinic, by the BCH team attending their appointment at the adult hospital.

Birmingham Children's Hospital is involved in many research studies ranging from; longitudinal surveillance studies, registries and early and late phase clinical trials of new drugs. We pride ourselves for being an active research centre that has recruited the first international participants onto registries and even first national participants on to complex clinical drugs trials. BCH is an accessible research centre for local, national but also international patients.

Consultants:
Dr Saikat Santra, Dr Suresh Vijay, Dr Julian Raiman

Clinical Nurse Specialists:
Rachel Gould, Louise Simmons, Catherine Stewart, Elaine Salmons, Kirsty Darling

Admin Staff:
Jenny Beardmore, Anna Hughes

IMD Coordinator:
Theresa Stokes

Research Staff:
Alice Stewart, Jitendra Sheinmar

Laboratory:
Tim Hutchin, Hayley Sherrod-Cole

Allied Health Professionals:
Emma Scobie (Speech & Language Therapist), Liz Wright (Physiotherapist), Rosie Jones (Dietitian), Shauna Kearney (Psychologist)

Consultants with Interest:
Alison James & Victoria Clarke (Dentists), Dr Evangeline Wassmer (Neurology), Mr Joe Abbot (Opthalmology), Dr Sarah Lawson (Stem Cell Transplant), Mr Guirish Solanki & Mr Desi Rodrigues (Neurosurgery), Shauna Kearney (Neuropsychology), Dr Ashish Chikermane (Cardiology)

Remembrance

A reflection of our lovely daughter Lorraine and her so charming brother Christopher who both had Morquio MPSIV

As parents you have so much love and hope for your children and you take on that role with great pride. But when you are told that your children have a life limiting condition it throws all your hopes into disarray. The emotional pain you feel goes so deep that you think it will never go away, as time passes you learn how to live with it and adapt to all the changes that their medical condition brings with it.

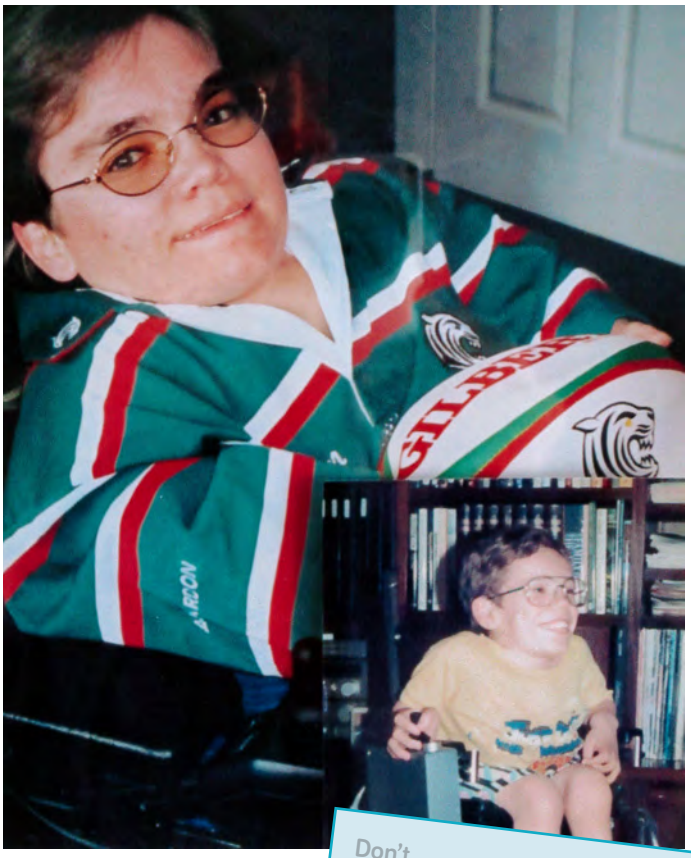
We said our sad goodbyes to Christopher in March 1998 at the age of 20. How Lorraine must have felt at that time and since must have been heart wrenching for her. Christopher was charming and good natured lad who would make you laugh at his jokes even if he did not get them quite right. While at college he showed a flair for IT work and his skill with recording equipment that earned him the nickname "fast forward". With his love for cars we were sure he could have become a racing correspondent or a racing driver in another life.

Lorraine showed from an early age that she was determined to do what she wanted and by and large she did. When her condition affected her mobility and she had to take to a wheel chair you could see the frustration in her, which at times would make her angry and difficult, but we learnt how to cope and win her over.

> **They both showed courage and determination to overcome the operations and hospital visits they had over the years, and they would be there to help support mum and dad and each other.**

When they were more able they travelled overseas with us to many countries. Their favourite destination was America where we travelled to various states. These trips where not always problem free and on one occasion Lorraine was stopped at Nashville airport by the FBI after they detected traces of nitro-glycerine on her. After a few hair raising minutes they realised that one of her meds contained the nitro-glycerine and it had shown in the palms of her warm hands. As the FBI cannot remove reported data, she became about 4 millionth on their wanted list which caused a few double looks on her passport checks at subsequent airports.

When at college Lorraine spent one day a week helping at a local charity called "Work Link" that helped people get back to work after being long term ill. They had an opening ceremony which the Honourable Terry Waite came to cut the ribbon. Lorraine had the honour of handing him the scissors and afterwards he chatted with her for some time, much to the annoyance of other dignitaries present.



Lorraine was a supporter of the Leicester Tigers Rugby for many years and would attend home matches until it became too difficult for her to go, and had to watch the games on TV. She also went to see England and the Tigers play at Twickenham, and was also invited by the then president Mr P Turnfield as guest for the day, and given a tour of the stadium and museum and over dinner together they discussed the England performance.

Don't

Don't leave me 'cause I'm small
Don't judge me by my looks
Just take me for the way I am
I'm one of you, but I'm made differently
Let me join in with your games

Don't push me aside
Let me be your friend
I don't want to sit alone
In the corner of the yard
In silence not noise
Watching you playing, having fun
Don't be afraid, accept me for who I am
Don't leave me in the dark.

Lorraine Rock, Aged 13

Sadly Lorraine passed away in January 2016 aged 41

We were so privileged to have two lovely children that grew up to become terrific adults and are missed so much.

Helen and Peter Rock

Bereavements

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

Elizabeth Kirkpatrick who suffered from Morquio and passed away on 8th February 2016 aged 45 years.

Helen Skidmore who suffered from Hurler and passed away on 26th May 2016 aged 37 years.

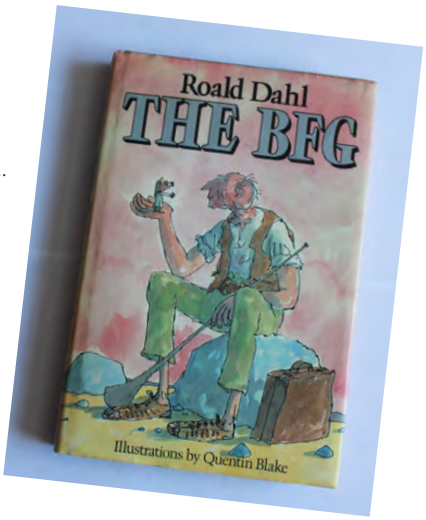
Your stories

Sally Mitchum and her family made the difficult decision to part with a very personal possession in order to raise money for the MPS Society. Here Sally explains why.

Once upon a time in a place not so far from here, a little boy was bursting with excitement. He had just met a real live author and now had a shiny new copy of a book signed by this famous man. His parents told him to look after the book as it was special, so he did. He read it carefully, marvelling at all the delumptious words, then he put it away on his bookshelf.

Years later, and that boy had grown into a man. He got married and had two boys of his own. As the eldest son grew and developed a love of stories, the man got down his book from the shelf and they started to read it together. Until this strange new invention called the interweb told the man that the book could be worth some money. The book was returned to the shelf and a less whoopsey-splunkers paperback was purchased.

Then the man's second son was diagnosed with a rotsome genetic disorder. They got through the next year somehow. One day, the man suggested to his wife that they sell the book to raise funds for the people who help his son, and his wife said it was a scrumdiddlyumptious idea.



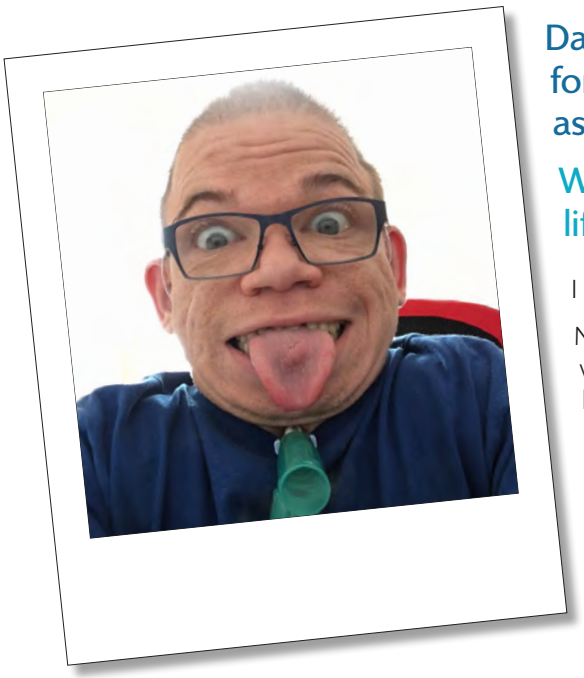
After all, what's the point of holding onto a book – however precious or important? Family is what really counts.

So, I'm becoming an expert on the condition of old books. Is it a first edition? Yes. Is it a first printing? Ooh, yes. Is that foxing or tanning on the upper edge? Bit of both. Any marks on the pages? A spot on the signed page and one on page 26. The level of detail that's needed is remarkable...

We've received an offer from one bookseller already, but can we do better? I don't know – watch this space.

Originally published on *A Hunter's Life - Learning to Live with MPSII*, June 2016, www.hunterslife.co.uk/bfg

If you know someone who might be interested in a first edition of *The BFG* or can spread the word please visit Sally's blog (www.hunterslife.co.uk) or email magazine@mpssociety.org.uk and we'll pass it on.



Daniel Newton supplied this fun photo for our Wear it Blue campaign so we asked him for a few words to go with it.

What we got was a real insight into the life of someone living with MPSII.

I was in Bolton Hospital from 1st April till 5th April.

My CPAP was beeping on that Friday morning and my mum and came in, I was cold, blue and wasn't breathing. My father got me breathing again but he couldn't wake me up. He was shaking me, poured a cup of water on my face but I still didn't open my eyes. The ambulance turned up and I woke when we were halfway near the hospital. They have to find out what is causing this. I fell asleep at the table in A&E when the doc was planning on sending me home and the machine started beeping, my oxygen saturation dropped very low and my face started turning blue. The doc understood what my mum was saying then.

They said that I had respiratory failure and the doctor said that I shouldn't be here now because I wasn't breathing for longer than 25 minutes.

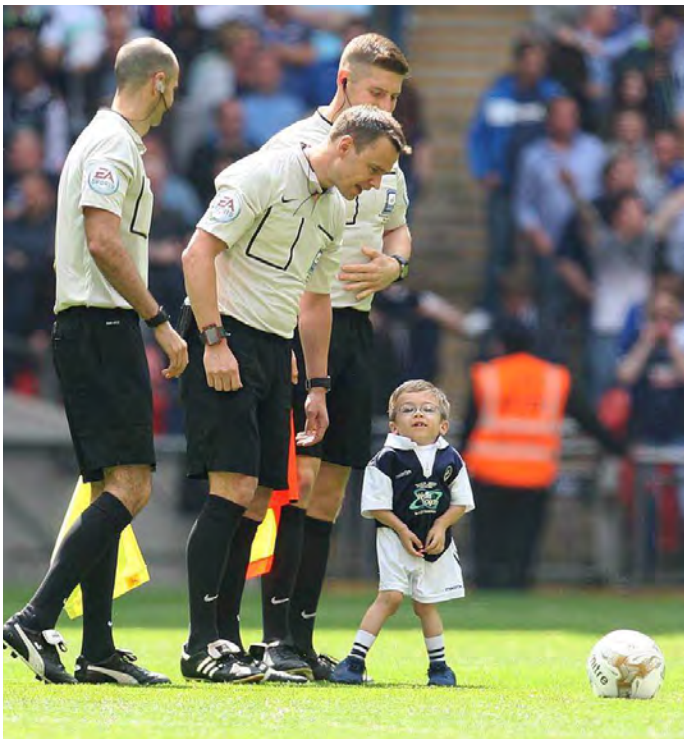
I am home now but am waiting for an echo heart scan appointment. I am still staying strong for my family...I may have lost the battle but I haven't lost the war.

Harvey Brown chosen as Millwall mascot at Wembley

Harvey Brown, lifelong Millwall Football Club supporter, was chosen as their mascot for the League one play-off final against Barnsley at Wembley. Harvey, who has Morquio, was chosen to be the Millwall mascot and went out with the team, led by Tony Craig, Millwall's captain. He travelled with the club to Wembley and after smiling and waving to fans from the pitch he got to watch the match from Millwall's dugout and then explore the famous grounds after.

Harvey's dad, Dean, said "I was so proud of him. He stood there before the match on the pitch like a little soldier" and mum, Vikki describes Wembley as amazing: "they made Harvey feel like he was on top of the world. He was so excited and treated like a superstar. The fact Millwall didn't win didn't spoil the day he still came back on the Millwall bus singing songs."

> "They made Harvey feel like he was on top of the world



Our first ML Conference

The MPS Society's first conference focussing on Mucolipidosis was held on the 9th and 10th April 2016. It offered individuals, parents, partners, carers and professionals the chance to listen to presentations from experts in the field, as well as from those who have been directly affected by an ML. It also gave families and professionals an opportunity to share experiences and knowledge.

There was a mix of presentations from professionals and expert speakers as well as from families affected by ML. The speakers were all highly received and here are some of the highlights of the weekend.

> "The most important part of the meeting was getting to meet other families and share similar experiences that other families wouldn't understand. It can be a lonely life living with a rare condition, but when you meet other families, they become your support network and share in both the good and not so good times.

Fiona Stewart – Genetics of MLII and MLIII

Fiona is a Consultant in Genetic Medicine at Belfast City Hospital. Her presentation on the genetics of MLII and MLIII explained the science using easy to understand analogies including spelling, cars and even baking a cake!

Looking at chromosomes is a bit like looking at a book shelf to see if any books are missing. Looking at single gene disorders is like seeing if there is a spelling mistake anywhere in any of the books. For some diseases everyone has the same gene mutation so it's a bit like the spelling mistake always being in the same place, for others there are many different mutations so you have to look all the way through the book.

Until now clinicians have had a clinical suspicion of a diagnosis in a patient and have then done a test to confirm this. Usually testing enzyme levels first before genetic testing. Now we have panel tests that can look at man different genes at once e.g. Manchester panel has more than 200 genes known to cause metabolic disease. These tests are useful when the diagnosis is not obvious or where a number of different conditions can give a very similar clinical picture.

John O'Connor – Our life with MLII/III

John is dad to three girls including Aoibhe who has ML. She was just like a normal new born and even very normal up to six or seven months, where we noticed a curve in her spine. Dr Tangeny who we now owe a great deal, diagnosed her at 9 months. Other cases we have read about weren't diagnosed until they were 2 or 3 years old. He spent some time checking Aoibhe and had seen things that we'd never seen. We made another appointment for a week later and we both went. I remember him looking very sorry for us, he got a huge Medical text book up on the table and took out a photocopied page from it and handed it to us. He told us

Thank you to Blake's Genes who supported the event.

> It's always an amazing opportunity to 'pick the brains' of professionals at these kinds of conferences.

that Aoibhe had MLIII and told us to read the information on the page and there was more information on the internet, but that's all he could tell us because he had never seen it before because it was so rare. We were sent for a skin biopsy to make sure Aoibhe had ML, the biopsy was sent for testing and it came back that Aoibhe didn't have MLIII, but was more rare again, it was in between MLII and MLIII, or as we call it ML 2.5.

Since then we have had numerous trips to Temple Street and Crumlin Children's hospital in Dublin for MRI and x-rays and operations. Her airway is very narrow and getting the tube in is getting more difficult every time she needs these operations.

Aoibhe is now 9 years old and is a happy child. She is only around 81cm in height and weighs 13kg. She's quite bright and is a year to a year and a half behind her peers. She finds it hard to get from a sitting position to standing, she can't straighten her fingers and finds it hard to grip, she can't reach above shoulder level and her balance can be affected due to her curve in her spine.

She loves going to play parks and is sports mad. She plays Boccia and attends a sport group for the physical disabled. Aoibhe is very good at finding a way to do what is normal to another child, it might take a while, but she finds a way.

Sarah Burgess – Living with MLIII

Sarah is a part-time youth worker living in Luton. She is 34 and was diagnosed with MLIII at 5 years of age. In her presentation she talked about what it is like living with MLIII including the diagnosis process, her various surgeries, the treatment she receives and her own 'keeping my marbles guide'. Her guide covered mental health issues, coping strategies, having support, asking for help and being allowed to say no as well as yes to things in her life.

Jenny Noble – Managing bone density & pain with Pamidronate infusions

Jenny is currently the Field Officer/Administrator for Lysosomal Diseases New Zealand and is Vice President for The International Advocate for Glycoprotein Storage Diseases (ISMRD). She is also a parent of two adults with the rare metabolic disease Mucolipidosis III alpha/beta.

Her presentation talked about the symptoms her children suffered with such as chronic pain, chest infections, difficulty sleeping, poor concentration and how many different drugs for pain just didn't work. Until a light bulb moment at an MPS meeting in Australia when a presentation about pain management in Sanfilippo patients brought up the idea of bisphosphonates and Pamidronate, a drug usually used to treat osteoporosis. She described a quick response to the treatment:

- Pain Free after first infusion
- 3 months treatment – Sarah walks again
- 6 months treatment – Sarah walks unaided
- 18 months treatment Hayden walked with a walking frame
- Sleeping through the night smiling again

Brian Bigger – The challenge of developing a therapeutic treatment for MLII/III

In 2006 Brian set up the Mucopolysaccharide Disease Stem Cell Research laboratory (now the Stem Cell & Neurotherapies group) at the University of Manchester in collaboration with Dr Rob Wynn, the director of the Blood and Marrow Transplant Unit at the Royal Manchester Children's Hospital and Professor Ed Wraith, the director of the Genetic Medicine Unit at St Mary's Hospital.

His presentation concluded that:

- A direct gene therapy approach to target as many cells in the body as possible – but particularly the bones and brain – will probably be the most effective approach for MLII/III.
- Chaperones and stop codon read through may provide some benefit to subsets of patients (missense and nonsense mutations respectively) but these approaches are often marginal.
- Management of specific symptoms of disease via bisphosphonates or perhaps specific targets of the immune system could be beneficial.
- Ultimately combination therapies are likely to be the best approach for MLII/III.

> Thank you for putting on this conference for Mucolipidosis – a disease often overlooked, both in MPS conferences as well as research.

But what about the children?

As no children under 18 years were allowed into the conference, the Society made alternative childcare arrangements so parents and carers could attend the conference in a relaxed manner, knowing that their children are safely cared for and entertained.

Some of the childcare took place on-site including a sensory area, a play area, arts and crafts, music time, build a bear and memory making. A few children went on outings to the cinema and bowling

Thank you so much to our volunteers, Sophie, Helen, Matthew, Ahmed, Anna and Veronica, who did an amazing job looking after the children over the weekend.

> **"I think it was Veronica and Helen who kept them busy and safe. Please pass on our thanks to them. The boys liked them both, probably because they let them loose on the Pick & Mix.**

> I would like to specially thank the volunteers for their time entertaining Zoha at the creche. Without their support I don't think either of us would have been able to attend and gain as much as we learnt from this meeting.

> It was a very informative event, we learnt from every session. It was also a great opportunity to meet other affected families, share our stories and learn from each other.

> Well organised, very informative, really appreciated. Thank you.

Gulliver's Land

Giant adventures on MPS Awareness Day

Milton Keynes was engulfed in a sea of cyan as our members and staff met at Gulliver's Land for MPS Awareness Day.

We wanted to do something special as 15th May fell on a Sunday this year so we organised a trip where our families could meet up and enjoy lots of rides.

The day started with an exciting song and dance from the Gulliver's Land mascots, Gilly and Gully, before the ribbon was cut and the azure army descended on the theme park.

We had a brief get together in the Main Street Theatre, giving everyone the chance to chat and ask questions about the day, then crowded outside Lilliput Land Castle for a photo opp – and what a great shot we got. Everyone really got in the awareness mood by sporting every shade of blue going!

The day was then ours to do with as we pleased. There were so many rides to choose from that we didn't know where to start, the carousel, The Python, the Flying Carpet, the Runaway Train? But we soon got round them all and it was great to see others from the MPS Society around the park, identified by their aquamarine attire.

We definitely made the most of the day staying until the park closed and riding as many rides as possible that were guaranteed to make you sick with dizziness. We didn't make it to the Dinosaur & Farm Park but I hear it was great fun and there were some very cute animals to cuddle.

As a new member of staff at the MPS Society this was the first opportunity I'd had to meet anyone with MPS or a related disease. My early days at the Society were of course spent familiarising myself with the diseases and their symptoms but this doesn't really give you an idea of what it's like to live every day with MPS or Fabry. It's hard. And yet all the families I met were so positive and up beat and were just having fun. Just getting on with life. I think you are all amazing and I look forward to seeing you again at the next event we organise.

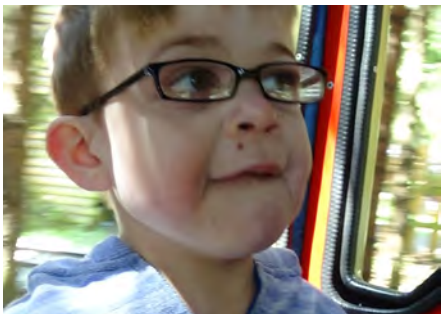
But a word of warning for the next trip to Gulliver's Land – avoid the cycle monorail, boy did I feel it the next day!

Helen Crawley
Fundraising & Information Officer

Thank you to Sanofi Genzyme, BioMarin Pharmaceutical and Shire Pharmaceuticals who sponsored our trip to Gulliver's Land.

“We would all like to thank the MPS Society for arranging the wonderful day at Gulliver’s Milton Keynes we all had a great time making many memories that will stay with us forever, it was lovely meeting new families so we don’t feel so alone.

Trudy, Nick, Mollie and Mason Goodwin



International

Reports of breaches of the ABPI code of practice at the Fabry International Network (FIN) meeting

In the interests of transparency and, as reported to the Fabry International Network (FIN) membership at its Annual General Meeting in Japan in May, FIN has inadvertently been linked to a complaint made by Sanofi Genzyme about Amicus to the Prescription Medicines Code of Practice Authority (PMCPA) alleging breaches to the Association of British Pharmaceutical Industry (ABPI) Code of Practice.

The events that led up to this complaint being made occurred at the FIN Expert Meeting held at Latimer House, England, 20–22 November 2015. FIN invited in good faith, and at its own expense, two representatives each from Shire, Sanofi Genzyme and Amicus to present and participate in this meeting.

FIN first became aware that their Expert Meeting had been used in this way when several people saw the 'Fabry International Network' name associated with the case AUTH/2809/12/15: on the PMCPA website (www.pmcpa.org.uk). Whilst FIN accepts that regulations are in place to protect the reputation of the pharma industry and indeed patients this FIN Expert Meeting was NOT a patient meeting and in the ultra-rare disease field individual pharmaceutical companies play an important role working with the patient organisations to ensure they have accurate and up to date patient relevant information.

Immediately prior to the start of the FIN Expert meeting, members of the FIN Board attended the Amicus opening of their European Office. Amicus had some leftover champagne and offered it to me for FIN. As I was not driving, for health reasons and could not lift, it was agreed the Champagne was transported to the FIN Expert meeting where it was used at the FIN 10th Anniversary dinner on Saturday night to provide each delegate, including the pharma industry delegates, one glass each. Genzyme reported this as a breach of the ABPI Code but in inter alia company discussions Genzyme accepted an apology from Amicus that this was a one off and would not happen again! Nevertheless this left a 'trust' issue in many of our minds. In monetary terms each delegate technically received a gift of less than €5! I am sure there is not one FIN member who felt this one glass of champagne was an inducement. I think we are made of stronger morals than that!

More seriously, Genzyme alleged further breaches against Amicus including promotion of an unlicensed medicine and failure to show reference numbers on the presentation raising concerns over a robust review and approval process from appropriately qualified personnel. Genzyme also alleged that the breaches were broad and gross in scope constituting a failure to maintain high standards and undermine the standing of the pharmaceutical industry.

Amicus provided the PMCPA with a detailed response in which it refuted the allegations made by Genzyme stating that no clinical results from the studies were given in the presentation and the audience was high level representatives from global Fabry patient associations.

On appeal, the PMCPA Panel considered the statements and discussion about amenable mutations and the implied positive regulatory status of Migalastat. Although much of the information is in the public domain, on balance the appeal board considered that the presentation had raised the prospect of a new treatment for Fabry patients with amenable mutations and, in that regard, had promoted Migalastat prior to the granting of a marketing authorisation.

The appeal board noted its ruling above and considered that as the promotional presentation was not formally certified it upheld a breach of the Code. The appeal panel also considered that as the presentation was aimed at patient organisations and had not been formally certified it also upheld a breach of the Code.

The appeal board noted its comments and rulings above and considered that high standards had not been maintained and consequently upheld a breach of the Code.

Although noting its comments above, **the appeal board however did not consider that in the particular circumstances of the case a ruling of a breach of clause 2 – discredit to, and reduction of confidence in, the industry – had taken place**

The FIN Board met to discuss how the PMCPA had without even contacting us published on their website during the investigation and appeal process the full name of the Fabry International Network as the meeting where the then alleged breaches took place. At their meeting in Tokyo in May the FIN Board agreed to write to Heather Simmonds, Director of the PMCPA requiring FIN's name to be removed from the website and from the final published report. I am pleased to report that the PMCPA have now removed all reference to FIN in the report now uploaded to the PMCPA website and to be published in the May Code of Practice Review.

Not directly related to the matters above, to those of us working in the ultra-rare disease field, patient organisations want to embrace and welcome further drug development and a professional working relationship with pharmaceutical companies is essential. Increasingly, this means new generations of drugs whereas ten years ago there was only one drug per disease or none at all. It is therefore an anathema that the ABPI Code can be used as a vehicle for anti-competitive behaviour by bigger and stronger pharmaceutical companies against smaller pharma companies doing their best to find treatments for ultra-rare diseases in this important rapidly emerging field for patients affected.

In truth nothing was shown or said by Amicus back in November that most of the Fabry patient organisation leaders didn't already know! Has the

ABPI and PMCPA not heard of the internet and social media? Today we don't need to travel on the Queen Mary to gather information from across the Atlantic. How can it be that American ultra-rare disease patients can access all the information they need on current treatments and clinical trials but in Europe we are all considered so corruptible that we need protecting from ourselves through a flawed ABPI Code of Practice?

In fact things are much worse. As Group Chief Executive of the MPS Society I employ four highly skilled social workers who are members of and regulated by the Health and Care Professions Council and yet, because they are employed (paid) by the MPS Society, these colleagues who advocate for patients (our members) day in day out are NOT recognised as healthcare professionals by some pharmaceutical companies fearful of the ABPI Code of Practice. This is in contradiction to a healthcare professional working for a patient organisation but whose salary and employment is channeled through an NHS Trust but does the same job. This discriminatory behaviour cannot be allowed to continue.

History has shown that regulation of the pharmaceutical industry is necessary but for ultra-rare disease patients and their patient organisations perhaps this should be left to the Medicines and Healthcare Products Regulatory Agency (MHRA), a professional organisation and equipped for the role, overseeing compliance of the Medicines Act 1968.

If I was the CEO of a Biotech Company or small pharmaceutical company with an eye to the European market for ultra-rare diseases today one thing is for certain I would not undertake to comply with elements of the ABPI Code of Practice outside of the Medicine Act 1968 or join the self-regulating ABPI relying on the funds of big pharma and the fines of penalised smaller pharma for its existence.

Christine Lavery

President of the
Fabry International Network

Group Chief Executive of the MPS Society

Fabry International Network Expert Meeting in Japan

20–22 May 2016

From the evaluation forms and talking to everyone the FIN Expert Meeting in Tokyo was as huge success and this was somewhat of a relief for our FIN Co-ordinator, Toni Ellerton, and the FIN Board as it was a great challenge to organise this meeting at such a distance. Indeed it wouldn't have been possible without the help we received from Professor Eto, Mr Harada and the Japan Fabry Disease Patients and Association, (JFA) team. *Read the full story on p.24.*

The FIN Board arrived in Japan in time to have a productive full day of one to one face to face meetings with representatives of Shire, Genzyme and Amicus. These were followed by a closed meeting of FIN members who gathered for the FIN Annual General Meeting. Although not many attended the AGM those that did asked valuable questions and scrutinised the Director's Report and Financial Report. The re-election of Directors was held and I am delighted that Lut De Baere, Anna Meriluotoand Jack Johnson were re-elected. Lut took over the Chair to oversee the appointment of Vice President and President. Jack Johnson was duly appointed Vice President and Christine Lavery, President.

> "Medical students, junior doctors and consultants chose to give up their Saturday to increase their knowledge of Fabry disease.

By the time we all gathered for dinner on the 33rd Floor of the Prince Park Tower Tokyo Hotel nearly all the 51 FIN members and speakers had arrived. With a glass of Champagne, donated by the hotel management, in hand the FIN delegates were welcomed to Tokyo by Christine Lavery and Hasio Harada. As the evening came to a close Professor Yoshikatsu Eto, Director, Jikei University, gave an enthusiastic insight into how to enjoy the sights of Tokyo and beyond. Sadly most of us will have to return again if we are to savour these wonderful sights and enjoy the amazing Japanese hospitality.

The FIN Expert Meeting was held at Jikei University and started promptly with an introduction to Fabry disease by Professor Eto. This set the scene for presentations by Professor Atul Mehta (UK), Dr Dominique Germain (France), Dr Dau-Ming Niu (Taiwan), Dr Uma Ramaswami (UK), Dr Derralynn Hughes (UK) and Professor James Moon (UK) to speak on the clinical aspects of Fabry disease and there treatment. Professor Moon described some innovative work he is pioneering at University College Hospital, London on advance cardiac imaging of the Fabry heart. Megan Fookes (Australia) shared her experience of Fabry disease in children.

As well as the speakers and FIN members this meeting was opened to 40 Japanese medical students, junior doctors and consultants who chose to give up their Saturday to increase their knowledge of Fabry disease. This initiative came at the request of Mr Harada and is certainly something FIN should consider doing in the future when the programme is medically orientated as part of our educational programme.

On Saturday night a buffet dinner was held at the Green Rattan restaurant in Roppongi Hills a lively part of Tokyo. I would like to say our FIN members then worn out made their way back to the hotel but I can't! Led astray by Mr Harada and some FIN Board Members many headed off to a Karaoke Bar to sing their hearts out and then try and find their way back to the hotel. (Jack and Christine preferred the bar on the 33rd floor back at the hotel!)

On Sunday most of the delegates took early flights or had a well-earned lie in. For many of the FIN Board and some speakers it was back to Jikei University to make presentations at the Fabry Tokyo Symposium 2016.

So you might well ask were does FIN's Expert meeting go from here? It has been agreed we return to Europe in May 2017. The precise country and venue is currently being worked on and will be announced soon.

Research & Treatment

MPSI and MPSII Regenxbio

REGENXBIO Inc. is a leading biotechnology company focused on the development, commercialization and licensing of recombinant adeno-associated virus (AAV) gene therapy. The company's mission is to transform the lives of patients suffering from severe diseases with significant unmet medical needs by developing and commercializing in vivo gene therapy products based on our NAV Technology Platform.

MPS I

RGX-111 is REGENXBIO Inc.'s product candidate for the treatment of Mucopolysaccharidosis Type I (MPS I). Individuals with MPS I have a deficiency of alpha-l-iduronidase (IDUA), an enzyme found throughout the body, including in cells in the central nervous system (CNS) that is responsible for the breakdown of polysaccharides. RGX-111 uses the AAV9 vector to deliver the IDUA gene to the CNS creating the potential that normal IDUA enzyme can be produced. Delivery of the gene encoding the enzyme could provide a permanent source of secreted IDUA beyond the blood-brain barrier, allowing for long term cross-correction of cells throughout the CNS. This strategy could also provide rapid IDUA delivery to the brain, potentially preventing the progression of cognitive deficits that otherwise occurs in MPS I patients. The U.S. Food and Drug Administration (FDA) granted Orphan Drug Designation and Rare Pediatric Disease Designation to RGX-111 for the treatment of MPS I. Initiation of a Phase I/II clinical trial is planned for mid-2016.

MPSII

RGX-121 is REGENXBIO Inc.'s product candidate for the treatment of Mucopolysaccharidosis Type II (MPS II), also known as Hunter syndrome. Individuals with MPS II have a deficiency in the lysosomal enzyme iduronate 2-sulfatase (IDS), an enzyme found throughout the body, including in cells in the central nervous system (CNS) that is responsible for the breakdown of polysaccharides. RGX-121 uses the AAV9 vector to deliver the human IDS gene to the CNS creating the potential that normal IDS enzyme can be produced. Delivery of the gene encoding the enzyme that is deficient within cells in the CNS could provide a permanent source of secreted IDS beyond the blood-brain barrier, allowing for long term cross-correction of cells throughout the CNS. This strategy could also provide rapid IDS delivery to the brain, potentially preventing the progression of cognitive deficits that otherwise occurs in Hunter syndrome patients. The U.S. Food and Drug Administration (FDA) granted Orphan Drug Designation to RGX-121 for the treatment of MPS II.

For more information, visit http://www.regenxbio.com/

MPSI and MPSII Armagen, Inc.

MPS I/Hurler Syndrome Trial

ArmaGen, Inc. is conducting a Phase 1 study of its investigational enzyme replacement therapy for Hurler syndrome, AGT-181.The study aims to test the safety and determine a well-tolerated dose of the medication in adult male patients with Hurler-Scheie and Scheie syndromes. ArmaGen's reimbursement plan addresses travel, subsistence (meals and lodging) and stipend (given by study site institution directly to participant) based on specific patient needs.

Additional information is also available at www.clinicaltrials.gov using the identifier number NCT02371226.

Currently approved treatments for Hurler syndrome are unable to cross the blood-brain barrier (BBB), a filter that prevents toxins as well as therapies from entering the brain. As a result, they do not address many of the severe and neurological complications of Hurler syndrome. AGT-181 is designed to cross the BBB in the same way insulin does.

MPS II/Hunter Syndrome Trial

ArmaGen, Inc. is conducting a Phase 1 study of its investigational enzyme replacement therapy, AGT-182, in adult male patients (18 years and older) with Hunter syndrome. The study aims to test the safety and determine a well-tolerated dose of the medication. ArmaGen plans to enroll 12 patients into the study. The first cohort/group of four patients has been enrolled, and the company is now enrolling the second cohort.

ArmaGen's reimbursement plan addresses travel, subsistence (meals and lodging) and stipend (given by study site institution directly to participant) based on specific patient needs.

Currently approved treatments for Hunter syndrome are unable to cross the blood-brain barrier (BBB), a filter that prevents toxins as well as therapies from entering the brain. As a result, they do not address many of the severe and neurological complications of Hunter syndrome. AGT-182 is designed to cross the BBB in the same way insulin does. AGT-182 is designed to treat both the body-related (somatic) and central nervous system (CNS) symptoms and complications of Hunter syndrome.

Shire's Humanitarian Programme for MPSII and Fabry Disease

It is generally accepted that people affected by ultra-rare diseases across the globe face challenges getting reimbursed high cost market approved treatments.

Like many LSD patient organisations across the World the MPS Society receives 100–200 emails each year from patients and their families pleading for help to access enzyme replacement therapy (ERT) for MPSI, MPSII, MPSIVA, MPSVI and Fabry disease. For most we can only respond with a level of empathy, kindly advice about joining forces with others in their country similarly affected and engaging with clinicians and the Departments of Health. Many of these enquiries come from the Middle East, India, Pakistan and Bangladesh where mistakenly people are under the impression that everyone else in the World is receiving reimbursed ERT unhindered. As many families even in Australia, New Zealand, Canada, South Africa and parts of Europe know this could not be further from the truth.

But there is a chink of light for some MPSII and Fabry patients thanks to the Shire Charitable Access Programme which is inviting clinicians in Ecuador, Dominican Republic, Egypt, Albania, Belarus, Tunisia, Malaysia and Pakistan to apply for their patients online at:
www.directrelief.org/rare-diseases/mps-ii-hunter-syndrome
or www.directrelief.org/rarediseases/fabry-disease

- If you are a patient with MPSII, Hunter disease in Ecuador, Dominican Republic, Tunisia, Egypt or Pakistan please tell your doctor about the Shire Humanitarian Programme and ask the doctor to apply
- If you are a patient with Fabry disease in Ecuador, Dominican Republic, Tunisia, Egypt, Albania, Belarus, Malaysia or Pakistan please tell your doctor about the Shire Humanitarian Programme and ask the doctor to apply
- If you are a patient organisation, doctor or relative who knows of eligible MPSII and Fabry patients in these countries please tell them about the Shire Humanitarian Programme

How to Apply

Physicians must complete the online application in order for their patients to be considered for enrolment. The applications will be reviewed by an independent Medical Expert Committee (MEC) and the physicians will be notified regarding their submissions after the MEC makes their decision. Shire and Direct Relief are not involved in the patient selection process.

Eligibility for MPSII Hunter disease

Physicians are eligible to apply on behalf of their patients if the patient resides in a country where Shire's humanitarian program is available and the patient has been diagnosed with MPS II (Hunter syndrome).

Inclusion Criteria

- A confirmed diagnosis of MPS II by documentation of absent or very low (<10%) I2S (iduronate-2-sulfatase) activity.
- Documentation of normal enzyme activity of at least one other sulfatase is required, as low levels of I2S activity are also characteristic of multiple sulfatase deficiency.
- Evidence of the following disease manifestations:
- Cardiac involvement evidenced by valve disease, ventricular hypertrophy, or arrhythmia
- Airway disease as evidenced by abnormal PFTs, obstructive airway disease or sleep apnea
- Decreased Joint Range Of Motion (JROM) or Joint arthropathy or disease limiting the activities of daily living (spine deformities, contractures, joint stiffness and claw-like hands)

Exclusion Criteria

- Patients with an advanced or a non-responsive neurologic state that would be unlikely to benefit from therapy.
- MPS II patients diagnosed with any condition that would prevent them from experiencing clinical benefit with enzyme replacement therapy.

Eligibility for Fabry Disease

Inclusion Criteria

The applicant must meet the following criteria to be eligible for participation in this Program:

- Applicants must have a genetically confirmed diagnosis of Fabry disease. Patients with a plasma GAL activity 1.5nmol/hr/mL or leukocyte GAL activity 4 nmol/hr/mg are eligible if their Fabry disease can be documented by genetic analysis, positive histopathology, and/or a family history of the disease.
- Applicants must have symptomatic Fabry disease as reported by the treating physician, for example: significant abnormalities in renal function, cardiac function, or cerebrovascular function, peripheral neuropathy, severe proteinuria and/or severe pain or crises.
- Female applicants of childbearing potential must have a negative pregnancy test prior to starting Replagal infusions. Periodic pregnancy testing should be performed as medically indicated. The therapy of pregnant or lactating females should only be considered in severe circumstances.

Exclusion Criteria

- Female applicants who are pregnant or nursing
- Applicants who have a clinical condition or health problem that may prevent any potential benefit of therapy (as assessed by the INCAP Medical Advisory Board).
- Applicants who are currently on another enzyme replacement or substrate inhibition therapy.

Lysogene launches a prospective, observational study of MPS IIIA

Lysogene has launched a prospective, observational study of patients, who are aged nine years old or younger, with mucopolysaccharidosis IIIA (MPS IIIA) to better understand their health problems and how to measure these problems over time. This is an observational study, so no experimental drug will be given. The study is planned before the start of Lysogene's gene therapy trial to function as a non-concurrent control. The information to be learned will help lead to faster and better trial design and understanding future therapeutic effects.

Study details

- Patients should have a confirmed diagnosis of MPS IIIA and be aged nine years or younger
- Patients participating in the study will attend the study site, for a day or two, every 6 months for up to a maximum of 2 years. Clinical study sites have been selected in France, UK, Germany, and the Netherlands. Patients might be eligible to participate from other geographical locations (more information on the clinical study sites).
- Neuropsychological tests will be completed to measure your child's cognitive development which include exercises, games and quizzes. These help measure your child's attention span, language skills, visual organization, memory, and learning abilities.
- Questionnaires about your child's behaviour, quality of life and your child's sleep habits will need to be completed every three to six months.
- Travel and study related expenses will be reimbursed as set out by a reimbursement policy.

What Next?

- If you decide that you would like your child to participate in this study, we will look at your child's medical records and collect your child's medical history.
- Individuals included in the observational study will be provided the opportunity to enroll in subsequent trials if they satisfy the specified enrolment criteria. However, please note that participating in this study does not guarantee a place in the clinical trial

Lysogene is currently planning a gene therapy clinical trial with its next generation formulation LYS-SAF302 to start in 2017 and more details will be made available in due course.

Off to university?

Recommend MPS Society to your RAG committee and get your institution fundraising for MPS.

BioMarin MPS IIIB (Sanfilippo B syndrome) study information for families and patient groups

BioMarin has recently started a programme for children with MPS IIIB (Sanfilippo B syndrome). This will involve multiple centres around the world. The program will enrol children between the age of 1 and 10 years who qualify, although the majority of children will be under 6 years old. The program consists of two studies.

The first study is an observational study of children with MPSIIIB and includes testing of cognitive and adaptive function, as well as assessments of behaviour and quality of life. This study is intended to provide baseline information to help in determining whether these children subsequently benefit from treatment in the next phase of the programme. This observational study is now open.

In the near future, BioMarin plans to start the second study. The second study will be a treatment study in which children will receive an investigational enzyme replacement therapy. This will be provided as an infusion via a port which delivers the enzyme into the brain. This study will be run at the same centres as the observational study. The first phase of this study will enroll a small number of patients in early 2016 to assess safety at several doses. The second phase of this study is targeted to start in mid-2016. In order to enroll in the second phase of the treatment study the child must have completed the observational study or the first part of the treatment study. The intention is to provide treatment to all of the children who have entered into the observational study.

Enrolment in this program will be limited to children from countries in which there is a study site. Please visit www.clinicaltrials.gov for more details.

Amicus Therapeutics Announces European Commission Approval for Galafold™ (Migalastat) in Patients with Fabry Disease in European Union

On the 31 May 2016 Amicus Therapeutics announced that the European Commission granted full approval for the oral small molecule pharmacological chaperone Galafold™ (migalastat) as a first line therapy for long-term treatment of adults and adolescents aged 16 years and older with a confirmed diagnosis of Fabry disease (alpha-galactosidase A deficiency) and who have an amenable mutation. Amicus has already begun supplying the market in Germany and has commenced the reimbursement process with NICE and other healthcare authorities in major European countries.

Galafold is the first oral treatment as well as the first precision medicine for Fabry disease. The broad label includes 269 Fabry-causing mutations which represent between 35 and 50 percent of all patients with Fabry disease.

> "The EU approval of the first oral precision medicine for Fabry disease is a major step forward for patients in Europe.

John F Crowley, Chairman and Chief Executive Officer of Amicus Therapeutics, Inc. is quoted as saying, "This EU approval for Galafold is a significant advancement in the field of precision genetic medicine and a tremendous milestone for the Fabry community."

The European Commission approval was based on clinical data from two Phase 3 pivotal studies in both treatment naïve Study 011 (FACETS study) and enzyme replacement therapy (ERT) switch patients Study 012 (ATTRACT study), as well as ongoing long-term extension studies. AS it is known Fabry disease is a rare genetic disease and potentially life-threatening condition caused by the accumulation of disease substrate (globotriaosylceramide, GL-3) in the lysosome due to a dysfunctional or deficient enzyme. Galafold works by stabilizing the body's own dysfunctional enzyme, so it can clear the accumulation of disease substrate in patients who have amenable mutations. An amenable mutation is one that is responsive to therapy with Galafold based on predefined criteria.

"As principal investigator in both Galafold pivotal studies, I have experience treating both naïve and treatment-experienced Fabry patients with Galafold," said Derralynn Hughes MA DPhil FRCP FRCPath, Senior Lecturer in Haematology at University College London, UK with clinical responsibilities in haematology and lysosomal storage disorders. "I am pleased that the European Commission has approved this new treatment option and I believe it has the potential to address unmet needs among Fabry patients who have amenable mutations."

"The EU approval of the first oral precision medicine for Fabry disease is a major step forward for patients in Europe," said Christine Lavery, President of the Fabry International Network (FIN). "We appreciate Amicus' commitment to the Fabry community and its dedication to develop high quality therapies for Fabry disease. For the first time in more than a decade, patients with Fabry disease who have amenable mutations now have a choice for an innovative new treatment option."

This information is taken from a comprehensive Press Release issued by Amicus Therapeutics on 31 May 2016 and published in full on the MPS Society website: www.mpssociety.org.uk

Patients/Families Impacted by MPS I and MPS II:
Please Participate in a Market Research Interview!
$120 (USD) Honorarium Provided

***Topic*:** How patients and families impacted by MPS I and II make treatment decisions, and their reactions to a potential new therapy

***When*:** Interviews will be conducted between now and mid-July by telephone *and* at the 14th International Symposium on MPS and Related Disease that is taking place July 14-16 in Bonn Germany.

Eligibility: To participate, you must be….
1) A patient or parent of a patient diagnosed with MPS I who:
-Has undergone bone marrow or hematopoietic stem cell transplant (BMT of HSCT) within the last 5 years
OR -Is currently in the process of deciding to undergo a BMT or HSCT
2) You are a patient or parent of a patient with MPS II who;
-Has been diagnosed within the last three (3) years
AND -Has experienced cognitive / intellectual impairment due to the condition

To Participate: visit the following link to arrange an interview time;
http://tinyurl.com/MPS-RSVP
Or email marketresearch@engagehealth.com

ENGAGE

Information & Resources

Disability Rights UK have published an employment skills guide for people with newly acquired disabilities or health conditions.

Get back to where we do belong will help those exploring their education, training and employment options to learn how to manage change, what to expect from employers, where to access training and much more. With practical advice and personal stories of people's own experiences it's a helpful and reassuring guide. Download it here: www.disabilityrightsuk.org/sites/default/files/pdf/GetBack16.pdf

Learning Support Centre

A member recently accessed support from The Learning Support Centre. They are an organisation of dyslexia and disability specialists who can provide study skills and support to students such as scribing and note taking, mobility and personal support.

Find out more from their website:
www.learningsupportcentre.com

Budget 2016
Childcare and parental leave

Chancellor George Osbourne's budget in March 2016 talked about putting the next generation first: "providing security and opportunity from childhood to working age and through to retirement" but how does this affect you if you have younger children?

Childcare

From early 2017, the government is introducing Tax-Free Childcare to help working parents with the cost of childcare, ensuring more parents who want to can go out to work or increase the number of hours they work. The youngest children will enter the scheme first, with all eligible parents brought in by the end of 2017. Existing Employer-Supported Childcare will remain open to new entrants until April 2018 to support the transition between the schemes.

As well as this the government plans to double the free childcare entitlement from 15 to 30 hours for families with three and four year olds from September 2017.

Shared parental leave

There will be a consultation on how to extend Shared Parental Leave and Pay to working grandparents. The consultation was planned for May 2016 but there has been no conclusion reported yet.

Air passenger duty for children

The abolishment of air passenger duty for children under 12 is already in place but since 1 March 2016 it has been extended to children under 16. If you've already booked a ticket you will be entitled to a refund. Although most airlines will do this automatically, for some you will have to make a claim. If you're unsure check with the airline who you have booked your flight with to make sure you are getting a refund.

References

www.gov.uk/government/publications/budget-2016-documents

www.gov.uk/government/uploads/system/uploads/attachment_data/file/385074/Air_Passenger_Duty_-_child_exemption.pdf

Welfare Change in Northern Ireland:
What do you need to know?

Benefits are changing and that can be a bit of a scary prospect. At the MPS Society we are experienced in supporting families to communicate their needs effectively to ensure that they get the benefits they are entitled to. If you are being affected by Welfare Change please contact the MPS Society sooner rather than later so that we can guide you through the process and hopefully lessen the stress associated with such a big change.

So, what do you need to know...

Benefit Cap

The Benefit Cap is a limit on the amount of benefit a household can receive. For the Benefit Cap to apply, someone in the household must be in receipt of Housing Benefit. This will be introduced in Northern Ireland from 31 May 2016.

Some key points :

- Benefit cap limits will be applied
  - £500 a week if your household is made up of a couple (with or without children) or you are a lone parent and you have children living with you, you are responsible for
  - £350 a week if you are a single person and have no children living with you
- Some of the benefits that are received by you, or a member of your household, are added together to determine if you are above the limit.

These benefits count towards your household income:	**The Benefit Cap will not apply if you, or a member of your household, are in receipt of one of:**
• Bereavement Allowance • Child Benefit • Child Tax Credit • Employment and Support Allowance (except where the support component has been awarded) • Guardian's Allowance • Housing Benefit • Incapacity Benefit • Income Support • Jobseeker's Allowance • Maternity Allowance • Severe Disablement Allowance	• Working Tax Credit • Attendance Allowance • Carer's Allowance • Disability Living Allowance / Personal Independence Payment • Employment and Support Allowance (where the Support Component has been awarded)

Some other benefits may also impact your eligibility – a list can be found in the Welfare Change section of www.nidirect.gov.uk

- Initially Benefit Cap will be delivered through a reduction in your Housing Benefit.
  - You will receive a Housing Benefit award notification from the Northern Ireland Housing Executive.

  Please read this documentation carefully and report any discrepancies.

> "The Benefit Cap is a limit on the amount of benefit a household can receive.

Please do not hesitate to contact the MPS Society Head Office on 0345 389 9901 if you receive a letter and aren't quite sure what you need to do.

Personal Independence Payment

Personal Independence Payment (PIP) will replace Disability Living Allowance (DLA) for eligible people aged 16 to 64 years from 20 June 2016. The change from DLA to PIP will happen in stages:

- From 20th June it will no longer be possible to apply for DLA – all new claimants will be required to apply for PIP
- Existing DLA claimants aged 16 to 64 years, whose benefit is due to come to an end or who report a change in their care or mobility needs, will be contacted about reassessment from 20 June 2016
- Claimants with an indefinite/lifetime award for DLA will not be contacted until December 2016 at the earliest
- All those aged 16 to 64 years will be invited to claim PIP by December 2018.
- Young people in receipt of DLA will not be invited to claim PIP until they are 16, and their parents or guardians will be contacted before they turn 16.

As with DLA, PIP will be made up of two parts, a Daily Living component and a Mobility component. Each component will have two rates, standard and enhanced.

There will be four steps to make a claim to PIP

1 by phone

- You'll be asked for basic information including names of key doctors involved in your care.
- Someone else can call on your behalf, but you'll need to be with them when they call.

2 written submissions

- You will be sent a 'How your disability affects you' form.
- It is important that you explain how your condition affects you as clearly as possible.
- You can also send additional evidence such as support letters and medical reports.

3 assessment

- You may be requested to attend a medical assessment with an Independent Healthcare Professional.
- The MPS Society can help to prepare you for questions you might be asked.

4 decision

- A case manager will make a decision about your claim.
- If you are determined to be eligible for PIP your award will be regularly reviewed to ensure you get the support you need

During the reassessment process you will continue to receive your DLA award until your eligibility for PIP is determined. You must fully engage with this process. Failure to respond to letters from the PIP office could result in your DLA payment stopping before you are assessed for PIP.

Other benefits

An update on changes to other benefits will be included in the next edition of the magazine.

We would encourage you to contact the MPS Society as soon as you receive paperwork relating to changes in your benefits.

The MPS Society Advocacy Support Team are experienced in supporting families to explain the conditions that affect them.

Meet Laura – Students 4 Rare Diseases (S4RD) new employee

'If You Hear Hooves, it May be a Zebra...A Medical Student Empowerment Project' is a joint project between the UK LSD Patient Collaborative, of which the MPS Society is a member, and a Committee of Medical Students originally from Barts London School of Medicine and Dentistry, now qualified doctors working as F2's in hospitals around the country.

Following the success of this project the UK LSD Collaborative secured funding from Biomarin, Genzyme, Shire and Ultragenx to fund a post to continue to develop rare disease societies throughout medical schools in the UK. I am delighted to report that Laura Curran was appointed for 15 hours a week to be the projects new Educational Co-ordinator. Laura is based at MPS House in Amersham and is managed by the UK LSD Patient Collaborative.

Laura writes; "Hi I'm Laura and I joined Students 4 Rare Diseases (S4RD) in March as Educational Co-ordinator, working on a part-time basis.

My main responsibility is to raise awareness of the organisation and promote the work of the S4RD amongst both our current medical school societies and within non-member medical schools. I am tackling this by utilising social media to try and enter their sphere and by contacting the schools directly to help us advertise our events and who we are. At our recent symposium in London we had 18 medical students (55 delegates in total) in attendance, and I am hoping that this will be the start of relationships within many of the medical schools around the country! I am hoping to use social media as a place we can share important educational information provided by the LSD Collaborative and other reliable medical sources on the individual rare diseases, to our medical student following.

One of my aims is to update our current website and make it more multifunctional where medical school societies can log in and obtain lecture notes from events they could not attend and share them amongst their fellow students. And, also upload lecture notes from their own events so that they can be shared equally. It would also be a great place to store information on event speakers and other useful information.

As well as being here to support the S4RD committee I am also a single point of contact for the students in the medical societies and potential medical schools. It is important that I am here to support them in any way I can, and also as medical students move up through the ranks, I want to nurture and develop relationships with the society leaders and the rest of the society so we always have other students in line to take over.

I have really enjoyed my first few months and am really excited about assisting the committee in progressing the fantastic work that has already been done. This is a completely different role to what I have done previously, and a new post for the committee, hopefully I will be able to use my previous experience in making this role work for the committee!"

Laura's contact details

Students 4 Rare Diseases (S4RD), MPS House,
Repton Place,
White Lion Road,
Amersham,
Buckinghamshire,
HP7 9LP.

T: 01494 764 788,
W: students4rarediseases.org

Working hours:
Tuesday 12.30–14.00
Wednesday 11.00–17.00
Thursday 12.30–17.00.

Brain tissue donation

We recently received a call from a family who were considering donating a family member's brain to medical research upon their death. The family wanted to know if the MPS Society had any information on this matter. I had insight into what the family were considering, as my sister and I had first-hand experience of making that decision on behalf of my Dad, who had a neurodegenerative condition.

> "On the day he died, all we had to do was to make a telephone call to the Brain Bank to let them know.

Whilst Dad was still alive we got in contact with the Brain Bank at Kings College London, gave permission for them to access his medical records and signed the necessary consent forms. On the day he died, all we had to do was to make a telephone call to the Brain Bank to let them know.

From this point on they dealt with all the arrangements and liaised with his doctors and the funeral directors. The funeral plans went ahead as normal and were not affected by the donation process.

Our decision to donate brain tissue brought us comfort, knowing that it may help other patients and their families in the future.

Points to consider when making the decision to donate

- It is best to begin to consider donation in advance of the person dying. Family members need time to discuss the issue and to explore their feelings before making a decision.
- The decision to donate usually is made before the death of the person, so that sufficient information about the person's medical history can be collected and necessary arrangements made to ensure that donation occurs within the required time frame.
- Brain tissue donation needs to occur as soon as possible after death, usually within 72 hours. For this reason, donation is unlikely to delay funeral arrangements.

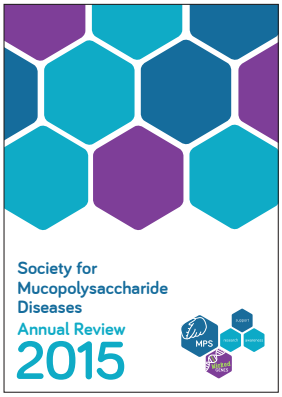
Further reading

Medical Research Council - Donating Brain and Spinal Cord Tissue
www.mrc.ac.uk/research/facilities/brain-banks/donating-brain-and-spinal-cord-tissue/

Human Tissue Authority - Body and Brain Donation Information Pack
www.hta.gov.uk/guidance-public/brain-donation

Jacqueline Adam
Managed Access Programme
Support Officer

A look back at 2015

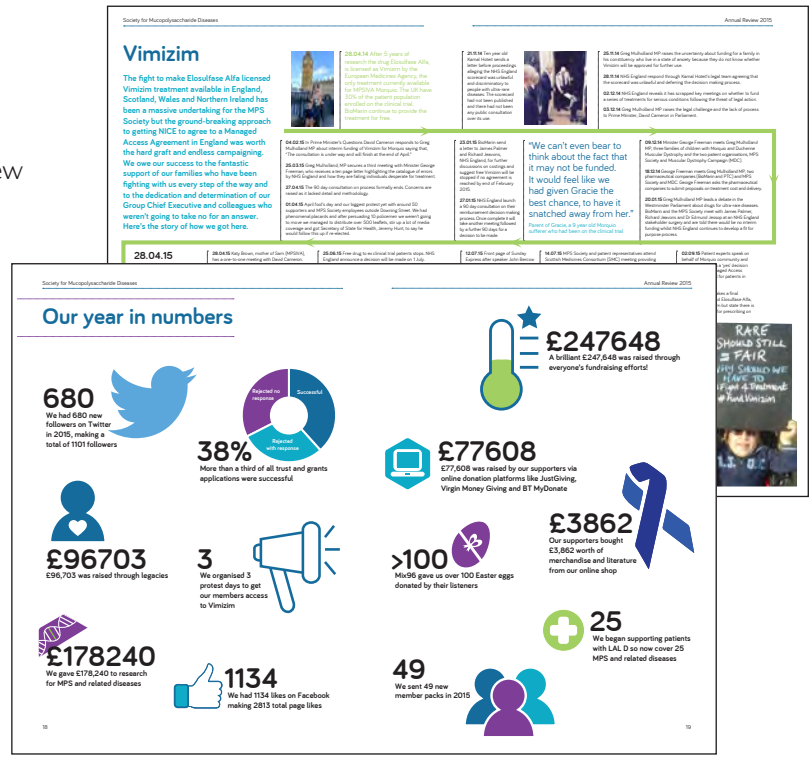


The MPS Society's Annual Review of 2015 is now available to download here:
www.mpssociety.org.uk/en/about/accountsreports/

It sets out our objectives and activities for the year as well as how we fulfil our public benefit as a charity.

The review includes highlights of our year including a timeline of the Vimizim campaign, a look back at the achievements of our Advocacy Support Team and how we have funded research throughout the year.

There's even a handy 'Our year in numbers' spread showing some of our best achievements from 2015.



First ever Velo Boot Tour fundraises for MPS

Fundraising

Thank you so much for all the fundraising you have done over the last few months. As you'll see our Wear it Blue section is crammed with pictures and snippets of what you got up to. As well as wearing blue and fundraising you made a big difference raising awareness by changing your profile picture on Facebook and Twitter and flying a plane for MPS!

We also want to thank all our London Marathon runners who tested their fundraising skills as well as their fitness in April over the 26 miles.

And thanks to all our other sporty fundraisers who have been training and fundraising all in the name of MPS.

You are marvellous!

Helen Crawley
Fundraising & Information Officer

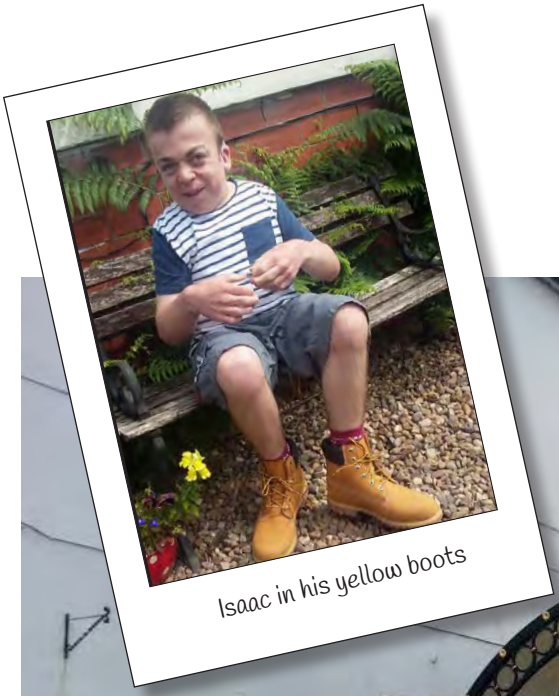
The Velo Boot Tour

On Thursday 12th May Timberland UK and their franchise partners for UK and Ireland embarked on the first ever charity fundraising cycling event, "the Velo boot tour" (named after their iconic yellow boot – the brand's signature piece).

The tour itself tackled the challenging "Ring of Beara" starting and finishing in Kenmare County Kerry and covering 144 kilometres of rugged terrain across the Southern Irish Coast.

The event was well supported, with 35 participants all raising money for different charities that they had their own personal affinities to.

For me, it was an easy decision to raise for the MPS Society. My nephew Isaac has suffered with this condition since birth, and I was delighted to have the opportunity to support a cause that I had a close affinity with and support Isaac and other children suffering with MPS.



Isaac in his yellow boots

Isaac's story

Isaac was born in November 1998 and soon after birth health problems materialised and he was eventually diagnosed with the condition Hurlers syndrome, (mucopolysaccharidosis) at 18 months of age.

The condition has proven to be a harsh and unforgiving disease and has directly and indirectly led to a number of serious ongoing health problems that Isaac has had to learn to live with.

Amongst others, partial hearing, impaired mental development, reduced skeletal growth, liver, spleen and heart problems are all ailments Isaac continues to cope with.

To date he has undergone 29 major operations, including 2 bone marrow transplants and having a shunt fitted in his brain. He has consistently defied expectations and recovered from setbacks with his health and maintained positivity even when the outlook has seemed bleak. It is abundantly clear that Isaac will not be beaten by these conditions and he has proven himself to be an extremely resilient person with an infectious personality and maintains a great sense of humour and a positive outlook despite the challenges he faces every day.

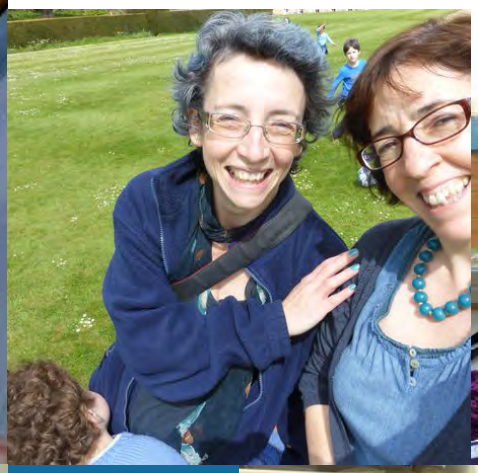
Despite the limitations Isaac has endured since childhood, Isaac has always and continues to live his life to the full and touches everyone he meets. Isaac has lived with these life threatening setbacks every day with a big smile on his face and he shows a relentless positivity that makes him a remarkable young man whom I am proud to call my nephew. Isaac's story is genuinely exceptional.

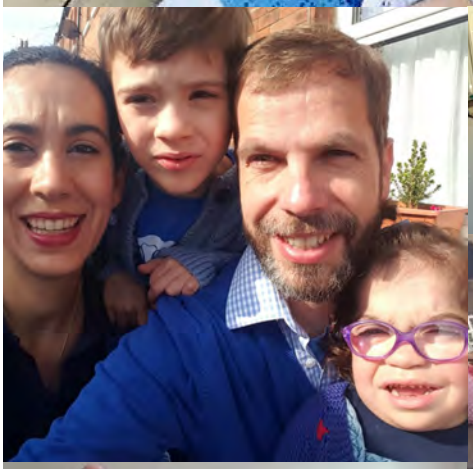
To dedicate my 140km bike ride to Isaac was an easy choice to make and I have been overwhelmed by the level of support people have shown to me and this most worthy of causes.

When tackling the steepest and most severe of the climbs on the "Ring of Beara" I frequently reminded myself that this is only a fraction of the mountain Isaac climbs and continues to climb every day of his life.

Thank you for

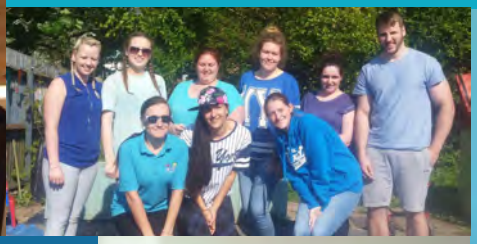
A huge thank you to everyone who got involved for MPS Awareness
sale or raffle, you all did a great job of raising awareness and vital fund





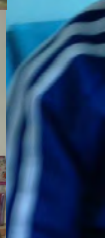
Vivienne Beckett got in touch to tell us that Genzyme held a Bake it Blue competition!

Barbara Newson involved the Hartlepool branch Boots in MPS Awareness Day and raised £91.46



Louise and Merlin Penny, and friends, organised a cake sale at Woolaston Primary School. They raised £250.89 through everyone at the school wearing blue and selling cakes. While Merlin's grandma Barbara raised £570 through various events and donations from friends and family.

Michelena Burke, who has Fabry, and her twin sisters Chrissie and Sammie approached their head teacher and asked the school to show support and Wear it Blue at Barnfields Primary School in Stafford. They raised £429. Sammie and Chrissie were also celebrated in the school assembly for arranging the day and showing how kind and supportive they are of their little sister. Their mum, Elaine, is very proud!

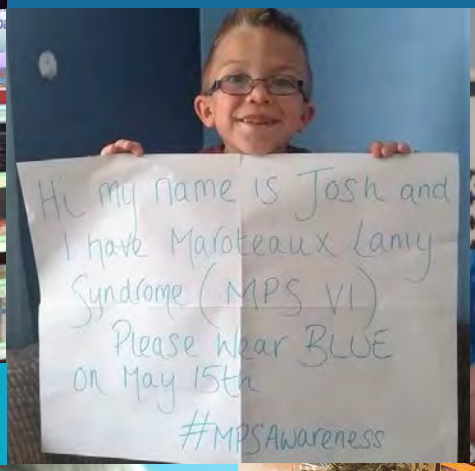


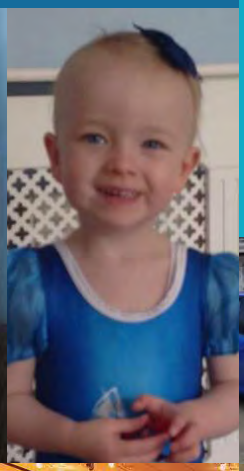
Charlie
was a

wearing it blue

Day! Whether you Wore It Blue, Took to the Skies, held a party, bake ds. Here are some of your photos and snippets of what you got up to.

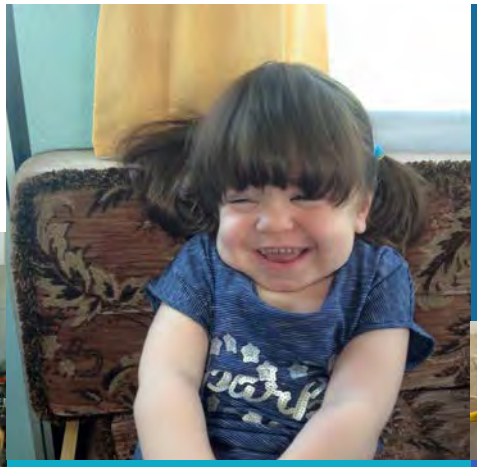
Wear it blue day at Boudoir Wine Bar in Glasgow raised £303.





of

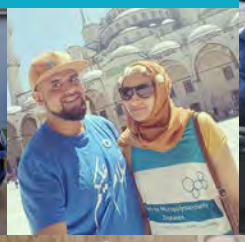
and Paul Dodd have organised many events this year, among them Wear it Blue day at Charlie's office which raised £250.



Ian Evans once again organised all the Arriva bus drivers of Aylesbury to wear blue on their buses while he dressed as Paddington Bear!

Aisha Seedat supported the campaign and told us:

"Well it was a sea of blue in Leicester for me this year. Born and bred in Leicester we certainly have made our mark and what better way to celebrate in supporting my charity with it. I was amazed at how many people supported the MPS Society not only in England in wearing blue but around the globe too. Malawi, India, Zimbabwe, South Africa, Zambia and Turkey. My phone bleeped all day with endless messages and photos."



Rachel Healy and her family held a pudding party and raised £200 by all bringing a pudding and donating money to enjoy all the cake! Rachel also involved her family and friends on MPS Awareness Day who all wore it blue in support. This is Rachel's 4th fundraising event for the MPS Society. Fantastic!

Surrey Square School

Surrey Square School organised a brilliant day on the Friday before MPS Awareness Day. Harvey Brown who has Morquio and attends the school said he wanted cake and balloons, and that's what he got! Millwall Football Club donated enough balloons for every child to release one each and a local chef donated a slice of chocolate cake for every child. The school also organised a raffle and a guess the number of sweets in the jar competition, had a visit from the Millwall FC mascot and wore it blue.

"The day on Friday was amazing" Kathryn Puch, the school's SENCo told us. "We have had a few replies to say that balloons were found in East Sussex. It was so lovely that those people took the time to let us know and write back."

The day raised over £650 through the various fundraising which was amazing!

Coffee, cakes and haircuts

On Friday 6th May Corey's school (Preston Primary) had a 'blue' mufti day to raise money in aid of the MPS Society. Everyone paid £1 and the total raised was £261

On Saturday 6th May 2016 I hosted "Coffee, Cakes and Haircuts"at Preston Baptist Church. This church is where Corey went to preschool and now his younger brother Joshua attends.

Over 200 people turned up on the day, which turned out beautifully sunny, to enjoy a cuppa and the homemade cakes that friends and Karen from Karen's Cakes supplied. The Hawk and Owl Sanctuary came along to talk about their birds of prey and let people hold them. Chase from Paw Patrol greeted visitors as did our very own football club mascot Gilbert the Gull.

The tombola was a great hit with over 100 prizes donated from local businesses and family and friends. A family friend, Marcus, kindly donated a ride on his Harley Davidson which was on show and children and adults could sit on rev the engine! A bring and buy sale, where 2 of Corey's friends donated their bikes. There was also face painting by Lorraine and Mandy, tattoos, guess the name of the teddy, a lollipop game, biscuit decorating and handmade blue lavender bags from Alison and heart purses from Corey's Nanna.

The main event of the morning was 7 of us having our hair cut off. Myself, Aimee, Vikki, Jo, Dawn, Chloe and Corey's classmate Amelia (8 years old) collectively had 82 inches of hair cut off which was sent to The Little Princess Trust to make wigs for children going through cancer treatment. We were sponsored to do this with the funds going to The MPS Society.

The total amount raised for this event was £2510.46

The atmosphere was amazing and there really was a true community spirit as so many people turned up to support the event who all had fun and hopefully learnt something about MPS. I am completely overwhelmed by the support, generosity and kindness of everyone especially all our friends who helped out preparing for the event and gave up their time on the day to help man stalls and even have their hair chopped off! All of this came about because of our son, Corey, who happened to be born with this rare genetic condition which no one had heard of and our determination since, to raise awareness.

Teresa Jeffery
Corey's mum

London Marathon 2016


Jade Arthur and Gemma Cantor


Alan McKenna

Well done and thank you so much to all our fantastic London Marathon runners. You did an amazing job in collectively raising more than £12,200 with more still coming in! We hope you had an amazing day and it's great to hear from so many of you wanting to sign up already for 2017.

Alastair Shields ran the London Marathon and raised a spectacular £2124.63. Alastair also managed to obtain a matched donation from the John Laing Charitable Trust for £1500, giving him a grand total of £3624.63!

Ed Smith ran the London Marathon and raised an enormous £3824.97! Ed will also be entering the Great North Run later on this year in support of the MPS Society.

Alan McKenna had a great time running the London Marathon for the MPS Society and raised a fantastic £1558.39.

Sisters **Jade Arthur** and **Gemma Cantor** ran the London Marathon together in support of the MPS Society. Jade raised £1637.80 and Gemma raised £1353.08, giving the sisters a combined total of £2990.88!

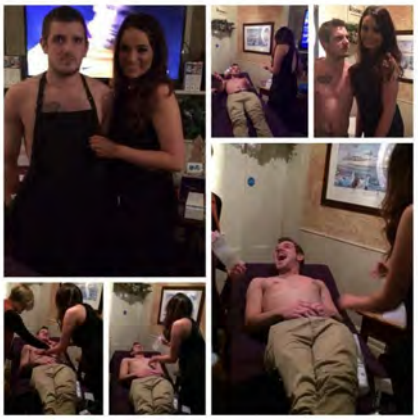
Scott Bennett secured a place for the marathon via the ballot but still chose to raise £298.12 for the MPS Society.

Marina and Friends Fundraisers is a charity shop in Bristol opened by Marina after her grand daughters both died from MPS. Marina started raising money from car boot sales and made £22,000 before opening the shop.

Now over 80 years old Marina still works in the shop which raised £7497.47 last year bringing the total raised to an absolutely amazing £149,325.95 since they opened.

We want to say an enormous thank you to Marina and her team for all their hard work.

You can find Marina and Friends on Facebook: www.facebook.com/MarinaAndFriendsFundraisers or if you're near Bristol why not pop in!



Wax for Jack

Fundraising is still continuing in memory of Jack for the MPS Society. Jack's friend, Sophie Bennett recently launched her new business venture "Sophie's Beauty Spot". She marked the occasion with a sponsored chest wax!! Jack's friend Rick Woolford kindly braved the pain and a fantastic £150 was raised for MPS. In addition to this, Sophie donated money raised from a raffle to a fund for a memorial bench for Jack.

Thanks so much to Sophie and Rick and all who donated or sponsored!

Amanda Stuart

Sporty fundraisers

Amy Wright

I completed my 2nd half marathon of the year yesterday in Stratford-Upon Avon which was good fun, and me and Robyn are running the Vitality Half Marathon in Hackney, London in 2 weeks time as our number 3. We will also be running Bournemouth and Winchester in the Autumn, and others when we have decided on dates! I believe we are up to just over £700 on our just giving page and still going strong.

Anna Rigden

Last year I started working for a play scheme that looks after children with all kinds of complex needs and there I met Ella and Daniel, two gorgeous children who both have MPSIII (Sanfillipo). As I worked with them more and more I got to know them, how smiley they were, very gentle and also very active (non-stop). I absolute love working with them and always look forward to when they are at the scheme as they are an honest joy to look after. One day when speaking to some family of one of the children I was told that what they both have is degenerative and terminal. I felt sick to my stomach to know that down the line, these amazing children won't be with us anymore and currently there is no cure for MPS diseases. So, I went home and thought about what I could do and having just taken up running, training for a half marathon seemed like the logical thing to do. I set myself 12 weeks doing three runs per week, to reach the 21km goal. Training was tough. I love running, but it can be tedious and also mentally

so challenging. Sunday March 13th came around quickly and I completed the run in under 2.5 hours, alongside my Dad who encouraged me the whole way. It was a hot day and I struggled round the course, but the thought that people had sponsored me and were supporting me kept me pushing on. I raised £500 for MPS and hope that the money can go towards providing care and support for all those affected and also to one day finding a cure. Daniel and Ella have provided so much happiness in my life and I hope to give something back to them and their families, who are truly inspirational people.

Sarah Jane Upton

Freddie De Gennaro has MLIII, complex epilepsy and autistic spectrum disorder. On Easter Saturday 2016 my very good friend (and Freddie's godmother) Sarah Jane Upton ran the Cardiff half marathon along with her partner, Stuart, and son, Tomer. Sarah Jane had spent many weeks and months preceding raising sponsorship, organising supporting events and raising awareness.

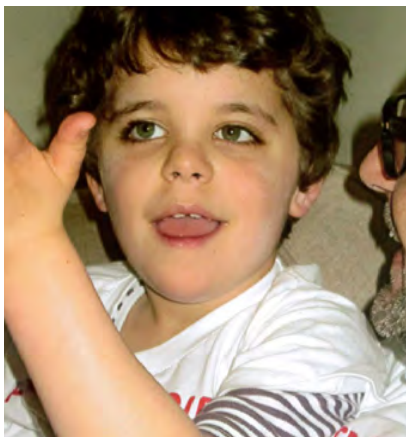
Alongside this my ex employer Alison Brown raised over £1000 at Fishers Restaurant in Bristol, Crockerne School (Freddie's old school) held a dress up day in Freddie's favourite things (Peppa Pig and Wotsits) where the children donated £1 each, and close friend Fabienne Vailes organized an 80's disco in the local village hall to raise money and awareness.

The amount of money raised communally was enough to fund a travel safespace and 2 epilepsy monitors for Freddie as well as a donation of £830 to the MPS Society

– such an amazing charity who offer invaluable support to us and to others.

Huge heartfelt thanks to Sarah Jane and to all who overwhelmingly contributed their time and money so generously – just amazing!

Justine De Gennaro
Freddie's mum



Thank you to all our donors including...

Rick Woolford and **Sophie Bennett** held a sponsored chest wax and raised £150 to mark what would have been Jack Stuart's 21st birthday. His mother, Amanda, shared the news with us.

Sarah Wolfe is celebrating her 30th birthday and remembering her cousins, Stephen, Ian and Mark Harvey, by running a number of events throughout 2016. So far she has raised £253.75.

Anna Dryer and colleagues at PwC donated £615, half of the funds raised at a charity quiz they organised.

Robert Baines and his daughter, **Cheree**, ran the Vitality Brighton Half Marathon to support William and raised a fab £85 for the MPS Society.

Angie Naish organised a charity night with the Wurzels and has so far raised £71.25 through JustGiving. Sounds like a fun evening!

The Amersham Playgoers Theatre Group chose us as this year's local charity and have given us their annual donation of £100.

Sally Mitcham sent us a cheque for £100 raised by the church where her son Danny's playgroup is based.

Donna Bown sent us £10 which was kindly donated by her son Luke's Great Nan.

We were nominated to receive £347 from the sale of scrap at **Emerson Industrial Automation by Kayleigh Humphrey** who works there.

Sarah Upton also ran the Cardiff half marathon and has raised £337.50 so far.

St James the Less church wives group in Hadleigh, Essex have donated £200 from their fundraising efforts throughout the year in support of one of their members who has a grandson with MPS.

Simon Greening ran the Cardiff half marathon and a Tough Mudder (hopefully not on the same day!) and has so far raised £285.

The final total raised from the **BADCo pantomime**, The Princess and The Peastalk, featured in the last magazine was £1250. Oh no it isn't...oh yes it is...

The film makers of Atomic: living in dread and promise have donated the £450 proceeds of a screening which was held to launch the CD of the original score by the band, Mogwai. They decided the proceeds would be donated to the MPS Society as the film featured archive footage of Elisabeth Peach who died from Hunters, and her family.

Debbie and Mark Burniston and their team completed the Great North Run and the Great Manchester run raising £1025 in the process.

The Torbay Methodist Luncheon Club collectively donated £206, which included Teresa Jeffery's speaker fee.

LV= Ipswich held a company raffle and raised £1009 for the MPS Society as one of their employees has a son with MPSIII.

The **University of Sussex** student Union Dance Society have raised £154.

Mick Yates completed the Bespack Gear 10K and raised £58.50.

Sarah Cutler worked hard to raise funds and awareness in her local area, which included cake sales at Sandford St Martin's Primary School and at the Co-Op petrol station in Sandford. In the last few months Sarah has raised £421.20, adding to the total of £2053.45 on her JustGiving page!

Nathaniel Ives and his team completed the Rat Race Dirty Weekend, a very muddy obstacle course, and raised £250.

Katy and Simon Brown, along with friends Ian, Lindsay and Dan ran the Leeds Half Marathon and the Great Manchester Run and raised £1367.50.

Louise Hiller did a sponsored skydive and raised £1948.75 on her JustGiving page.

Josh Ramshaw and his team took on a sponsored walk in memory of Jack Stuart, who sadly passed away from MPSII.

The Meadows Primary School in Madeley Heath wore it Blue and raised £150, MPS is a Charity close to their hearts as a very special person called Emily who sufferers from MPS attends the school community.

Ysgol Maes y Coed school in Bryncoch held a 'Blue Day and raised £105.74

Kathryn Wallis, grandparent to Archie and Isaac Eaton who have Morquio held a Garden Party and raised an amazing £1,690.

Chris Merrick from Leicester walked 100K from London to Brighton and raised £110.

Donations

Brenda Weston; Sovereign Health Care Charitable Trust; The Clipper Foundation; The Florence Turner Trust; The City Bridge Trust; S Michael Brecker; Hillpark Secondary School; Riversdale Surgery; Westcliff High School for Girls; Benham Charitable Settlement; Mrs A Baker; Oakdale Trust; The Highcrest Academy; Gill Gardner; Darryl Brook; Mrs L A Sutton; Barbara Newson; Mr & Mrs Shaikh; The Joseph Strong Frazer Trust; Baron Davenport's Charity; Amanda Scott; Mrs P Booty; Kevin & Bernie Drayne; Mr & Mrs Lavelle; Linda Rowland; Mr & Mrs Cooper; Kerry Stratton; Alison Powell, Jean Armsby; Sally Sutcliffe; Graham Nance; Kerry Marshall; The Clover Trust; Rachael Sumner; Nicholas Winder; Abby Thomas; Amanda Laycock; Elizabeth Merryweather; Francesca Arrigoni; George Kingston-Rayes; J Garthwaite; Jodie Harrington; Karen Hewitt; Matt Mould; Michael Morris; Nick Miles; Paul Berg; Peter Rennoldson; Tim Peach; Tmara Senior

In Memory

Lady Shauna Gosling; Jack Stuart; Daniel Singh; Gareth Evans; Alan Poole; Barbara & James Hill; Emily Otway

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

David Fowler; Marilyn Eggleton; John Casey; Mrs D Bown; Your Derwent & Solway Housing Association; Ellen Graham; Mr & Mrs Eggleton;

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

P J Martin, J Casey, D Palmer, J Hastings, J Winzar, D Winzar, EM Lee, K & J Hudson, R & K Dunn, Saville Norman, M Newell, M Tosland, S Bhachu, C Cullen, L Brown, V Lucas, D Forbes, S Winzar, E White, C L Hume, C Sullivan, A Byrne, D M Robinson, N Cadman, J Wilson, A Tresidder, M Rigby, Stuart Robinson, K Osborne, E Cox, Mr Nicholas Thompson, M Peach, R Arnold, J Ellis, I & V Pearson, C Gibbs, D Peach, A Cock, A Dickerson, M Kalsi, D Holmes, P Summerton, R Gregory, L Stillwell, R Henshell, K & S Bown, S & J Home, V Little, S & D Greening, Z Gul, M Reeves ,J Dalligan, M Malcolm, E Mee, M Hahner, K Brown, E Moody, E Brock, M Fullalove, M Leask, G Ferrier, E Parkinson, R Taylor, John Scott; A Weston; Evelyn White; C Hume; W Cavanagh; A Sabin; B Harriss; L Brodie; A Ephraim; C Dalligan; M Malcolm; E Mee; E Moody; Mr & Mrs Hahner; K Brown; M Fullalove; Margaret Leask; Gordon Ferier; E Brock; E Parkinson; R Taylor; R & N Gregory; Lindsay Stillwell; K & S Bown; V Little; Michael Reeves; Z Gul; S & D Greening; J Casey; D Palmer; J Hastings

Sandra and Rash Singh held their 4th Fete in memory of their son Daniel on Sunday 15 May and raised an amazing £1500. The event was attended by Sue Peach, Chair of the MPS Society and Jane Roberts from the Willink who lent a hand. The Sun was shining down, there were lots of stalls, face painting, a fair, live music and a tombola.

Barnfields Primary School in Wildwood raised £429.10, the children brought in a donation so they could wear blue for the day.

On Easter Saturday **Alex Walker** and **Matthew Reading** were married at St Mary's Church, Haddenham. The Organist and Singers at the wedding waived their fees and Alex and Matthew have decided to donate the money to MPS. Alex and Matthew knew Matthew Hardy who died in 1994, his parents and Jenny and Andy Hardy are great supports of the Society.

Copmanthorpe Primary School in Low Green held a non-uniform day and raised £157.68.

Tracy Cunningham raised £1,074.88 via a bag pack at Marks and Spencer's for MPS Awareness Day.

The Hampden Arms held a quiz for MPS Awareness day and raised £110.

Velma Lazenby sent in £40 collected from her friends in Wetherby U.3.A. Theatre Group on MPS Awareness Day in memory of her granddaughter Emily Otway.

Thank you to everyone who took part in the Weather Lottery:

Mrs O Megoran, Mrs J Edwards, Mrs C Lavery, Mrs G Plummer, Mr M McCawille, Mr A Selwood, Mrs T Brown, Mr A Dickerson, Mrs M Crespin, Miss D Halleron, Ms C Halleron, Mrs D Bown, Mr M Hughes, 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"

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

MPS Commercial

MPS Commercial

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Board of Directors

Chair
Bob Stevens

Board
Jessica Kafizas
Sophie Thomas
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.

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For more information about MPS Commercial and clinical trials visit our website:

www.mpssociety.org.uk/commercial

Patient Access to Clinical trials and Treatment

In 2013 the MPS Society set up MPS Commercial; a wholly owned subsidiary of the MPS Society whose social objectives are to reinvest any profit into the MPS Society so that the MPS Society can offer additional support and activities to its members.

Last year MPS Commercial's donation to the MPS Society included €20,000 to take members and their families to Bonn for the International Conference in July 2016 and €20,000, which was matched funding from the Gosling Foundation, to take families to Lapland in December 2016!

The MPS Commercial team are really excited by the opportunities that their hard work has provided.

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 Lead 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 supports the newly introduced Managed Access Programme for Vimizim

Jackie is a Vimizim Managed Access Programme (MAP) Support Officer, for those with MPSIVA, Morquio.

Pauline joined MPS Commercial in February 2016 as a part-time accounts assistant.

So how did we do this?

Supporting patients and their families whilst participating in a clinical trial

MPS Commercial are fully aware of the demands of being on a clinical trial and what it entails. We aim to make the process as stress free as possible. Over the last ten years (initially as MPS Society providing the logistics and latterly as MPS Commercial) our expertise and support role has grown from the early days of UK MPS patients attending UK hospitals to now supporting patients with MPS and other ultra-rare conditions across mainland Europe, Asia, Australasia and both North and South America.

We work with patients and their families taking the pressure off families and clinical trial sites by supporting and managing the logistics involved with participating in a clinical study. We book accessible accommodation, trains, planes and taxis and ensure that the travel arrangements include the necessary assistance. During the clinical trial start-up phase MPS Commercial risk assess accommodation in close proximity to the hotel to ensure it is appropriate for families' and patients' needs.

MPS Commercial works with the clinical trial site and reimburses any out of pocket expenses once claims have been validated against the Clinical Trial Sponsor's Travel and Reimbursement Policy.

Managed Access Agreement

MPS Commercial has responsibility for administering the Quality of Life assessments (QoL), one of the five criteria for continuing treatment for those with MPSIVA under the Managed Access Agreement (MAA) agreed by NICE. The first Managed Access Agreement Programme was established in December 2015 for Vimizim (elosulfase alfa) and was developed from the MPS Society's suggestion of a robust procedure whereby all patients that met a set of criteria would be able to access reimbursed treatment.

In the case of Vimizim the QoL assessments are four questionnaires administered over the telephone or face to face at defined points in the treatment schedule. This is the first ever MAA and MPS Commercial have worked in close association with the LSD clinical experts, NHS England, NICE and pharmaceutical company to provide accurate and timely QoL data.

As a first we have had to develop processes of communication, scheduling and reporting. We have been proactive in our continuous assessment of the managed access process looking not only at our own practices, but in reviewing how the QoL assessments used meet the aims of the MAA, seeking the patients and hospitals perspective and keeping the pharmaceutical company informed of progress. We have organised a 6-month review with all the stakeholders to ensure that key information and issues are shared and actioned as necessary.

Survey design, collection and analyses

Our services are expanding rapidly and we can complete outcome research from survey design and data collection through to analysis of the results. We are thrilled that the team have had four abstracts accepted at the International Symposium on MPS and Related Diseases in Bonn and are busy designing the posters!

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