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MPS
support
research
awareness
Wicked GENES

The MPS Magazine

Spring 2013

Society for Mucopolysaccharide Diseases

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

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

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.

The MPS Society:-

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

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

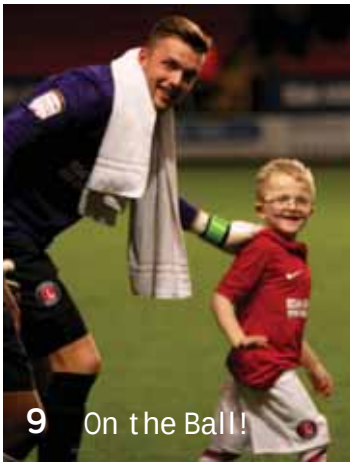
Brings about more **public awareness** of MPS and Related Diseases

MPS and Related Diseases

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

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

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



Front Cover
Anabelle, see also p10

The MPS Childhood Wood 12

Contents

The Chief Executive's Report

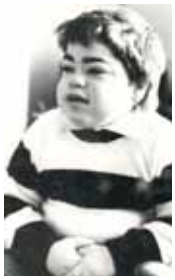
As we go to print I am sure I am not alone in trusting that the worst of the winter weather is behind us and we can focus on warmer weather to come.

Talking of warmer weather I have had the opportunity to escape the worst of the snow to hit the Chiltern Hills in both January and February. On Sunday 20th January after sitting for five hours on the runway (not literally) at Heathrow Tanya Collin-Histed, CEO of the Gaucher Association and I left for Mumbai, Bangalore and Chennai in India. This was truly a memorable and humbling week that included seeing the launch of Shire's new Humanitarian Aid Programme for India and working alongside Dr Vellodi from Great Ormond Street, Dr Suresh Vijay from Birmingham Children's Hospital and Dr Uma Ramaswami from Manchester Children's Hospital in the MPS Clinics. I think we were all moved by the stoicism and dignity of all the families we were privileged to meet. However, for me, the family with a daughter with Sanfilippo who'd travelled 1,700kms one way on a bus to see the 'Sanfilippo doctor from England' left me speechless, not least for forty minutes later they were heading for the bus station and the 1,700kms home!

Two weeks later and we were again battling snow on the M25 this time to get to Gatwick for the Orlando flight and the WORLD Lysosomal Storage Disease Meeting. This has grown from an acorn of an idea by the University of Minnesota to a huge oak tree of a meeting with nearly 1,000 delegates all focusing on Lysosomal Storage Diseases. Having worked out the presentations on MPS, Fabry and related diseases I wanted to hear, the rest of my programme was wall to wall meetings and networking. But in truth this is where anyone who is anyone in the LSD field, be it clinician, nurse specialist, researcher, scientist, academic, pharmaceutical company or parent organisation spends the second week in February, and in case you are thinking, there was no time for even a quick visit to Mickey Mouse!

In the last MPS Magazine, our Chairperson, Sue Peach, updated you on the progress of Wicked Genes. In this MPS Magazine you will see the Wicked Genes Campaign coming alight and we are all expectant that with your help we will be able to reach out to many people for support raising funds for pioneering research that may deliver new therapies for our children and adults with MPS, Fabry and related diseases as well as funding the indispensable MPS Advocacy Team and support activities provided by the MPS Society.

I want to give my own thanks to the entire MPS team who over the past year have worked tirelessly to support you all. Thirty years on from when my own son, Simon's hands, classic of many children with MPS, were depicted as the logo for not just the MPS Society but many other MPS Societies around the world I am pleased to commend the new futuristic branding of the MPS Society. Whilst we can't live on history the team have incorporated the hands and the blue colouring in the new 21st century MPS and Wicked Genes brand that I sincerely trust will serve the MPS Society well as it aspires to help more families in innovative ways and be at the forefront of greater understanding of these truly devastating conditions. We can only achieve these goals with your support .



Christine Lavery MBE
Chief Executive
c.lavery@mpssociety.org.uk

News from the Board of Trustees

The Society's Trustees meet regularly. Here is a summary of the main matters that were discussed and agreed at the Trustee Board Meeting held on Saturday 1 December 2012.

Treasurer's Report

The draft Income and Expenditure budgets which had been received by Trustees prior to the meeting were considered in detail. The budgets were agreed unanimously.

Risk Management

The Trustees considered the Health and Safety report. There were no issues raised and this was agreed unanimously.

Personnel

Subject to some minor changes, Trustees approved the new employee performance management document.

Clinical Management

Trustees were informed that Dr Maureen Cleary at Great Ormond Street Hospital is now recruiting children with MPS IIIB to a Shire sponsored natural history study.

Trustees were updated on the two new companies, BUPA and Central who came into the Home Care space as of 1 October 2012 providing ERT to MPS and Fabry patients outside of the hospital environment.

Trustees were appraised of the current situation with regards AGNSS and the new NHS Commissioning Board. Trustee Bob Stevens, spoke of the engaging and helpful meeting he and his wife, James and Claire Garthwaite and the Chief Executive had with Secretary of State for Health, Jeremy Hunt. *(There is a full update on the new NHS National Commissioning Board in this Magazine.)*

Advocacy Support

The Chief Executive advised Trustees of the ongoing difficulties in Wales for clinicians wanting to treat MPS and Fabry patients with Enzyme Replacement Therapy (ERT). One piece of good news was the MPS Advocacy Team's success in achieving ERT for one of our adult MPS I members who has been denied treatment for eight years.

Recognising the distress of many of our MPS IVA member families regarding the 17th November episode of Casualty, Trustees reviewed the MPS Society's response which included a formal complaint to the BBC and a letter to 'Points of View'. Trustees concluded that this was a thoroughly regrettable piece and deeply flawed storyline but agreed that a majority of MPS IVA families wanted the matter put to bed. It was agreed as an MPS Society there was no more the Society could usefully do.

The Chief Executive reported back on the meeting, held at the behest of the Trustees, with representatives of Peninsula University, Exeter, where concerns were raised around communications and lack of communications with our members who had in good faith participated in the Health Technology Assessment (HTA) study which was looking at cost effectiveness of ERT. At the meeting the Chief Executive secured copies of the Patient Summaries for MPS I, MPS II and Fabry disease and these were distributed with the Winter MPS Magazine.

Fundraising

The Chief Executive confirmed that the Charity Commission has been asked to approve Wicked Genes as a working name under the MPS Society's registration. *(Approval has since been granted by the Charity Commission.)*

Trustees discussed the importance of the MPS members and supporters understanding that Wicked Genes is the working name for the fundraising initiative of the MPS Society and that Wicked Genes is raising funds for research across all the MPS, Fabry and related diseases as well as all the Society's advocacy and support activities. Trustees acknowledged that fundraisers can restrict their money to a specific use and indeed the MPS III families are doing this to raise funds for the Genistein Clinical Trial.

The Chair of Trustees, Sue Peach oversaw the MPS draw.

Welcome to:

Debbie Cavell

Hi! My name is Debbie Cavell and I joined the Advocacy Team as an Advocacy Support Officer in January. The team have been very supportive and helpful and I have been made to feel very welcome by both the team and the families I have met so far. I am inspired by the commitment and dedication of the team and I am looking forward to a challenging and rewarding role.

I will be working with and supporting families with MPS I, MPS IV, MPS VI, MSD and ML II and visited Great Ormond Street Hospital BMT Clinic on my second day!

I am a newly qualified social worker having graduated from Bucks New University in June 2012. Previously I worked for local authority children's services and a number of local schools in support positions.

I have three older children and enjoy being a Mum. I also spend my time going to the cinema and pub quizzes and listening to live bands in my local pub.

I am very much looking forward to meeting and getting to know more families over the coming weeks.

Another new face to introduce to you. Elizabeth Hardy has been working in the Amersham Office during the six months of Sue Cotterell's maternity leave, which finished at the end of February. However, Elizabeth will continue to work alongside Sue after her return, part-time, helping to raise the necessary funds for the MPS Society's work from grant-making trusts and foundations.

Elizabeth Hardy

Hi, my name is Elizabeth Hardy – usually known as Liz! I joined The MPS Society in September last year to stand in for Sue Cotterell while she and Steve welcomed their new daughter into the family. Everyone has been very friendly and I've so enjoyed working for the Society that I've been asked to stay on, for two days a week, as part of the fundraising team. This means that I'll be able to come to the MPS Conference in Coventry in June and meet some of you and find out more about how the Society works with and for you. It will all help me, hopefully, to put in more persuasive appeals to the trusts and foundations!

I've worked for lots of different organisations over the past !!!!! years, including Buckinghamshire New University, Buckinghamshire County Council and the National Film & Television School in Beaconsfield – also a fundraising post. Working for the MPS Society has been and will continue to be very fulfilling, not to say an education about the gritty determination of human nature to overcome adversity in any form.

Outside work I enjoy singing with a choir and writing – helps a bit with the applications! With adult children no longer living at home I'm able to indulge my passions a bit more these days, though nothing beats meeting and working with the lovely and hardworking people at the MPS Society.

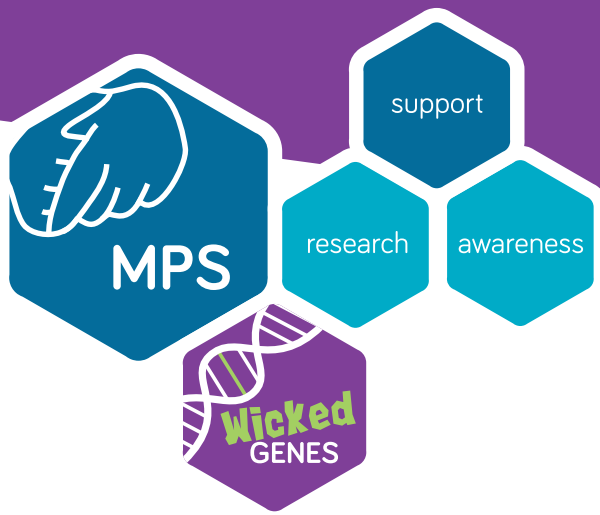
Book Now!

MPS Society Conference Weekend

28 - 30 June 2013, Hilton Coventry

The full programme and booking form is available to download from our website

www.mpssociety.org.uk



Brand launch

The MPS Society gets a new look!

In this competitive and uncertain economic environment we simply have to reach out to new and existing supporters. It was decided that after 30 years the MPS branding needed modernising, to remain engaging and to stand up to other charities in a competitive sector. An updated MPS logo would also have more synergy with our new fundraising logo of Wicked Genes.

While it was felt that some modernisation was due, we didn't want to lose the historical integrity of the MPS Society logo and it was therefore felt we should retain the blue colour of the MPS logo and the MPS 'hands' due to the high recognition in the field of MPS/rare / Lysosomal Storage Diseases.

The research we conducted found that you, our supporters and members wanted co-branded materials (MPS and Wicked Genes) wherever possible.

The new MPS and Wicked Genes logos work together but can also be used apart. The use of hexagons means that everything fits together and has a medical feel, but the use of rounded edges softens this. The additional use of smaller hexagons with the words; support, research and awareness allow us to highlight the charity's objectives.

The 'hands' are still used but have been adapted slightly so that they are of a similar style to the other elements of the logo.

In line with other UK charities, we will also be moving from our current web and email address that includes the use of **.co.uk** to **.org.uk** to reflect our not-for-profit status. Our website will be **www.mpssociety.org.uk** and our email will be **mps@mpssociety.org.uk** .
However, we will have a redirection in place from our old address so don't worry if you don't remember to use the new one.

The new MPS branding is clean, simple and modern but has maintained its historical integrity.

Let us know what you think at mps@mpssociety.org.uk

New members

Zubeda has recently been in contact with the Society. She has Fabry and the family live in West Yorkshire.

Lauren has recently been in contact with the Society. She has a diagnosis of Fabry disease. She lives in Aberdeen, Scotland.

Natalie has recently been in contact with the Society. She has a diagnosis of Fabry disease and lives in the South East.

Mr Matthew Sims and Miss Lauren Jowett have recently been in contact with the Society. Their daughter **Gracie** has been diagnosed with ML II. She is three months old (born 3rd December 2012). The family live in the South East.

Jason Liversidge has recently been in contact with the Society. He has a diagnosis of Fabry disease. Jason is 37 years old and lives in the North East.

Sally has recently been in contact with the Society. She has a diagnosis of Fabry disease. The family live in North Yorkshire.

Congratulations

Professor Bryan Winchester, a Trustee of the Society, has been awarded a "Clé du Lysosome" for his research on lysosomal storage diseases by our French sister organisation, Vaincre les Maladies Lysosomales. The award was presented to Bryan at the annual Award Ceremony in Paris in November 2012.

The Ceremony was held in a glittering State Room in the Hotel de la Marine, the headquarters of the French Navy on Place de la Concorde in the heart of Paris. Bryan was the first British recipient of the award, which was initiated in 2007. As he had to give his acceptance speech in French, Bryan was very glad of the Champagne reception that followed!

Family home videos needed!

The MPS Society are looking at creating some short videos to raise awareness. These will be used online on our website and on social networking sites such as Facebook and Twitter. We hope that people will share these far and wide, connecting with new supporters.

To make these films engaging and appealing to viewers we would like a selection of snippets of home videos from families affected by MPS diseases. We are looking for a variety of settings – playing with siblings, family meal times, celebrations and holidays. We would be interested to have footage of treatments (in hospital and at home) and anything else you may have filmed!

Footage will be selected, edited and put to music. We don't need Hollywood quality. We need real life home videos.
If you can help us please contact us on 0845 389 9901 or wickedgenes@mpssociety.org.uk today.

What's on in 2013

CONFERENCES and REGIONAL EVENTS

MPS Awareness Day
15 May

Launch of Wicked Walkabout
18 - 19 May

MPS National Weekend Conference
28 -30 June Hilton Coventry

Childhood Wood Planting
13 October

MPS REGIONAL CLINICS 2013

MPS IV Clinic GOSH
9 July

Manchester BMT Clinic (under 6 years)
12 April, 12 July, 11 October

ON THE BALL!

Dylan is a very big fan of Charlton Athletic Football Club. On November 27 he was lucky enough to be a team mascot in their game against Peterborough at the Valley.

Demelza South East London had been approached by the **Free Kicks Foundation** and asked to nominate a local child to be a mascot at a forthcoming game. They knew Dylan was a keen footballer and were kind enough to put him forward.

Dylan's day began at the Players Entrance at the Valley where we were met for a tour of the stadium and the trophy room. Then he went off to meet the players and the manager in the home team dressing room – and got his programme signed by every one of them.

When the players went out onto the pitch Dylan went with them. He was able to practice his skills with one of the coaches while the team warmed up.

When the team came back out onto the pitch for the match Dylan walked on alongside his hero, the goalkeeper, Ben Hamer. Dylan was very, very proud of his role in front of 17,377 fans. After he shook hands with the other team he had his photograph taken with the captains and the referees before he took his place in the stands to watch the game.

He had a splendid time, made even better when Charlton beat Peterborough by two goals to nil! Obviously they had a lucky mascot that day.

Dylan and his family have been members of the MPS Society since he was diagnosed with Alpha Mannosidosis four and a half years ago. He had a bone marrow transplant in May 2009. His MPS Advocate, Rebecca Brandon, has been a great help to his family, most recently by giving a talk about his disease to the teachers at his school.

Thanks to all at Charlton, Free Kicks and Demelza for a day he will never forget.

Manchester BMT Clinic (over 6 years)
3 May, 5 July, 18 October

BMT Clinic GOSH
26 March, 24 September

Fabry Clinic GOSH
9 April, 27 August

MPS III Clinic GOSH
14 May, 13 August, 12 November

Birmingham MPS and Fabry Clinic
29 April, 14 June, 8 November

Adult Fabry Clinic Birmingham
11 June, 13 August

Bristol MPS Clinic
27 March

Northern Ireland Clinic
24 May

Anabelle 18 February 2011 - 25 December 2012

I thought I would talk about the three important things which come to mind when I think about Anabelle; **her life; her laughter and her love**.

Her Life

Anabelle came into this world six weeks early and weighed a mere 3 and half pounds; she could practically fit into the palm of my hands. She showed us at this stage her strength and resilience and overcame her premature birth by putting on weight and learning how to feed.

In March the following year she was diagnosed with MPS I Hurler and although looking back they were dark days, having her there helped us deal with this particularly with the support of friends, family and also the MPS Society.

Her condition meant that initially we had to go up to Great Ormond Street weekly so our little one could have Enzyme Replacement Therapy. We then practically moved to London so that Anabelle could have a Bone Marrow Transplant. Again she showed us her strength and courage by going through Chemotherapy and the transplant including all the drugs that went with it. Although there were some hard times we don't really remember those as Anabelle did not let these get her down always wanting to smile and play.

In September she came home to semi isolation which meant Leanne and myself having to work part time in order to care for her. This meant we had so much quality time with her and helped me understand the importance of family life.

She began to catch up with her milestones and wanted to be on her feet all the time, her makaton sign language was really progressing and she even began to say a few words. Sharon a friend from Great Ormond Street had bought her a talking teddy and Anabelle constantly played with it. I can tell you it drove us mad but Anabelle learnt to say Hi yer and bye, bye by playing with it.

Her Laughter

Throughout her life she always carried the most infectious smile and you could not help by smiling in return. During the days of Enzyme Replacement Therapy we would take the train home which would be filled with world weary London commuters. On the packed train she would poke her head around the seats and give the commuters smiles and laughter. They had no choice but to laugh and smile back.

Myself and Leanne have never laughed so much as we did in Anabelle's company. Anabelle could be a bit of a diva as well (I don't know where she got that from). She was a massive Peppa Pig fan and also loved the petit filous yoghurts so we would buy her the brand yoghurts. She would check the side of these yoghurts and if wasn't a Peppa Pig one she would refuse to eat it and throw a strop.

Her Love

She was an extremely affectionate child and an extremely loved daughter, granddaughter, sister, niece and friend. Wanting nothing but to snuggle up and cuddle. She would blow people kisses. Her final Christmas present which was from her sister Leah was a book titled 'this is how much I love you' and this pretty much summed up her life.

During the Great South Run her love inspired many friends and family to run for the MPS Society. It also helped some of us lose their beer bellies!

I just want to finish off with the lessons Anabelle has taught us;

She reminded us about the important things in life. It isn't just about work and earning a decent salary. It is about making sure you have good quality time with your family. Within the first year of her life, during the week I would be lucky to see her five minutes in the morning and then getting home to read her a story and put her to bed, just like any other dad really. However after she came home from Great Ormond Street I would look after her Mondays and Tuesdays and these were the best days of my life and I look back on them with fondness. I no longer get stressed about small things such as work and things that happen in the street or on the road. These are small inconsequential things.

After her diagnosis I viewed disabilities in a new light. Even though she was not really affected by her condition I was able to see the child before the disability, rather than just seeing a disabled child. For these lessons I thank her and am eternally in her debt. You are our shining star, Love you our little angel.

Lee Shepherd

Bereavements

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

Ian Fred Shead and his Baby Holly

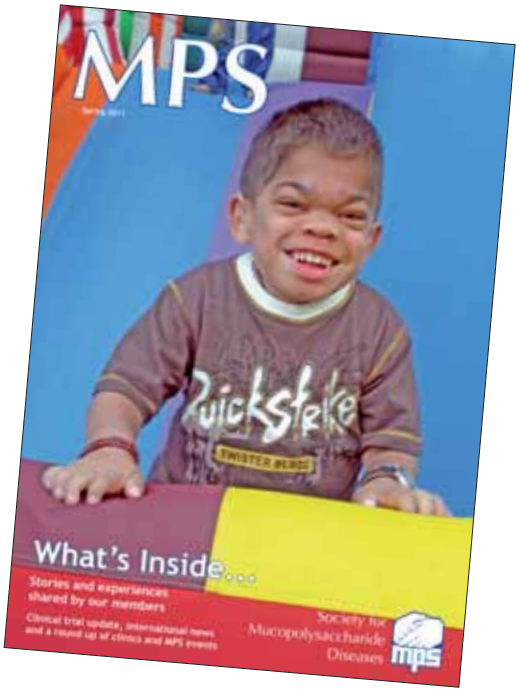
Ian Shead who suffered from Hunter disease and who passed away on 14 January 2013 aged 40 years.

"Ian was an inspiration to us all, and to whoever met him. Ian did not let life stop him or beat him down. Ian was and is loved by all his family and friends. Sleep well my baby love."

Mummy & Daddy XXX

Naim Anwar who suffered from Hurler Scheie and who passed away on 12 February 2013 aged 17 years.

Parth from Delhi, India who died on 12 January 2013. Parth suffered from Hunter disease and featured on the front cover of our MPS Magazine in Spring 2011.



Wear It BLUE

MPS Awareness Day
15th May 2013

Help us mark International MPS Awareness Day

This is a day for all the individuals and families worldwide who have been affected by MPS diseases. It's a day to celebrate the advances in research undertaken by dedicated scientists over the years and an opportunity to celebrate the treatments now available and those yet to come.

Join us in marking this significant day in the MPS Calendar

- **Take part in an event, be it big or small, simple or crazy.**
- **Wear an MPS awareness ribbon and share with people what it means to you.**
- **Wear it Blue and get those around you to do it too.**

Read on in this MPS Magazine for fun ways to raise much needed funds whilst raising awareness.

Visit **www.mpssociety.org.uk** for more information or give us a call on **0845 389 9901** to explore the different ways you can get involved.

Childhood Planting

Sherwood Pines, Nottinghamshire
21 October 2012

1

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On a beautiful autumnal day, and for the first time on a Sunday, the MPS Society held its 20th planting in the Childhood Wood for children and adults who have lost their lives to MPS, Fabry and related diseases.

The day started at the Clumber Park Hotel where families and dignitaries gathered and enjoyed Sunday lunch together. Commander Judith Helen Swann, Councillor Keith Walker and Lady Inga Walker welcomed everyone to Nottinghamshire and told of their ongoing interest in the Childhood Wood.

We all then drove in convoy to Sherwood Pines where we met in the car park and walked up to the Childhood Wood along with the balloons.

The Foresters had done an amazing job clearing and preparing the area for the new saplings right down to digging the holes. The MPS Remembrance Boards were in place depicting the names of the seven children and adults to be remembered at the planting. During the short ceremony the names of those remembered were read out by Sue Peach (MPS Chair) and Wilma Robins (MPS Trustee) read the poem by Christina Rosetti, 'Remember'.

Christine Lavery

1. Daniel Singh's family
2. Archie Rudham's family
3. Colin Arrowsmith's family
4. Sara Hyde's family

MPS Awareness Day
15th May 2013

Help us mark International MPS Awareness Day

This is a day for all the individuals and families worldwide who have been affected by MPS diseases. It's a day to celebrate the advances in research undertaken by dedicated scientists over the years. It's an opportunity to celebrate the treatments now available and those we hope are yet to come.

Join us in marking this significant day in the MPS Calendar.

You'll find more information about how you can get involved in MPS Awareness Day in this magazine.

Whether you want to **Wear It Blue**, go for a **Wicked Walkabout**, or simply make a donation by text, there is something for everyone.

Donate a £1 and Wear it Blue
for MPS Awareness day on 15th May!

With MPS Awareness Day rapidly approaching it's time to dig out those navy slacks, dust off that royal blue sweater and accessorise that sky blue boater.

This is your opportunity to encourage colleagues, friends and classmates and spread the word by proudly wearing blue to raise awareness of these genetic diseases.

The premise is simple – encourage people to pay a £1, wear blue, help to save lives. Easy.

Order your posters and leaflets today: wickedgenes@mpssociety.org.uk

Or . . .

An all blue outfit not your thing? Accessorise with a sapphire coloured scarf or let us sneak a peak of your azure blue socks.

Go Bonkers in Blue – wear fancy dress for the day by turning up ship shape Bristol fashion as a sailor or walk the beat dressed as a local bobby.

Whatever you do, pay £1 and Wear It Blue!

Visit **www.mpssociety.org.uk** for more information or give us a call on **0845 389 9901** to explore the different ways you can get involved.

Where does your money go?

A donation of £2 per month could help us to offer so much more support in so many ways:

- Access to clinical management and palliative care
- MPS Regional Specialist clinics
- Support with disability benefits
- Paving a child's way in accessing education
- Upholding rights in employment
- Advising on home adaptations
- Bereavement support

Please donate **online** at
www.mpssociety.org.uk
phone 0845 389 9901
text MPSS01 £2/£5/£10 to **70070**
or **post** your donation to our office, MPS House.

The MPS Advocacy Service

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

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

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

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

Support provided by the team

- **Telephone Helpline** – 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 Disability Living Allowance claim forms, the Society continues to provide help and support in completing these forms and, where needed, will take a representative role in appeals and tribunals
- **Housing and equipment** – the Society continues to take a major role in supporting and advocating appropriate housing and home adaptations to enable the needs of an individual with an MPS or related disease to be met. Where requested, we can provide comprehensive and detailed housing reports based on individual need
- **Education** – the Society helps members to access appropriate education and adequate provision for its implementation. This is achieved through providing educational reports used to help inform and educate professionals, and in many instances, to inform Statements of Special Educational Need. Where requested, we also provide information days/ talks to schools and relevant professionals
- **Respite Care** – the Society continues to work closely with a number of respite providers and, where appropriate can make individual referrals
- **Independent Living/ Transition** – the Society provides advice, information and support through the transition from child to adult services. This could include access to independent living, learning to drive, further education and employment
- **MPS Careplans** – the Society undertakes a comprehensive assessment of the issues which need to be addressed when caring and providing support to a specific individual diagnosed with an MPS or related disease, as well as other family members through the writing of a careplan
- **Befriender Service** – the Society links individuals and families affected by MPS and related diseases for mutual benefit and support
- **Bereavement support.**

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

Email:
advocacy@mpssociety.org.uk

Telephone: 0845 389 9901

Advocacy Resources

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

- Disability Living Allowance
- 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

SOPHIE
Manages the MPS Advocacy Team

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

Team members

REBECCA
Fabry
MPS II Hunter
ML III / ML IV
Mannosidosis
Fucosidosis

STEVE
MPS III Sanfilippo
MLD
AGU
Winchester
Geleo Physic Dysplasia
SLY
Gangliosidosis
Sialic Acid Disease

DEBBIE
MPS IV Morquio
MPS 1 Hurler BMT,
Hurler Scheie, Scheie
MPS VI Maroteaux-Lamy
MSD,
ML II

ALISON
Supports all members living in Ireland

CHANGES TO HOUSING BENEFIT - WHAT YOU NEED TO KNOW

Changes to how housing benefit is awarded will be coming into force in April 2013.

If you live in a Council or Housing Association home and you have a "spare" (please see below for government definition of a spare room) room then your benefit could change.

This will affect everyone between the age of 16 – 61 years and it does not matter how much or how little benefit you receive, even if you are working. It could also affect you even if you are sick or disabled. You won't be affected if you live in a one bedroom flat or bedsit or if you or your partner receives pension credits.

Definition of a "spare" room

If you have more bedrooms than the government says you need, you will lose part or all of your housing benefit. The new rules state that you need one bedroom for;

- Each adult couple
- Any single person over the age of 16 years
- Two children of same sex under the ages of 16 years
- Two children under the age of 10 years, regardless of their sex
- Any additional child
- A carer (who does not normally reside with you) if you or your partner requires overnight care.

These new rules apply to everyone, even if:

- You and your partner sleep separately because of a medical condition
- The main residence of your child / children is another address but you have a "spare" room for when they stay with you.

So what does this mean?

If you have one "spare" bedroom your housing benefit will be cut by 14% of the rent you pay every week. If you have two or more spare bedrooms you will lose 25%.

This means that you will have to pay the additional cost of rent no longer covered by housing benefit.

Example

Mr & Mrs Briggs have two children aged 3 & 5 years and live in a three bedroom house. Their rent is £120 per week and they receive £20 toward this through housing benefit. Under the new rules their children must share a bedroom, leaving one "spare" room. Therefore, their housing benefit will be reduced by 14% of their rent (14% of £120 = £16.80). Under this new scheme this will mean that they will only be entitled to a contribution of £3.20 from Housing benefit instead of the £20.00 they were receiving and will have to top up the rest themselves.

Your local council should be contacting you directly regarding this new proposal and will undertake an assessment of your current living arrangements.

What if I have a severely disabled child ?

If you have a severely disabled child who needs a room of their own, they may not be required to share a room. You should speak to a benefit advisor regarding this.

What if I live in an adapted home?

If you live in an adapted home you will not automatically be exempt. However, if it is found that it is not practical or affordable for you to move you may be able to apply to your local council for a discretionary housing payment if you are having trouble paying rent or council tax and qualify for either benefit.

Applications should be made to your local council who will assess each case individually. However, councils are only given a small pot of money each year and once it is spent, there is no more available until the following year.

For further information please go to the Department of Work and Pensions

www.dwp.gov.uk/adviser/updates/housingbenefit/

or contact the
Advocacy Support Team
by phoning 0845 389 9901
or emailing
advocacy@mpssociety.org.uk

All Ireland Advocacy Support

It's 2013! Can you believe it? It seems like only yesterday I was writing my final 2012 magazine update on the All Ireland Advocacy and Support Service. I hope that you have had a good start to 2013 – but for those of you who have been facing more difficult times, I hope that you have been able to access the support that you need.

2013 has had an exceptionally busy start. I am delighted to have been invited, by local paediatricians, to the multidisciplinary team meetings for some of our MPS Society members; I have visited four schools (two in Northern Ireland and two in Southern Ireland) to provide education sessions about MPS; and I have been asked by 6 families to support them in drawing up care plans for their children and young adults. I have also welcomed new families to the MPS Society and supported patients at clinic appointments and meetings. I am so pleased that families are accessing the services available through the All Ireland Advocacy Support Service and would encourage you to point others in our direction.

MPS Clinic in Dublin

In February the Temple Street Metabolic Unit in Dublin ran a 2 day clinic for MPS and related diseases – this was a new clinic and we look forward to hearing feedback from those who attended.

At this clinic patients had access to Specialist Metabolic Consultants, a Dentist with a special interest in MPS, a Neurological Surgeon, and Cardiology and Radiology review. The clinic was an excellent opportunity to meet the families that I had previously only had telephone contact with – it's always great to put a face to a name! I also got to catch up with a few old (and not so old!!) friends.

This clinic highlighted a number of unmet support needs, both on individual and service wide levels, that I will be spending the upcoming weeks and months addressing. I look forward to updating you on our progress in Southern Ireland.

Alison Wilson a.wilson@mpssociety.org.uk

Rare Disease Day

28th February 2013

As I have explained before, I am part of the Northern Ireland Rare Disease Partnership (NIRDP). Although this is not part of my 'day job' it is an excellent opportunity to fly the flag for MPS and related diseases amongst those with an interest in developing services for patients with rare diseases.

Alison Wilson

On 28th February (International Rare Disease Day) the NIRDP were involved in the Rare Disease Without Borders Event in Dublin – this was a cross border event, so Northern Irish and Southern Irish representatives were in attendance. I was delighted to be able to represent both the MPS Society and the Northern Ireland Regional Genetics Service at this event.

Please see a copy (on the right) of the All Ireland Advocacy and Support Service poster that was displayed at the event.

For me the highlight of this event was hearing Minister Edwin Poots MLA (Minister of the Department of Health, Social Services and Public Safety N.I.) and Minister Alex White TD (Minister of State for Primary Care in the Department of Health R.O.I) pledge their support for cross-border work for Rare Diseases.

We look forward to developing these North/South links in the year ahead and hope that by Rare Disease Day in 2014 we will have major advances to report.

Alison Wilson

All Ireland Advocacy and Support - The Society for Mucopolysaccharide Diseases (MPS Society)

Adapting to Changing Needs

The Society for Mucopolysaccharide Diseases has provided an Advocacy and Support Service for its members in the United Kingdom for over 30yrs. Over time this service has changed to meet the changing needs of a cohort of patients who have seen major advancements in the treatment and management of their conditions.

However, the core aims of the MPS Society have remained the same:

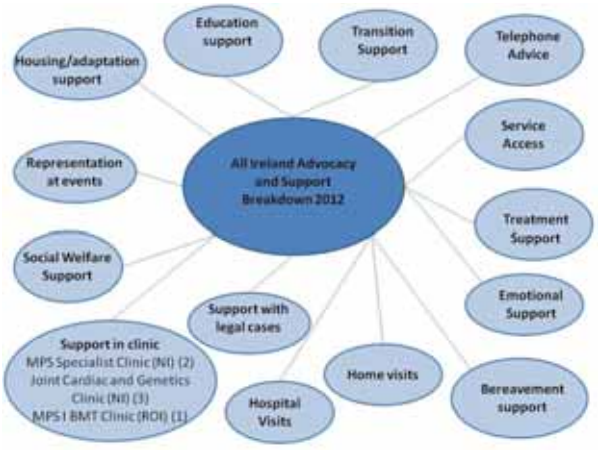
To act as a **support** network for those affected by MPS & related diseases
To promote and support **research** into MPS & related diseases
To bring about more public **awareness** of MPS & related diseases

The **All Ireland Advocacy and Support Service** was established in 2010.
The aim of this service is to provided individuals across the island of Ireland with a local an accessible source of support.

Importance

Condition	Number known to MPS Society	
	NI	ROI
MPS I – Hurler/Hurler Scheie/ Scheie Disease	10	45
MPS II – Hunter Disease	4	10
MPS III – Sanfilippo Disease	4	2
MPS IV – Morquio Disease	13	6
MPS VI – Marotaeux Lamy Disease	1	1
GM1 Gangliosodisis	1	
Metachromatic Leukodystrophy		1
Fucosidosis	1	
Mucolipidosis Type II (Psuedo- Hurler Polydystrophy)	1	1
Mucolipidosis Type II (i cell Disease)	1	1
Winchester Disease		1
Fabry Disease	26	19

Advocacy and Support Breakdown 2012



A New Service – Providing Cross Border Support

Advocacy and Support Officer based in the Department of Medical Genetics in Belfast.

This worker travels throughout Ireland to meet families in their homes and attend meetings.

Support is also provided by letter and/or telephone where this is sufficient.

Society for Mucopolysaccharide Diseases

The Society for Mucopolysaccharide Diseases is a registered Charity No. 1143472. Registered as a Company limited by guarantee in England & Wales No. 7726882.
Registered office at MPS House, Repton Place, White Lion Road, Amersham, Buckinghamshire, HP7 9LP. Registered as a Charity in Scotland No. SCO41012.
Tel: 0845 389 9901, Fax: 0845 389 9902
www.mpssociety.co.uk


Sultan (MPS IV)

Birmingham MPS Clinic

8th January 2013

Steve and I had been up to Leeds the day before on visits and then stayed overnight in Birmingham.

Luckily there was no snow but it was very cold.

Sultan arrived with his mum and we learnt all about the marine life in the Barrier Reef on the TV while he was waiting, we did eventually have to turn it off as CBeebies can only be tolerated for a short time.

Ali was camera shy so we left him in peace.

Xena and **Roman** started to decorate paper hands that had been left for the patients to decorate for Rare Disease Day. Unfortunately they didn't have time to finish them so Steve and I had to attempt to finish them off!


Caitlin (MPS III)

Caitlin was a little star and sat and watched what was going on, I didn't get too close as she took a liking to my glasses and I really needed them in one piece.

There was lots of comings and goings and eventually it was time for us to drive home. It was a lot easier driving out of Birmingham than it was getting in the night before.

Thanks to all the staff for looking after us, see you all next time.

Rebecca Brandon & Steve Cotterell


Jack (MPS III)

Newcastle MPS Clinic

I stumbled bleary eyed into the Royal Victoria Infirmary on the 26th February and was met by an equally tired Dr Jones he too had a 4.30am start, although he was canny enough to have bought a coffee to perk himself up. Dr Rylance soon arrived and the clinic began. It was good to see cheeky **Jack** and chatty **Dominic**. **Daniel** I think would rather have been in bed as would **Luke** after a fairly early start. We would like to thank Dr Rylance and the team for hosting the clinic and look forward to the next one.

Steve Cotterell,
Advocacy Officer


Daniel (MPS II)

Great Ormond St. MPS IV clinic

An MPS IV clinic was held on 8th January at GOSH, our new worker Debbie Cavell accompanied me on her second day with us to come and meet the families that she will be supporting. It was good to see **Hayaan** here, there and everywhere as usual, and it proved difficult to drag **William** away from the games console. We also had the opportunity to meet some new families as well and will look forward to seeing them again. We would like to pass our thanks to Dr Vellodi, Niamh and the team for supporting this clinic.

Steve Cotterell,
Advocacy Officer


Xena (MPS IV)


Roman (MPS IV)


Dominic (MPS IHS)


Luke (MPS III)


Hayaan (MPS IV)

Wicked Walkabout Launch!

Launch of the Wicked Walkabout

You'll all be familiar with other big charity events such as coffee mornings, races and red noses. Well we thought it was time we had our own and so we're delighted to announce the launch of the MPS Wicked Walkabout programme, to coincide with MPS Awareness Day on May 15th.

The US MPS Society has a Walk/Run programme that has been running for 12 years and has raised nearly $4million. Most importantly it has raised awareness of MPS and related diseases across the USA, with events taking place all through the year and nationwide. We hope to have Wicked Walkabouts throughout the year for the MPS Society.

What is a Wicked Walkabout?

It's up to you! A Wicked Walkabout can be a gentle walk, a run or a cycle. We recommend a minimum of 5km, but that is just a guide and could be longer (or shorter if it's a Wicked Wellie Waddle!). The Wicked Walkabout is about raising awareness through fundraising for those affected by MPS and related diseases. Have a think about how you would like to do this, it may be you set a minimum sponsorship for people to enter your event or perhaps charge an entrance fee. It's your event, your choice!

How:

If you think you would like to hold a Wicked Walkabout, get in touch and we can send you the Wicked Walkabout Guide email **wickedgenes@mpssociety.org.uk**

Wicked Walkabout Update

You may remember in the last edition of the magazine we launched our new event, the Wicked Walkabout. We are delighted to announce we already have 10 events planned for this year and we'd love to get to 20. If you are interested in finding out about Wicked Walkabouts then check out this magazine, or drop us a line on wickedgenes@mpssociety.org.uk

28th February 2013

Rare Disease Day is an international awareness campaign that takes place on the last day of February every year. The main objective of the day is to raise awareness about rare diseases and their impact on patient's lives.

The campaign targets the general public and policy makers but everyone is welcome to join in; patients and their families, patient organisations, health professionals, researchers, drug developers, public health authorities.

Rare Disease Day was first launched by The European Organisation for Rare Diseases (EURODIS) and its Council of National Alliances in 2008. Since then, thousands of awareness raising activities have taken place all over the world, reaching thousands of people and generating national and international media attention.

Rare Disease Day has notably contributed to the advancement of national plans and policies for rare diseases in a number of countries. The campaign which started as a European event has progressively become a world event with a total of 62 countries taking part in 2012.

2013 Rare Disease Day's slogan was 'Disorders without Borders' and a 1 minute promotional animated film was produced to highlight the international dimension of the campaign.

The Independent Newspaper's Rare Disease Supplement

To coincide with Rare Disease Day the Independent Newspaper produced a Rare Diseases Supplement. We are delighted to announce Christine Lavery, Chief Executive of the MPS Society, was interviewed for an article about how patient power can transform access to treatment.

See below.

WITHIN THE INDEPENDENT FEBRUARY 2013 · 11

How patient power can transform access to treatment

Patient groups are making their voices heard in the reformed NHS. The ideal result? Fairer access to treatments, greater patient support and increasing research.

SPEAKING OUT

Christine Lavery, chief executive of The Mucopolysaccharide Society, for people affected by Mucopolysaccharide disease (MPS), is a firm advocate of patient power.

"Over 95 per cent of people with MPS are members of the society, which offers an advocacy service to individual patients," says Lavery.

Its work benefits doctors and researchers too. "We inform patients about research and clinical trials and provide financial support for patients who otherwise could not take part," says Lavery.

The 2010 Equity and Excellence White paper included the phrase "Nothing about me without me". However, there have been concerns that the patient voice hasn't yet been hard-wired into the new NHS structures. The MPS Society is part of a broader coalition of LSD Patient Associations representing all 50 lysosomal storage diseases which have been lobbying Government.

Christine Lavery
Chief executive, Mucopolysaccharide Society

Patient involvement

Lavery continued: "In partnership with our clinical and commissioning colleagues we have contributed significantly to the shaping of the current service for the very rare, ultra orphan diseases. During this period of change within the NHS we feel it as important as ever that the patient perspective has strong representation. The patient must be central to all decisions not only into existing and new services but into the evaluation of new technologies."

The MPS Society was instrumental following the last major NHS reorganisation. "We won the battle in 2005, so patients with LSDs in England can access the same level of treatments without being subject to a postcode lottery," says Lavery.

"Recently, we also won a commitment from the Department of Health that enzyme replacement therapy, used to treat many LSDs, will continue to be offered to patients England wide under the new NHS commissioning board."

LINDA WHITNEY
info.uk@mediaplanet.com

ERT for LSDs Standard Operating Procedures

We all know that these are difficult times financially for the public services as well as for individuals and families personally. Over the past six months the AGNSS LSD Expert Advisory Board have had to agree some efficiencies in current prescribing practices that will ensure benefit to newly diagnosed patients now and in the years to come. These decisions have not been made lightly and the LSD Patient Organisation along with clinicians have all been asked to buy into these efficiencies. Recognising the significance of these decisions the LSD Collaborative with each CEO accompanied by two Trustees from each of the Patient Organisations met to consider the efficiencies proposed. Sue Peach, Chair and Jessica Reid (Trustee and lawyer) represented the MPS Board of Trustees. It was recognised that whilst there may be a small number of patients and their families unhappy with the efficiencies the LSD Collaborative and attending Trustees agreed unanimously that these were in the best interest of the LSD community as a whole and communicated this decision to Commissioners late last year.

Key efficiencies agreed:

- across all LSDs the maximum number of infusions a new patient will have at the hospital before transferring to Homecare has been standardised and shortened across England. Time scales vary from LSD to LSD. If clinically indicated hospital infusions will continue until patient can safely go on Homecare. ERT in hospital incurs VAT where as ERT delivered in a Homecare setting does not!
- In Pompe and Gaucher disease ERT will be prescribed in patients 18 years and over against a Body Mass Index (BMI) of 27 where a patient's BMI exceeds 27
- In Gaucher Type 1 disease VPRIV will be the first line treatment
- Criteria for not starting ERT in Fabry disease changed. *(If you are concerned please see your Fabry specialist)*
- Fabry patients on Replagal will not be switched back to Fabrazyme unless overriding clinical reasons (study pending)
- MPS II, Hunter – Patients with confirmed MPS II and no CNS disease will be treated with ERT
- MPS II Patients progressing to CNS disease the standard procedure will be to stop ERT. A phased reduction in weekly infusions may be offered. Palliative care arrangements should be in place before treatment is stopped
- Patients presenting with severe CNS disease will not start ERT

There were no changes at this time to the Standard Operating Procedures for MPS Hurler Scheie and Scheie.

If you are concerned by anything you have read in this article please do email me at c.lavery@mpssociety.org.uk

Christine Lavery

A Clinical Trial for Genistein Update

First of all I wish to thank all the families of children and young adults with Sanfilippo disease for their enormous fundraising efforts to secure the money needed to start the Clinical Trial for Genistein in MPS III, Sanfilippo, at the University of Manchester.

At the outset the overall cost of the trial was £800,000 and as a proportion of this had already been raised by the MPS Society, only a further £600,000 was needed. This was the target set as of October 2012.

In the meantime, the MPS Society has been working closely with Dr Brian Bigger at the University of Manchester to minimise the cost of the trial so that the total research grant now needed is £650,000 rather than the original £800,000. Meanwhile, Dr Bigger has also been able to budget to increase the number of clinical trial participants from 20 to 30 patients which is really good news.

The £200,000 that had already been raised by the MPS Society comes from a research grant from the US National MPS Society and contributions pledged by the MPS Societies around the world.

The income for funding the clinical trial currently stands at:

£150,000	**fundraised by the UK MPS Society**
£ 50,000	**Donation from the GEM Appeal (Karen Hoather)**
£ 50,000	**fundraising pledged to the MPS Society from fundraisers**
£ 30,000	**Contributions pledged by the MPS Societies around the world**
£170,000*	**Research Grant from the US National MPS Society**

TOTAL raised or guaranteed to date £450,000

TOTAL still to raise is £200,000

*It should be noted that the Board of Directors of the National MPS Society are only committed to paying the £170,000 Research Grant to this Genistein Clinical Trial up to 30th June 2013. We are advised that if the Genistein Clinical Trial is not started by 30th June 2013 their grant offer will be withdrawn. **The next 3 months are critical to securing the remaining £200,000 and we need all the help we can muster from not just the UK Sanfilippo families but all the Sanfilippo international community.**

Scottish Medicines Fund

The Scottish Government has launched a fund to cover the cost of medicines for individual patients with rare conditions, which are not available for routine prescription.

£21 million will be invested to pay for the cost of medicines known as "orphan drugs". These are medicines for illnesses which affect fewer than 1 in 2,000 people.

Only those medicines not recommended for routine use by the Scottish Medicines Consortium will be covered by the fund.

The new fund has been put in place following interim advice from Professor Charles Swainson who is examining the current Individual Patient Treatment Request (IPTR) arrangements in the NHS in Scotland, as part of the ongoing access to new medicines review.

The fund will be available from March this year through to April 2014 and will help to cover successful new Individual Patient Treatment Requests.

Health Secretary Alex Neil said:

"It is only right that Scottish patients with rare conditions have access to innovative medicines which are clinically justified and that they are not disadvantaged due to the very high cost of these treatments.

"I am therefore pleased to confirm today that the Scottish Government will establish a fund, which will ensure that the cost of successful new individual patient treatment requests for orphan medicines are met.

"This fund bridges the period to the establishment of next year's value based pricing for medicines and any changes that are made following the completion of the ongoing access to new medicines review.

"We recognise that the Scottish Medicines Consortium is globally respected and has the fastest and most efficient medicine review process anywhere in the UK, but that for some individual patients with rare conditions there is a need for further support. This new fund will complement it by making the IPTR processes fairer."

Requests to the fund will be assessed on a case by case basis, based on the clinical circumstances of individual patients. Detailed operational arrangements will be developed and announced in due course. The fund will be managed by NHS National Services Scotland.

The EU's definition of an "orphan medicine" is defined as one for which the frequency of the disease is less than 5 per 10,000 of the EU population.

The £21 million is in addition to the current board funding allocations.

The Scottish Medicines Consortium (SMC) appraises all newly licensed medicines and provides advice to NHS Boards on their clinical and cost-effectiveness.

Currently, if a medicine is accepted for use by the SMC then individual health boards set the criteria for prescribing it. If the medicine is not accepted then health boards do not make it routinely available. However, clinicians can prescribe medicines that are not accepted for routine use by the SMC for individual patients in certain circumstances by special request through Individual Patient Treatment Requests.

A review is underway to assess the current systems for making new medicines available across NHS Scotland. This review is being led by Professor Philip Routledge. Separate to this work the Chief Pharmaceutical Officer Professor Bill Scott, with Professor Charles Swainson, has been reviewing the Individual Patient Treatment Request arrangements to establish whether any further improvements can be made. This new fund is being made as a result of Professor Swainson's interim advice.

Enzyme Replacement Therapy in the Republic of Ireland

As we go to press we have learnt that due to budgetary constraints children with Hunter disease will not see an increase in their Enzyme supply as they grow and gain weight. The implication of this is that over time these Hunter children will be receiving lower and lower unlicensed doses of Elaprase.

As a consequence there are concerns that these children's Hunter disease will progress due to lack of adequate enzyme.

If you are a family affected by this decision in respect of Hunter, MPS I or MPS VI or your child is recently diagnosed and being denied enzyme replacement therapy please do contact Alison Wilson via e-mail at a.wilson@mpssociety.org.uk or telephone 0044 (0)2895 047779.

New NHS National Commissioning Board

AGNSS Update

First of all I need to thank all of you, the hundreds of MPS and Fabry families who last August and September wrote to your Member of Parliament, to Prime Minister, David Cameron and Deputy Prime Minister, Nick Clegg. I can tell you now that your efforts really made the difference and I can assure you that the importance of the Highly Specialised Service for Lysosomal Storage Diseases is now recognised at the highest levels.

Your letters opened doors for the LSD Patient Organisation Collaborative (LSD Collaborative) of which the MPS Society is a major player to advocate for you in the new NHS. It would be fair to say that your letters and our high level communications strategy has paid dividends. The LSD Collaborative has had constructive face to face meetings with Kate Caston, Head of Specialised Commissioning, and Barbara Howe, Lead Commissioner. In our discussions we have been keen to retain all that serves LSD patients best from the AGNSS model and to demonstrate the benefit the LSD Collaborative brings to the table

Since my meeting with two MPS II constituent families and Jeremy Hunt, Secretary of State for Health in November we have kept in close touch and have recently acknowledged the the understanding that is now developing between the LSD Collaborative and the NHS Commissioning Board. Having said that we have again highlighted our concerns relating to the yet to be agreed new appraisal system for high cost ultra orphan drugs to be rolled out by NICE and of concern to our MPS IVA families.

In our call to action we set out to safeguard for LSD patients four key areas that in the LSD Collaborative's view are integral to the future of the highly specialised service for LSD patients. What have we achieved?

- The LSD Expert Advisory Board (that includes the LSD patient organisations) will continue to meet regularly with clinicians and the National Commissioners for this highly specialised service. The LSD Collaborative will continue to be an effective partner in getting ERT and new therapies to patients whilst recognising that efficiencies that are fair, safe and equitable will have to be made.

- The LSD Collaborative originally sort to protect the current ring fenced budget for the cost of Enzyme Replacement Therapy. During our letter writing over 450 LSD families received reassurances from Earl Howe, UK Health Minister that patients currently prescribed enzyme replacement therapy are not at risk of this being stopped. Also during our discussions with Commissioners we could see that limiting the LSD ERT to a ring fenced budget which would not increase over coming years would mean no growth for newly diagnosed patients or new ERTs coming on stream. On this point the LSD Collaborative agreed in these times of austerity being a player for a much bigger pot of money offered LSD patients the best opportunity of their treatment being funded.

- Being grouped in the Metabolic Clinical Reference Group (CRG) which has over 700 rare diseases the vast, vast majority of which sadly there is no treatment was and continues to be unacceptable to the LSD Collaborative not least because marginalised we see our members facing a lesser service. Equally it would spell the end of the innovative collaboration and joint working that has evolved over the last five years between clinicians and the Commissioners. This point was fully appreciated by both Kate and Barbara Howe. I am pleased to say a CRG for Highly Specialised Services has been introduced and the LSD Collaborative looks forward to working in this space. Clearly we can't and wouldn't wish to be totally isolated from the metabolic disease family so the LSD Collaborative have sent a proposal to Barbara Howe of how we might link in.

- Finally we come to the abolition of the AGNSS model for appraising new high cost therapies for ultra-orphan diseases. This was the nut we were never going to crack easily if at all! With AGNSS gone the process for these appraisals had to be homed somewhere. It is no surprise the National Institute for Clinical Excellence (NICE) will now take over this role. The best engagement we have at the moment is taking part in regular stakeholder meetings with NICE on the design of the new appraisal system for ultra orphan drugs. There is no way of knowing at present whether the LSD Collaborative is there to tick a box or seen as a valued part of a process that we expect to achieve a fair, transparent and equitable appraisal process that wholeheartedly recognises that our members did not chose to have an LSD or have children with these conditions. I will attend the next NICE stakeholder meeting on 8 April in Manchester and only time will tell.

Christine Lavery

NHS Commissioning Board
A special health authority

Amicus Therapeutics presents Additional 6-Month Results from Phase 3 Fabry Monotherapy Study at LDN World Symposium

CRANBURY, NJ and ORLANDO, FL February 15, 2013 –
Amicus Therapeutics today announced additional 6-month (Stage 1) results from the first on-going Phase 3 global registration study (Study 011) of investigational oral migalastat HCl monotherapy(150 mg, every-other-day) in males and females with Fabry disease who had genetic mutations identified as amenable to migalastat HCl in cell-based assay.

Study 011 consists of a 6-month, double-blind period (Stage 1) when subjects received migalastat HCl 150 mg or placebo, a 6-month open label-follow-up period (Stage 2) when all patients received migalastat HCl, and an on-going 12-month open-label extension.

As previously reported in December 2012 the primary endpoint in Stage 1 did not meet statistical significance. The pre-specified primary and secondary analyses of the primary endpoint numerically favoured migalastat HCl over placebo. In the primary responder analysis, 13/32 (41%) in the migalastat HCl group verses 9/32 (28%) in the placebo group demonstrated a 50% or greater reduction in kidney interstitial capillary GL-3 from baseline to month-6 (p=0.3). Taken alone the secondary analysis of the absolute percent change in the kidney interstitial capillary GL-3 from baseline to month-6 showed a median reduction of 41% in the migalastat HCl group verses a median reduction of 6% in the placebo group (p=0.093).

Study 011 entry criteria particularly elevated urine GL-3, were intended to enrich for patients with higher interstitial capillary GL-3, and more measurable disease burden. Although all patients had detectable interstitial capillary GL-3 at baseline, a number of patients had low levels of GL-3 at baseline, making it difficult to detect a significant difference in responders between the two treatment groups. Clearance of kidney interstitial capillary GL03, a marker of treatment effect, is being measured by histology2 in evaluable kidney biopsies from baseline to month 6 (Stage 1) as well as baseline to month 12 (Stage 2). Stage 2 results remain blinded at this time.

Dr Ezgu reported at LDN WORLD that *"The 6-month data from Study 011 are encouraging, including a post-hoc subgroup analysis presented today, and we continue to evaluate patients in this on-going study. There is still an unmet medical need for Fabry disease treatments. Migalastat HCl may potentially offer an oral treatment for Fabry patients with amenable mutations based on these results from Study 011 and earlier clinical studies, and potentially forthcoming data from on-going studies."*

Study 011: Stage 1 (Baseline to Month 6) Updated Data Highlights and Presented at LDN WORLD Symposium:

- **Post-Hoc Subgroup Analysis of Primary Endpoint (modified intent-to-treat (mITT), n=60):** In a post-hoc sub-group analysis, patients with higher baseline disease burden (0.3 inclusions or more per interstitial capillary, n=25) were compared to those with a lower baseline disease burden (0.3 or fewer inclusions per interstitial capillary, n=35). In the 25 patients (14 males and 11 females) with higher disease baseline burden, 7/11 (64%) on migalastat HCl and 2/14 (14%) on placebo were classified as responders. Among the 35 patients (8 males and 27 females) with lower baseline disease burden, 6/19 (32%) on migalastat HCl and 7/16 (44%) on placebo were classified as responders.
- **Urine GL3:** The observed median reduction in urine GL-3 from baseline was 17% for placebo and 12% for migalastat. However due to unexpected variability in the pre-treatment urine GL-3 data (>1.6 fold difference between values at screening and baseline), any potential treatment effects on urine GL-3 cannot be determined.
- **Renal Function from Baseline to Month 6:** Renal function remained stable and changes from baseline were similar in both treatment groups during stage 1. The mean (SD) increase in estimated glomerular filtration rate (eGFR) was 2.7 (15.1) mL/min/1.73m2 in the migalastat HCl group compared to a mean decrease of 2.4 (10.8) mL/min/1.73m2 in the placebo group. No clinically meaningful changes in proteinuria were observed, and iohexol GFR data currently being analysed.
- **Safety:** During the first 6 months, no drug-related serious events have been observed. No subjects discontinued migalastat HCl therapy due to a treatment emergent adverse event and the majority of adverse events in both treatment groups were in mild nature.

John F. Crowley, Chairman and Chief Executive Officer of Amicus stated, *"The 6-month results presented today at the WORLD Symposium support our commitment to the on-going development of migalastat HCl monotherapy. We look forward to reporting the 12-month results from this study to add to the entirety of the data that the FDA has indicated would support a potential U.S. conditional approval."*

continued ▶

Enzyme Replacement Therapy Clinical Trial for MPS IH Patients Post HSCT

For some time MPS I member families have been expressing disappointment that having had a Bone Marrow or Cord Blood Transplant for their child with MPS I Hurler disease that is it as far as other therapies are concerned.

Bone Marrow Transplant was introduced as a therapy for MPS I Hurler by Professor Jack Hobbs in 1980. In the late 1990's Cord Blood Transplant was also introduced and these treatments now referred to as Hematopoietic Stem Cell Transplant (HSCT) is the recognised treatment of choice for children with Hurler disease.

Sixty or more children in the UK who have had HSCT owe their lives to this treatment but it is not a perfect science. Many of the children treated with HSCT whilst doing well cognitively have progressive musculoskeletal disease affecting their mobility and impacting on their education and everyday life.

Not surprisingly the question is often asked ***'could ERT help MPSI post HSCT children long-term?'.*** Dr Paul Orchard, a world renowned HSCT specialist in Hurler at the University of Minnesota is running a two year clinical trial to look at the benefits of treating pre-pubertal children who have undergone HSCT for Hurler disease. The inclusion criteria are for children aged seven years to puberty.

So you might ask how do children in the UK fit into this clinical trial? After considerable thought we are delighted that AGNSS (soon to be part of the new NHS National Commissioning Board) has agreed to fund up to three children in England to be part of Dr Orchard's clinical trial in collaboration with Dr Ashok Vellodi at Great Ormond Street Hospital.

As this clinical trial does not have a commercial sponsor, the MPS Society working with its members is committed to raising the funds to support UK children recruited to this clinical trial. If you would like to know more please do contact c.lavery@mpssociety.org.uk at the MPS Society or Dr Ashok Vellodi.

Christine Lavery

Study 011 results from Stage 2 are anticipated in the second quarter of 2013. The U.S. Food and Drug Administration (FDA) has indicated that it will consider the entirety of the Stage 1 and Stage 2 efficacy and safety data from Study 011. A meeting with the agency is anticipated in mid-2013 to discuss the U.S. conditional approval pathway for migalastat HCl under subpart H.

A second phase 3 global registration study (012) study is also underway to compare open-label migalastat HCl to ERT to primarily support global registration. Study 012 (The ATTRACT, or FAB-AT1001-012 Study) is a randomised, open-label 18-month Phase 3 study investigating the safety and efficacy of oral migalastat HCl 150 mg QOD compared to standard-of-care infused therapy using ERTs (Fabrazyme® and Replagal®). This study achieved final enrolment of 60 total patients in December 2012.

Subjective and Objective Assessment of Hand Function in MPSIVA Patients

Christian J Hendriksz et al
Birmingham Children's Hospital NHS Foundation Trust

This abstract that appeared in the Journal for Inherited Metabolic Diseases (2013) 36:161-170 on MPS IVA, Morquio diseases challenges why as a skeletal dysplasia the impact of the abnormalities on the upper limbs has not been described?

The aim of the study was to assess the level of subjective and objective functions of the hands and upper limbs in patients with MPS IVA in order to advance the understanding. New understanding may change future management in this complex group of patients.

The methodology was to evaluate a series of ten patients with MPS IVA who are currently part of the enzyme replacement therapy clinical trial programme at Birmingham Children's Hospital to establish functional levels. A questionnaire was developed based on the principles of the World Health Organisation International Classification of Functioning. Patients were also measured for grip function and range of motion.

The results showed that all patients showed hyper mobility and limited control of the wrist. Their grip strength was reduced and they showed higher difficulty levels for activities involving strength. The researchers surmise that it is feasible that the wrist abnormalities may be the major cause of functional loss for MPS IVA patients. They conclude that the findings suggest the wrist itself in Morquio patients represents an area that should be focussed on to be able to improve these patients' functional abilities and therefore their level of day to day independence.

Lionization in Fabry Disease

by Joan Keutzer

The α-galactosidase gene is on the X chromosome. Males who inherit a mutated α galactosidase gene on their X chromosome will be affected by Fabry disease (because they only have one X chromosome, they are called hemizygous). The severity and rate of progression of disease in males are variable, but those who inherit a mutation are extremely likely to have signs and symptoms of the condition. Females who inherit a mutated α galactosidase gene on one of their two X chromosomes (because they have two X chromosomes they are called heterozygous) may or may not be affected by Fabry disease. Some females have no symptoms while others exhibit the full spectrum of Fabry disease symptoms. While the percent of females that are affected by Fabry disease is unknown and the expression of Fabry disease in females is virtually impossible to predict, there is a growing body of evidence, including from the Fabry Registry, that a large proportion of females will in fact have signs and symptoms of the disease, although most will be less severely affected than the males and will suffer the complications at an older age.

The amount of residual α galactosidase enzyme activity in a Fabry patient is thought to affect the severity of Fabry disease. Some patients (male and female) with Fabry disease have a reduced level of enzyme activity; others have no detectable enzyme activity. This is influenced by the nature of the mutation. Patients with residual enzyme activity may have a less severe clinical phenotype. In addition, other factors are thought to contribute to the expression of Fabry disease in females, including lyonization.

Early in female embryonic development, one of the two X chromosomes is inactivated in each cell. Genes on the inactivated X chromosome are not expressed. This process is named lyonization after Mary Lyon, a British geneticist who first explained the phenomena. Lyonization assures that males and females have the same "dose" of genes that are found on the X chromosome. Otherwise, because females have two X chromosomes and males have one, females would have twice the "dose" of X linked genes.

Lyonization occurs early in embryonic life, beginning at about 3 days (when the embryo has about 36 cells) and is not complete until the end of the first week of development (when the embryo has about 200 cells). Lyonization is a random event and occurs independently in each cell. This means that the active X in any given cell may be the one that carries the Fabry mutation or the one that is normal. It is entirely a matter of chance which X chromosome remains active in any one cell. However, after one of the X chromosomes has become inactive in a cell, all of that cell's descendants have the same X chromosome inactive. The severity of disease in females is thought to correlate with the proportion of cells expressing the mutated gene.

In females, because of lyonization, some cells and tissues produce "normal" α galactosidase enzyme and others produce "mutant" α galactosidase enzyme. As lyonization is a random process, the number and types of cells and tissues that produce "normal" is different in each female. Thus, the level of clinical severity in females ranges from asymptomatic to very severe.

Unusual Suspects

1 February 2013

Barts and London School of Medicine and Dentistry

The Unusual Suspects, a day symposium on rare diseases was hosted by Barts and the London Society of Rare Diseases. This is a new Society aimed at educating future doctors about diseases that in total affect approximately 3.4 million people in the UK. 'An important part of the Society's educational programme is having expert patients/ parents give an insight into the struggle it can be to reach a rare diagnosis and how lonely it can be once you have arrived at one.' said President, Lucy McKay.

The Symposium was intended to spread the word further with talks that reminded medical students tow always 'keep rare diseases in the back of their mind' so that one day if they suspect a rare disease they can make the difference in a patient's journey to diagnosis and onward. From an MPS Society perspective we were delighted that Perry Elliott, Professor in Inherited Cardiovascular Disease at University College London was able to give his time to talk on Fabry disease.

I wish to thank the Symposium Organising Committee for a truly innovative initiative. I have no doubt that the 50 medical students attending the Symposium will when they qualify look for all the more usual diagnoses in their patients but when the obvious is not there 'think rare'.

Christine Lavery

Sharing Expertise in India

20-28 January 2013

As I alluded to in my blog/report I was privileged to be invited to work alongside Drs Ashok Vellodi, Suresh Vijay and Uma Ramaswami in MPS clinics in Mumbai, Bangalore and Chennai and I never cease to be humbled by all the MPS families I met.

Dr Mamta Muranjan and team at King Edward's Memorial Hospital Mumbai with Dr Vellodi, Tanya Collin Histed and Christine Lavery

My journey started in Mumbai on 21 January 2013 where Tanya Collin Histed, Executive Director of the Gaucher Association and I had been invited to an expert advisory board meeting in respect of Shire Pharmaceuticals Indian Humanitarian Aid Programme for Elaprase, Replagal and VPRIV.

As Tanya and I had a few hours of free time, we took a guide to show us round Dharavi slum, Asia's largest, situated in Mumbai where in an area of 1.1square kms over 1 million children and adults live. Over a period of two hours we walked through the slum which is the largest recycling plant in Mumbai and generates an income of £350 million per year. Dharavi slum provides cheap and affordable housing and is located between two main suburban rail lines convenient for those travelling out of the slum for work. The slum has a large number thriving small-scale industries that produce embroidered garments, export quality leather goods, pottery and plastic. The slum is one unending stretch of narrow dirty lanes, open sewers and cramped huts but what was striking was how clean and healthy the children appeared. What we did learn from this humbling experience was a necessary understanding of the conditions many urban and rural families live in albeit not necessarily a slum especially when we are involved in supporting LSD families whose child might qualify for ERT in the Shire Humanitarian Aid Programme.

Before leaving Mumbai on Wednesday 23 January we went to visit Dr Mamta Muranjan where she had arranged a parent and professional conference on MPS and Gaucher disease. I spoke on advocating for MPS families and the role of the patient organisation. It was then a two hour flight to Bangalore and an early night.

On Thursday 24 January Dr Suresh Vijay from Birmingham Children's Hospital ran an all day metabolic clinic at which I joined him. Amongst the patients we saw a man with Fabry disease not on treatment, a little girl with ML II and a child with Sanfilippo disease. There was a short break during the morning to attend

continued ▶

Dharavi Slum

Hindu festival to consecrate the ground at Bangalore Medical Genetics Centre for patient accommodation

Raising the Indian National flag at the Mediscan Clinic in Chennai

accommodation for the Medical Genetics Clinic. Dr Meenakshi Bhat who runs this amazing facility did her metabolic training with Dr Fiona Stewart in Belfast. On Friday morning Meenakshi had organised a one day conference on LSDs again for families and professionals. Tanya and I just had time to give our presentations on Gaucher, Fabry and MPS International Networks before taking a car with Dr Vellodi on the seven hour journey to Chennai.

Early on Saturday 26 January we were at the Mediscan Clinic for a whole day of 27 MPS patients. It was lovely to see some of the children I had met on my previous visit two years ago but there was one very special boy missing, Parth. It was with great sadness that I learnt Parth who appeared on the front cover of the Spring 2011 MPS Magazine died just two weeks previous at his home in Delhi. That evening we were treated to dinner with the Mediscan LSD group, a just celebration as it was India's National Day.

The next day, Sunday, Dr Ramaswami and I ran the Mediscan Sanfilippo clinic and saw 25 patients. It is difficult getting over to families that it is not because they live in India that their children do not get therapy for Sanfilippo and that the sadness they are suffering for their children is the same the world over as there is no licensed therapy yet for this devastating condition. A few hours after the clinic finished it was on to the airport and the long journey home.

Christine Lavery

Eurordis Black Pearl Gala Dinner

EURORDIS Black Pearl GALA DINNER 2013

26 February 2013
Hotel le Plaza - Brussels, Belgium

It was a great privilege to be supported by GSK to attend the Eurordis Black Pearl Gala Dinner in celebration of Solidarity & Hope for Rare Disease Patients Throughout Europe ahead of Rare Disease Day, 28th February 2013.

The Gala Dinner was the back drop to recognising the contributions individuals have made over many years to the rare disease space. In particular I was very pleased to see two good friends from the UK, Lesley Greene and Kay Parkinson receiving awards. Lesley Greene along with her husband Peter founded the Research Trust for Metabolic Diseases in Children, now known as CLIMB. Lesley was recognised for her dedication and outstanding contribution to the Eurordis community with the Volunteer Award. Kay Parkinson was given the Patients Organisation Award for innovative approaches in supporting patients with Alstrom disease.

This evening provided an invaluable opportunity to engage with the many players involved in rare diseases across Europe .

Christine Lavery

left to right: Vinciane Debroux-Pirard (Genzyme), Fabrizia Bignami, Christine Lavery (UK MPS Society), Valentina Bottarelli (EURORDIS)

Junior Lawyers Quiz Night raises £1000 for charity

18 January 2013

Money raised for genetic diseases charity. Three Blandy's teams rank in top four positions

Last night, the Berkshire, Buckinghamshire and Oxfordshire Junior Lawyers Division (BBO JLD) hosted a charity quiz night in Reading.

Despite the threat of snow, the quiz went ahead and was a great success, raising £1000 for the Society for Mucopolysaccharide Diseases (MPS Society) based in Amersham.

With 22 teams (approximately 120 people), the atmosphere was fun, yet VERY competitive. A team from Pitmans ran out winners, however the three teams entered by Blandy & Blandy LLP came a respectable 2nd, 3rd and 4th.

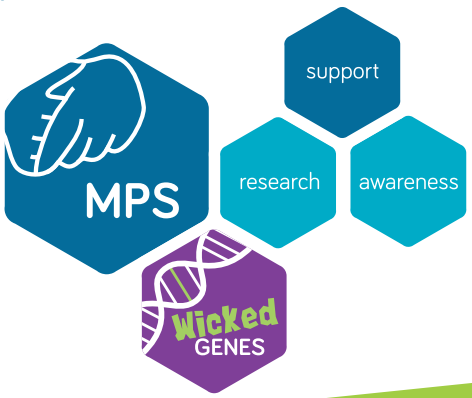
Money raised from the entrance fees and together with a raffle totalled a very respectable £1000. In attendance representing the charity was the founder and Chief Executive, Christine Lavery MBE, who commented: **"I am absolutely thrilled to see so many young lawyers come and support this event in aid of the MPS Society. I would like to say thank you to everyone for their efforts."**

As well as wearing the hat of regional JLD Sponsorship and Lecture Secretary, Blandy's family solicitor Jessica Reid is also a Trustee of the MPS Society. She commented **"There was a lot of organisation involved but it was great fun and a really positive atmosphere. The money raised will go a long way to help support individuals and their families affected by MPS and related diseases, which are progressive, limiting genetic conditions."**

Teams represented law firms from across the three counties, including Blandy & Blandy, Caversham Solicitors, Darbys, Field Seymour Parkes, Brookstreet DR, Gardner Leader, Henmans, Blake Lapthorn, Owen White, Pitmans, Boyes Turner, Shoosmiths, The Head Partnership and Vodafone Group. Barristers Chambers represented were from QEB, 1 Hare Court, Harcourt Chambers and 1 Garden Court.

The Berkshire Buckinghamshire and Oxfordshire Junior Lawyers Division is a local group of The Law Society's Junior Lawyers Division. Members include LPC students, paralegals, trainees, and junior solicitors in the three counties region. Blandy & Blandy lawyers have always taken an active role in supporting the JLD. Currently, as well as Jessica, above, trainees Jannicke Brar and Vicky Charlesson are joint Treasurers and Caroline Brady is one of four social secretaries – all of whom played a part in organising this quiz.

The Society for Mucopolysaccharide Diseases is the only registered charity in the UK providing professional support to individuals affected by MPS and related diseases. Established in 1982 by Christine Lavery MBE, its purpose is to provide support, research and awareness, working with medical professionals and scientists.



CONGRATULATIONS TO THE GAELFORCE TEAM!

Mick Kirwan, Neil Mitchell, Philly O'Carroll, Barry Hicks, Conor Swendell and Stephen & Eamonn Tighe completed Gaelforce on August 18th 2012.

The team completed Gaelforce in aid of the MPS Society. Their friend's young son Max suffers from the very rare, incurable condition known as Hunter Disease (also known as MPS II). Research is urgently needed to find a cure for this devastating and life-limiting condition.

Gaelforce West is the largest one day adventure race of its kind in the world. It is a multi-sport adventure race involving cycling, running, hiking and kayaking. The course of approximately 67km stretches from the stunning Glassilaun beach to Westport. In total they ran for 21km, cycled 45.5km, Kayak for 1km and climbed Croagh Patrick while we are at it!

The Gaelforce Team raised £2161.40 on their JustGiving page, www.justgiving.com/gaelforce

JustGiving

Happy Days Nursery is a children's nursery in Rayleigh, Essex where Archey Rawles (who is now at big school) attended. His mummy Tracey, wanted to raise money on behalf of the MPS Society and to raise awareness of Mucopolysaccharide (MPS) and related diseases that Tracey and Archey have been diagnosed with.

Last year, on Wednesday 16th May 2012 (a day that Archey attended) we had a **Wear it Blue Day** to raise money for the MPS Society. Not only did all the children and staff wear something blue and donate £1, we also had blue toys, resources and activities for the children to play with, baked cakes which were then decorated with blue icing and even had a story about a blue kangaroo. It was a fantastic day and lots of fun was had by all. **We raised £120 for the MPS Society.**

CONGRATULATIONS TO IAN EVANS who has been awarded a Gold Award from the Arriva Community Action Awards in recognition of his valuable contribution to the Society. It is wonderful that Ian's fundraising and support of the MPS Society which included Santa to drive a bus around Aylesbury at Christmas has been recognised in this way.

Thank you also Ian and Arriva for the kind donations of £1000 plus a further £85 being the proceeds from the sale of uncollected lost mobile phones plus £83.95 in loose change collected in an MPS collection box by Santa Claus.

Photo above, shows Ian receiving his award from Paul Adcock, Managing Director of Arriva the Shires and Essex. Santa Claus waiting for his next bus at Aylesbury Bus Station (below).

FUNDRAISING

CHAYSER GIG

Nick Travers raised £400 on his JustGiving page recently in memory of his friend's brother Jamie who suffered from Hunter disease. The money was raised from donations as well as the proceeds from a recent gig by his band 'Chayser' at the O2 arena in Sheffield.

PARACHUTE JUMP

Our son in law, Leigh Jones, did a sponsored parachute jump during October 2012 in memory of our daughter, Katie Smithers, who passed away on 17th October 2005 from MPS I aged 21 years. **Leigh raised a total of £500.** Thank you to staff at Remploy Port Talbot and Sandfields Primary School, Port Talbot and all family and friends who sponsored the jump.

Peter and Maureen Smithers

IN MEMORY

In Memory of Kieran Hughes

(4 June 1995 - February 2007)

On Saturday 6 October 2012, friends and family of Kieran Hughes, climbed the Cobbler in Arrochar in Kieran's memory. This was no mean feat with the Cobbler being 920 metres high.

The five intrepid travellers (Sarah McNeill (Aunt), Alison & John Tarkenter (family friends) and Angela & Gillian McPate – friends of Alison) set off from Dunoon at 9.30am. They reached the base of the Cobbler at approx 10.30am. It was a beautiful sunny day for their journey and resplendent in their specially designed tee shirts, with Keiran's smiling face, (courtesy of Stephen Ballantyne), they started the climb.

About three quarters of the way to the summit, they stopped for lunch and enjoyed the amazing scenery all around. The last part of the climb was difficult but they are sure that Keiran was with them all the way and gave a few pushes when needed. They reached the top at approx 1.30pm.

The climb down was not much easier and it was a sore but happy group who reached the base about two hours later.

Sarah, Alison, John, Angela & Gillian would like to thank, most sincerely, all those who sponsored their walk.

They raised the grand sum of £706.00 (added to this will be 25% Gift Aid).

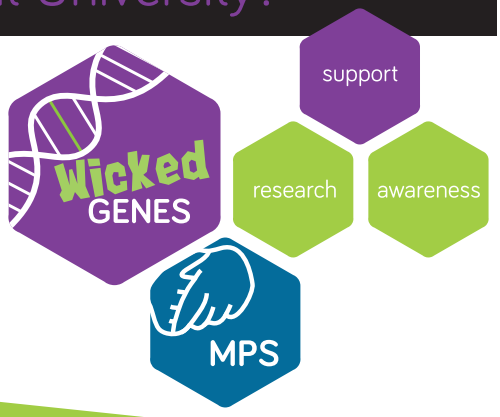
Raising and Giving at University

Calling all students past and present; can you help us get Wicked at University?

Most UK universities have amazing RAG Committees (Raising and Giving) and through various activities, ranging from the fairly standard to the utterly crazy, UK universities are responsible for raising millions of pounds for UK charities. (It is estimated that these student-run university organisations raised between £6m and £9m for charity in 2011).

Wicked Genes have been busy contacting universities to raise awareness of Mucopolysaccharide and related diseases and to put the MPS Society forward for RAG partnership consideration.

Wicked Genes would particularly like to thank the RAG team at the **Coleraine Campus of the University of Ulster**, who recently selected the MPS Society to be one of their three charity partners. Coleraine launched their RAG week at the end of February with all manner of sponsored events; sponsored waxes, bike rides, parties. Apparently, a particular highlight was a RAG Week blind date evening!



If you have any links to universities then please let their RAG teams, (normally based at the Student Union), know about the MPS Society. We would be delighted to support any student fundraising.

Special thanks to . . .

Raymond Webber
donated £70 towards research into Morquio.

Shabana Kausar
donated £20 in memory of her son Shujah.

Paul Bridges
donated £40 in memory of a friend's dad.

Steve Ford
donated £10 after seeing an article in the Metro newspaper.

Robert Kennedy
donated £225 as a family request for son's funeral.

Friends and family of Tillie Mae Mawdsley
have donated £50 to the Genistein trial.

Jenna Pollard
donated £185 in honour of Merlin Penny-Smith.

Ellen Graham
donated £25 to MPS in lieu of sending out Christmas cards.

The Hampden Arms pub
held their annual Boxing Day quiz raising funds for the MPS Society. They raised £170.

ProCo
held a fundraising day before Christmas and raised £110.

Sally Amos
sent in donations for £60.

Ian passed away on January 14th 2013 aged 23. At the suggestion of his father Chris, **Colin Gorman** made a donation in Ian's name.

Samantha Schefele
raised £110 being the proceeds of a bake sale for the Genistein Trial.

Mrs J Eve
kindly sent in a donation of £30 collected by the over 60s club of which Mr Walter Wheeler was a member. He sadly passed away on 14th December but his wish was that any money collected was to go to the MPS Society as he had a granddaughter who died of this disease.

Whartons Primary School
recently donated £260 being the proceeds of donations and a collection at their Christmas performance.

St Thomas More Catholic Primary School
held a collection for the MPS Society following their Christmas nativity play in December raising £230 for MPS.

Michala Mawdsley
sent in £50 for the Genistein trial donated by Clancy Docwra.

Ben Conlin and friends
at St Patrick's R.C. Primary School raised £383.50 for the MPS Society.

The Busy Bee MCC
held a number of fundraising events throughout the year and donated £400 to the MPS Society in memory of Lewis Browning, the son of one of their members.

Norton Rose LLP
have donated £2365 towards the Genistein trial for Sanfilippo. This was raised through various fundraising activities over Christmas.

Marina Foster
has raised a further £4883.55 from the sale of second hand items at her shop, Marina and Friends, in Bristol. This takes the cumulative total to £96,701.35.

Margaret and Graham Moore,
Grandparents to Samantha Brockie, have raised £150 being the proceeds of a pre-Christmas card party, raffle and donations.

David Cook's employer, UKAR,
donated £1000 to the MPS Society. This was match funded following David's fundraising charity football match towards the Genistein trial.

Many thanks to **the London School of Economics** for inviting the Wicked Genes team to host a stand within the university campus to raise awareness of the MPS Society and Wicked Genes. The LSE RAG team were very welcoming and it was great to find out more about student fundraising and learn about how we can get involved in the future.

The Rotary Club of Yeovil
have kindly donated £250 to the MPS Society. Christine Lavery, our Chief Executive, gave a very informative talk to the club to raise the profile of MPS and related diseases and the work of the MPS Society.

Many thanks to **Tom Gill and colleagues at 24/7 Media** for their kind donation of £1200 towards the Genistein clinical trial. Tom's son Bobby has Sanfilippo disease. His family have been very active in fundraising for the Genistein campaign.

Ann Parsons
has donated £101 being the proceeds from the sale of trolley key rings at Asda Eastbourne

The MPS Society received £208.92 in donations in memory of the late Robert Lunnon.

Emily Cannie
from Greenock, Scotland bought the contents, made up and held a raffle for Christmas hamper recently. She is related to the Ingram family. She had a fantastic response and we are all very proud of her for raising £200 for the MPS Society.
Russell Ingram

Marilyn Eggleton
kindly donated £20 in memory of her daughter Kim who suffered from MPS III Sanfilippo.

Michelle Brooker
donated £55 to go with Lily's ShareAGift page.

Hayley Vallance
took part in the Cardiff Half Marathon in October 2012 raising £311 in sponsorship money.

Dorothy Robinson
(nanny of Hannah Shannon MPS III) held a Third Craft Workshop with the proceeds of £170 being donated to the MPS Society. They had a wonderful day with homemade lunch for the break.

Michella Perry
took part in the Great North Run in September 2012 raising £155 for the MPS Society in sponsorship monies.

Louise Hiller
from Torus Insurance held a cake sale raising £58.57 for the MPS Society.

Mrs Whittaker
has kindly donated £20 in memory of Mrs Phyllis Mary Sanderson who passed away on 5 October 2012.

Lakeside School
in Herts who raised £200 towards the Genistein Trial with their Christmas activities, including carols round the tree.

Donations
K Spark • Emma Morrice • Emma Ashton • Mrs Baker • Diane Pierson • Mr J Scott • Mrs M Davison • Mr Denis Stevens • Isabelle • Karen Berry • Mr and Mrs W Pearson • Lionel and Janet Croft • Mrs Baker • Petra Koehne

In Memory
Lewis Browning • James Forsyth Nicholson • Anabelle Rose Shepherd • Christopher Croft

Collection Boxes
Victoria Atkinson

Stamps, foreign coins, mobile phones, ink cartridges; jewellery
Arriva The Shires • Molly Griggs • Sarah Swayne • Sue Lowry • Wilma Robins • Members of St George's Church, Brockworth

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

The importance of Social Media for the MPS Society

You have hopefully noticed we have been far more prolific in our updates both on Facebook and Twitter and early signs are that this is definitely a step in the right direction with activity on our MPS Society Facebook page up nearly 700% this week. We have a passionate and committed MPS community and our Social Network sites enable us to build on this.

We would like to continue to grow our audience to raise awareness of Mucopolysaccharide and related diseases and the work of the Society. Our Facebook and Twitter presence enable us to share MPS information, news and updates with you as soon as we get them. If you aren't following us on Twitter and Facebook yet . . . please do so and encourage everyone you know to do the same.

Our pages enable us to engage with you, our supporters in a way we have not been able to do before and hopefully give you the tools to help spread the word about the charity and the work it does and drive traffic to our website. It is also a different way of communicating with you in a way which might be easier and more convenient for some of you. Social Media is an open and honest way of communicating and we hope with your 'likes' 'shares' and 're-tweets' we can continue to grow and engage.

facebook.com/mpssociety

twitter

twitter.com/MPSSocietyUK

Other ways to support MPS Society:

Leaving a Legacy

What is a legacy?

A legacy is defined as an 'amount of property or money left to someone in a Will'. For many years not-for-profit organisations have benefited enormously from these charitable bequests. In fact, the single biggest source of unrestricted gifts to charities in the UK actually comes through Legacies.

Who can leave one?

Anyone - It's a common myth that only the rich and famous leave money to charity when they die. This couldn't be further from the truth. The reality is without the gifts left in Wills by people like you, many of the charities we know and support today wouldn't even exist. Did you know that 74% of the UK population support charities and when asked, 35% of people say they'd happily leave a gift in their Will once family and friends had been provided for.
The problem is only 7% actually do.

Despite this relatively small number of people leaving a charitable bequest in their Will, legacy gifts are worth almost £2 billion to the not-for profit sector. Legacy gifts provide around 12 per cent of all income for the largest fundraising charities.

How to leave a legacy?

Making a Will or making a change to an existing Will need not be complicated. If you already have a Will that you are happy with, but just wanted to add a gift to the MPS Society ask your solicitor to draw up a codicil – a simple legal addition to an existing Will. If you don't yet have a Will you can use a Solicitor, Will Maker or even use a Do it Yourself pack available from shops and on line. Leaving a Will is the only way to ensure our families, friends and any charities and good causes we support will receive what we want to leave them after our death.

Why leave a legacy to the MPS Society?

It is vital that the MPS Society has sufficient funding to be able to look forward to the future with confidence. One way in which you can support the Society in achieving its long term objectives is to include the Society when drawing up your Will. Such generosity will enable us to continue supporting those affected by MPS and related diseases, and honour commitments to research programmes and a multitude of projects designed to improve the quality of life of individuals with these rare, genetic, life limiting diseases. Legacies can make an enormous difference to the service we provide.

If you chose to remember the MPS Society in this way, you do not have to inform us of your decision if you prefer not to. However, if you do inform us of your decision, this information will be treated with the most upmost confidentiality.

Help us care for today and give hope for tomorrow.

If you would like more information about leaving a legacy please email **mps@mpssociety.org.uk**

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

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

www.mpssociety.org.uk

Hold the back page... stop the press
We have an announcement!

There are nearly 170,000 charities in the UK, some with some very immediately recognisable brands. While we recognise that MPS and related diseases are rare, we want to raise the profile of these devastating conditions across the UK and reach out to more families affected by them.

If you visit the MPS website at **www.mpssociety.org.uk** you'll see not only are we developing a new look but we have made our website far more accessible and easy to navigate so you can get the support you need quickly and with ease. Whether it's the latest information and support from Advocacy on the benefit changes, or research and treatment updates, the new site should make your search much easier.

We've also created a new fundraising pack which is packed with information about the MPS Society and great ideas for fundraising for us whether you are at school, work or in a local community. We wanted a pack that can shout 'we may be small but we mean business' when it lands on someone's desk. We understand that it's likely that potential new supporters haven't heard of Mucopolysaccharide Diseases before, our new pack ensures this doesn't matter. In a short booklet we have included all you need to know to inspire people to raise awareness of MPS and related diseases and help raise vital funds.

You can download a copy of the new fundraising pack or email us and we can send you one. We'd love to know what you think of it all too, so please email us at **wickedgenes@mpssociety.org.uk**

p.s
It doesn't stop there... we've also created a guide to holding a **Wicked Walkabout**! A one stop shop on how to hold a brilliant sponsored walk/run or cycle. Drop us a line if you'd like us to send you one.

p.p.s
If you or anybody you know wants to hold a **Wear It Blue** event to mark MPS Awareness Day on 15th May, email us for posters and stickers

wickedgenes@mpssociety.org.uk

Requesting more information by post

If you would like to receive more information by post, please tick the appropriate boxes, complete your contact details and return the form to us in the post.

- ☐ Fundraising Pack
- ☐ Wicked Walkabout Organiser's Guide
- ☐ Wear It Blue for MPS Awareness Day, 15th May
- ☐ Publication Order Form
- ☐ Merchandise Order Form
- ☐ Ink cartridge/jewellery re-cycling Envelopes
- ☐ Cardboard Collection Box
- ☐ Gift Aid Form
- ☐ How to make a regular donation
- ☐ Leaving a Legacy leaflet
- ☐ Payroll Giving
- ☐ Information on becoming an MPS Childcare Volunteer

Name: .....

Address: .....

.....

.....Postcode: .....

Email: ..... Tel. No. .....

“It was really hard in the last year when he stopped talking. Our greatest fear is having to watch the Tommy we know and love gradually disappearing from us. I would just love to be able to listen to him say mummy and daddy again”

Tommy’s Mum

Mucopolysaccharide (MPS) and related diseases are rare, genetic conditions causing progressive physical and in many cases neurological deterioration. These devastating diseases can result in death in childhood.

But there is hope.

Over the past 30 years the MPS Society has been involved in funding vital research in developing new treatments and supporting families like Tommy’s.

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

Text MPSS01 £5 to 70070

MPS

support

research

awareness

Wicked GENES

The Society for Mucopolysaccharide Diseases is the only UK charity providing professional support to those affected by 24 MPS and related diseases, funding research and raising awareness of these rare genetic diseases.

www.mpssociety.org.uk