MPS

Society for Mucopolysaccharide Diseases

Spring 2010

What's Inside...

Treatment and Clinical Trial update plus stories from our members

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 over 1200 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

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

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

MPS & 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, severe degenerative mental deterioration resulting in death in childhood.

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

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 to www.mpssociety.co.uk, phone 0845 389 9901 or post your donation to our office, MPS House.

Front cover photo:
Sibling Week group, April 2010

Society for Mucopolysaccharide Diseases

MPS House, Repton Place, White Lion Road, Amersham
Bucks, HP7 9LP, **www.mpssociety.co.uk**
T: 0845 389 9901, Out of Hours: 07712 653258
F: 0845 389 9902, E: mps@mpssociety.co.uk
Registered Charity No. 287034

Staff

Christine Lavery
Chief Executive
c.lavery@mpssociety.co.uk

Antonia Anderson
Communications Officer
a.anderson@mpssociety.co.uk

Kate Barker
Grants and Corporate Fundraising Officer
k.barker@mpssociety.co.uk

Sue Cotterell
Office Manager/PA to CEO
s.cotterell@mpssociety.co.uk

Fiona Hopson
Office Administrator
f.hopson@mpssociety.co.uk

Gina Page
Finance Officer
g.page@mpssociety.co.uk

Sophie Thomas
Senior Advocacy Support Officer
s.thomas@mpssociety.co.uk

Jolanta Turz
Advocacy Support Officer
j.turz@mpssociety.co.uk

Linda Warner
Advocacy Support Officer
l.warner@mpssociety.co.uk

Kirsty Wyatt
Special Projects and Communications Assistant
k.wyatt@mpssociety.co.uk

Magazine Deadlines

Summer	1 Jun 2010	Autumn	1 Sep 2010
Winter	1 Dec 2010	Spring	1 Mar 2011

Friend of MPS

Become a Friend of MPS to receive the Society's magazine and fundraising newsletter plus a range of other benefits. Contact us for more information.

IN THIS ISSUE...

Spring 2010

Our cover shot for this edition of the MPS Magazine is of MPS siblings on their trip to France during the Sibling Adventure Week. More photos and stories from this will be featured in our Summer magazine.

CHIEF EXECUTIVE'S REPORT

With Easter behind us, and I hope better weather on the way, we look forward to seeing as many of you as possible at the events we have lined up for you over the next six months.

By the time you read this, siblings of MPS brothers and sisters will have returned from their 'sibling week' at PGL Windmill Hill in East Sussex. I am sure the highlight of the week will have been the day trip to France, a visit to a chocolate factory and spending their Euros. Like you, I look forward to reading the childrens' stories in the next MPS magazine.

I am sure the MPS Young Adult Weekend to Blackpool at the end of April will also generate its fairshare of stories. Here at MPS House the challenge has been finding appropriate evening entertainment for the group! So let us see what the young people make of the weekend.

In 2010 it is the turn of Northern Ireland and Scotland to host regional conferences. This year it is with great pleasure that we have joined forces with the Irish MPS Society to organise All Ireland MPS and Fabry Conferences to be held simultaneously on 14 – 16 May at the Hilton Templepatrick. Whilst these conferences are planned with Irish families in mind, they are not exclusive to our Northern Irish members. If you live outside the Province but are interested in attending the MPS or Fabry Conference please do contact the MPS office for a booking form.

The Scottish meeting at the Edinburgh Airport Hilton Hotel, 23 - 25 September 2010, is for individuals and families affected by Lysosomal Storage Diseases (LSDs). The programme has something for everyone and we hope you will gain much from meeting with others affected in similar ways.

Following the success of our first disease specific 'Expert Meeting' for Morquio in 2008 we look forward this year to a full house at the 'Expert Meeting for Sanfilippo Disease' at the Hilton Northampton, 27 – 28 August. We are bringing together the world's experts not only on clinical management of MPS III but those who are working tirelessly to find long overdue therapeutic breakthroughs. There will be a full childcare and vulnerable adult programme to enable parents to attend all sessions of the conference and relax over a gala dinner on the Friday night. We also extended an invitation to this meeting to all the International MPS Societies to enable our members to have the opportunity to benefit from a global perspective.

Finally, there is the MPS and Fabry Family Weekend at Camelot, near Preston in Lancashire, 29-31 May. This is a unique opportunity to have a fun weekend away at a highly subsidised cost with no conference involved. On the Sunday evening the gala dinner, for which childcare will be provided, will be preceded by the Society's Annual General Meeting. Camelot is a land of great knights and amazing days, breathtaking rides, magical sorcery and shows for all ages, so I do hope as many of you as possible can magic yourselves there!

Christine Lavery
Chief Executive

MPS Annual Report and Accounts
The MPS Board of Trustees have decided that to save on costs, the MPS Society will no longer be producing a printed annual report and accounts distributed free to our members. The full report will instead be available to download from our website, www.mpssociety.co.uk, by 1st June 2010. If you require a hard copy, please request this by emailing accounts@mpssociety.co.uk. Please note, for this we will charge a fee of £5 to cover costs.

Highlights from the Management Committee

The Society's Board of Trustees meet regularly. Here is a summary of the key issues that were discussed and agreed at the Management Committee Meetings held on 5 December 2009 and 5 - 6 February 2010.

Personnel
The Chief Executive advised the Board that all staff appraisals have been completed. The Board were also informed that Fiona Hopson and Kirsty Wyatt have joined the team as Office Administrator and Special Projects and Communications Assistant respectively. The post of Advocacy Officer is currently being advertised.

Financial Management
Trustees agreed a number of changes to the MPS draft accounts for year ending 31 October 2009 required under SORP regulations. Trustees also agreed a programme of capital expenditure to replace the Society's oldest computers that are increasingly slow and unreliable. The Board approved the draft accounts at their meeting on 6 February.

Generating Income
Trustees were advised that the Society is now registered as a Charity in Scotland and is now in a better position to secure funding where previously excluded.

Risk Management
Trustees reviewed the Risk Register and considered and agreed a small number of changes to reflect advice received by Peninsula.

Policies
At their December 2009 Management Committee Meeting Trustees reviewed and approved the following policies: No. 22 Financial Controls; No. 23 Reserves Policy; No. 26 Financial Assistance; No. 28 Moving and Handling; No. 35 Gifts and Hospitality; No. 41 Regional Events; No. 53 Office Keyholders; No. 62 Data and Information Processing.

At their February 2010 Management Committee Meeting Trustees reviewed and approved the following policies: No. 51 Board Diversity; No. 52 Server Back-up; No. 61 Investment; No. 63 Anti Money Laundering

Clinical Management and Treatment
Trustees were updated on the current status of the Fabrazyme shortage for Fabry disease as well as the challenges of recruiting to the MPS IIIA Natural History Trial. BioMarin have reported positive results from their Phase I/II Clinical trial for MPS IVA.

Jeans for Genes (J4G)
Considerable discussion took place regarding governance concerns and cost income ratios related to the Jeans for Genes Charity. Barry Wilson, Chairman, advised Trustees that he had received an email from Ann Green, Chairman of J4G, stating that plans are in place to reduce the 70% cost income ratio. The MPS Trustees noted that a programme of change is underway. Trustees considered and rejected the proposal by Great Ormond Street Children's Charity (GOSHCC) to take over the management of the J4G Appeal for 2010 requiring the J4G Charity to be closed. The MPS Trustees agreed that the four partner charities would be best served if the J4G Chief Executive and her team continue to plan for the 2010 J4G Campaign. Following the resignation of the MPS nominated independent J4G Trustee in 2009, the MPS Board approved the proposal that Christopher Syrda be put forward as the MPS nominated independent J4G Trustee.

MPS Research Grants
Professor Bryan Winchester gave an overview of the projects supported and grants awarded since 1982. In proposing a research strategy to form part of the MPS Strategic Plan 2010 – 2015, Prof. Winchester suggested that the Society should look at broadening its sources of income to fund research. He also recommended that when the Society is in a position to, it also considers smaller pump priming research projects as well as major programme grants. It was agreed to use this document as the basis of information provided on the MPS Website.

Following a donation of £165,000, at their December 2009 Management Committee Meeting Trustees agreed to fund the first year of a five year research project to establish a clinical trial for Genistein at the University of Manchester. The Trustees also agreed two small grants of £6,000 and £7,000 respectively for its MPS III programme grant also at the University of Manchester.

International Collaboration
The Chief Executive gave details of the WORLD Lysosomal Storage Disease (LSD) Conference she had recently attended. The MPS Society had an abstract accepted reporting the results of 'A Pilot Assessment of Four Questionnaires for Assessing Functional Status and Quality of Life in Mucopolysaccharidosis Type I'. It was agreed that Sophie Thomas and Linda Warner will attend the LSD Roundtable meeting in Prague in April. The Trustees offered their congratulations to the five young 'MPS Ambassadors' aged 18 –24 years, two with MPS IVA, one with Mannosidosis, one having had a BMT for MPS I and one a sibling of a sister with MPS III who have been chosen to participate in the International Symposium for Mucopolysaccharide Diseases in Adelaide, Australia in June 2010. These Ambassador roles have been made possible with a restricted grant from the Shauna Gosling Trust.

NEW FACES AT THE MPS OFFICE

Fiona Hopson

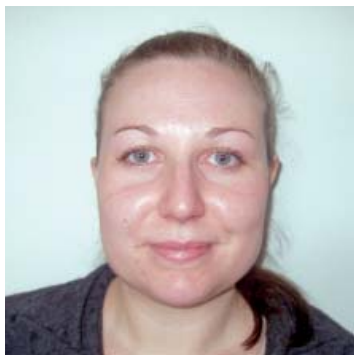
My name is Fiona Hopson. I'm 25 years old, and I joined the MPS Society at the beginning of February 2010 as the Society's full-time Office Administrator.

My background is in the military where I served 4 years in the Royal Air Force working with Aerospace Systems. I worked in NATO and in many underground bunkers across the UK. I left the RAF in 2007 to have my son Daniel. Previously in college I worked with children with disabilities, organising sporting events for the children.

My pastimes now are running around after my 2 year old son and my husband, but in the spare time I do get I enjoy seeing friends, hosting dinner parties, travelling, doing home improvements, and sleeping… all the "grown-up" things you're supposed to do as a mum and wife!

I find working for the Society very rewarding, yet also emotionally consuming. It makes me appreciate every moment with my son and to never take anything for granted. I feel honoured to be able to make a difference in any way I can for the MPS Society and its members.
Fiona Hopson f.hopson@mpssociety.co.uk

Kirsty Wyatt



My name is Kirsty Wyatt and I joined the MPS Society in February 2010 as Special Projects and Communications Assistant.

My background is within TV Production, working within a live TV environment as well as on location shoots.

In and around university and my working life over the past ten years, I have been heavily involved with the Pepper Foundation, a local charity that provides on call nursing care to children within the Chilterns Area who suffer from terminal and life limiting illnesses. Other than my contributions within Events Publicity I am also involved with the Pepper Summer Show where I have the opportunity of fulfilling my 'performing ambitions' on stage. I have also produced a mini documentary for this charity which has become a useful publicity tool.

In my short time here I have already been inspired by so many stories from both individuals and family members that are dealing with MPS and Related Diseases and hope that my experience can help promote the Society and the essential support that it provides its members.
Kirsty Wyatt k.wyatt@mpssociety.co.uk

MPS Annual General Meeting 2010

The 2010 Annual General Meeting of the Society for Mucopolysaccharide Diseases will be held at the Park Hall Hotel, Chorley, Lancashire at 6.30pm on Sunday 30 May 2010.

If you are interested in becoming a Trustee of the MPS Society please contact the MPS office.
We would particularly like to hear from any MPS Society members living in Northern Ireland as well as other parts of the United Kingdom.

Society for Mucopolysaccharide Diseases
MPS House, Repton Place, White Lion Road, Amersham, Bucks, HP7 9LP
Tel: 0845 389 9901, Fax: 0845 389 9902
www.mpssociety.co.uk, email: mps@mpssociety.co.uk

WHAT'S ON 2010!

MPS CLINICS

Friday 14 May	Northern Ireland Clinic
June TBC	MPS I BMT Teenage / Transition clinic
Wednesday 2 June	Bristol clinic
Friday 9 July	Birmingham clinic
Friday 16 July	MPS I BMT under 6 years
Friday 23 July	MPS I BMT over 6 years
Friday 15 October	MPS I BMT under 6 years
Friday 22 October	MPS I BMT over 6 years
Friday 26 November	Birmingham clinic
November TBC	Northern Ireland clinic
December TBC	Cardiff clinic

CONFERENCE EVENTS

14 – 16 May	All Ireland MPS and Fabry Conference
29 – 31 May	MPS Family Weekend, Camelot Lancashire
20-27 June	International MPS Conference, Adelaide
27 – 28 August	MPS III Expert Meeting, Northampton
12 September	Birmingham Family Day, Cadbury World
23 - 25 Sept	Lysomal Storage Disease Study Meeting
16 October	London Family Event

Book Now!

Expert Meeting on Sanfilippo Disease, MPS III

27 - 28 August 2010

Northampton Hilton

The MPS III Expert Meeting programme is available as a download from the MPS Society's website **www.mpssociety.co.uk**

A programme and booking form have already been sent to member families with MPS III children and adults but if you are a bereaved MPS III family and would like to attend, please contact the MPS office

Mark your calendars and hop down for the
11th International Symposium on Mucopolysaccharide and Related Diseases
Adelaide, South Australia, 23 – 27 June 2010
www.mps2010.com.au

Mucopolysaccharide and Related Diseases Society Aust. Ltd., Lysosomal Diseases Australia and Lysosomal Diseases New Zealand warmly invite you to join them in Adelaide,
23 – 27 June 2010 for the 11th International Symposium on MPS and Related Diseases.

The scientific and family programmes will be exciting and relevant with a focus on the areas of newborn screening, prognostics, understanding pathology and therapeutic options. Genuine opportunities for thorough discussion and debate will be a feature of the program.

Adelaide is a city surrounded by parklands, sports fields, a top class golf course, walking and cycling tracks and beautiful gardens.

We hope you will hop on down under and join us for five exciting days of cutting edge science, exciting family experiences and an enjoyable cultural experience.

Members' Announcements

New Members

Mr and Mrs Sainsbury have recently been in contact with the Society. William has a diagnosis of MPS I Hurler-Scheie disease. William is 2 years old. The family live in the South East.

Mrs Hanson has recently been in contact with the Society. Mrs Hanson and her two sons have a diagnosis of Fabry disease. The family live in the South West.

Mr and Mrs Brentnall have recently been in contact with the Society. Jacob has a diagnosis of Sanfilippo disease and the family live in the East Midlands.

Mr and Mrs Attride have recently been in contact with the Society. Their sons Kallum and Stefan have recently been diagnosed with Mucolipidosis Type III. The family live in the Midlands.

Births

We would like to congratulate Tim and Jessica Hooper on the birth of Martha Rose Hooper on 31 January 2010 weighing 8lbs 8oz. Martha is a new sister for Jamie who has MPS III, Sanfilippo disease.

Deaths

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

Joseph McDonagh who suffered from ML II and died on 30 December 2009 aged 6 months.

Helen Swiderska who suffered from Sanfilippo Disease and died on 15 February 2010 aged 30 years.

Catherine (Kate) Scott who suffered from Sanfilippo Disease and who died on 8 March 2010 aged 37 years.

Do you have a story to share?
Please email
newsletter@mpssociety.co.uk
or phone **0845 389 9901**

In Memory

Daniel James Anthony
Our Star

When God gave us our son Daniel,
He gave the brightest star from the sky.
He said Daniel will be so very different from others,
But he did not give us reasons why.
Take this star where ever you travel,
For so very many people Daniel will meet,
Because this star is so very, very special,
He will light up every dark cornered street.

His time on this planet has been very special,
The wonderful things he has done,
All the places he's been and the people he's met,
Made possible because of the love,
From his friends, Dad, Sister and Mum.

It's time for this star to go back
Now to be with the stars up above,
While on earth he has known nothing
But happiness and totally unconditional love
It's time for this star to go back now,
Please only be sad for a while
Remember he left us all something forever,
His humour, his laughter, his mischief
and his beautiful smile.

Daniel Ellis

25 August 1991 - 20 December 2009

The Sanfilippo Code of Conduct

By Penny Lister

Thou must never, under any circumstances, allow parent or carer to entice thou into a daytime nap. Should the disgraceful situation ever occur the offending parent/carer must be punished by at least 24 hours of sleep deprivation.

Thou must be constantly on the lookout for open doors and gates; these are invitations to forbidden pleasures and adventures.

Thou must always leap into active mode when thou hears the first tweet of a bird at sunrise. The only exception to this rule is on the first day of each school term, when a sleep-in until at least 9am is recommended.

When travelling in a car thou must always buck violently when stopped at traffic lights. This prevents the driver from becoming bored and dozing off to sleep.

There is only one correct way to empty the bath of its water and it is not by pulling out the plug.

To avoid the indignity of being washed with soap, choose one of two alternatives: a) eat it, b) hide it in the toilet bowl.

Thou must taste-test the environment at every given opportunity. Even the stringiest leaf, grittiest mouthful of dirt, crunchiest rock and mushiest pet food will taste better than the content of thy lunchbox.

Thou art quite entitled to swipe food and drinks from others if they are within thy arm's reach.

As a matter of self-preservation thou must save thou most charming and disarming smiles for the hours between midnight and 4am.

Thou must always remember that buttons, knobs, switches and taps are there for a reason – do not allow them to stay idle.

Singing at the top of thy voice, decorating the floor with magazines, and pruning the plants are highly recommended activities for Doctor's waiting rooms – thou will usually not have to wait long for attention.

Within five minutes of arrival at parks thou must fill thy pants. Ditto, regarding arrival at swimming pools.

Editor's Note: This article was found in the Australian MPS Newsletter in September 1997. We thought that parents of Sanfilippo children would relate to this code...

The MPS Magazine has changed many times over the years as the Society has developed and expanded. We thought you might like to read what we wrote back in our 1986 Annual Report...

The Newsletter

One of the principal objectives of the Society is to act as a parent support group. The quarterly Newsletter continues to provide a link between members, both individuals and families and the events taking place across the country: research generally, activities and fundraising by the Society, social and other events organised for families, personal news and social chit chat. As such, its value cannot be understated; for many families it is perhaps the single most important aspect of the Society's work and it continues to attract a high priority.

The last Annual Report (1985) noted a change in emphasis in the content of the Newsletter towards an exchange of experiences, solutions to problems, helpful advice and an insight to happy events around the country and, latterly, in other parts of the world. The evolution of the newsletter continued in the subsequent 12 months and its circulation, nationally and internationally, to professionals, institutions and families alike, has increased dramatically, with Israel, USA, South Africa, Austria, Germany, Canada, New Zealand, Australia, Italy and Japan now on our mailing lists.

Whilst the Newsletter continues to be produced by a member family in a domestic environment, 1986 saw the first tentative embrace of new technology by the production of the Newsletter on a 'home' computer linked to the printer purchased by the Society in 1985 with the aid of a grant from the Department of Health and Social Security. The use of a word processor has assisted in the production of the Newsletter and it is not difficult to envisage the application of other innovations in the near future.

It is inevitable that the Newsletter will include those moments of great sadness that are common to and shared by all who have been touched by MPS. However, its principal purpose is to provide a vehicle for optimism, to emphasise the positive aspects of life and to support the very special qualities of those to whom the Society is dedicated. The volume and regularity of contributions, the members' keen anticipation of its quarterly publication, suggests that whatever improvements are required, the Newsletter continues to fulfil its primary objective.

Mark's Story

Mark Robert Fitzgerald was born on 15 October 1977 at the University College Hospital London. He was a happy child, though he suffered a lot with ear, nose and throat problems.

When he was about 6 years old his Consultant, Professor Strang, was pleased to tell us that a bio-chemist had discovered that Mark had a gene deficiency and suffered from Hunter Syndrome. We asked about a cure for this illness and were introduced to a specialist at Great Ormond St Hospital, Dr Rosemary Stevens. She was a lovely lady and a very caring doctor and helped us understand about Hunter's, and provided help and advice about coping with this illness.

In 1981 we had another son Michael. He also had the same problem as his brother, Hunter Syndrome. Mark started nursery school and enjoyed his time there, then progressed on to big school as we called it aged 5 years. Mark made friends easily and played football as often as he could. He represented his school, St Dominic's at footie and was proud to do so, but struggled to keep up with his pals in the later years as they had out grown and towered above him. Mark had become a permanent fixture on the subs bench and he was happy to be involved, but hated not playing.

Mark went to secondary school aged 11 years and half way through his first year we moved to Bedfordshire. Both boys were apprehensive about starting new schools and making new friends but they did so easily. As we were buying our own home they liked having their own rooms.

We were by now involved with the MPS Society and were grateful for the help and advice we received. We learned a lot from the staff there on all sorts of topics that confronted Mark with his Hunter's. As Mark was getting towards ages 15 and 16 the illness was getting more severe. He was unable to walk far, needing to be helped more and more, and his breathing and chest was becoming more of a problem. Mark was becoming depressed and frustrated in his own body.

When Mark was seventeen he passed his driving test first time and this gave him more confidence and he could drive himself to Sixth Form College and visit his friends. Mark was slowly putting on weight and unable to walk far without resting, and catching his breath. Mark had lived a fairly normal life up till the age of 19. Mark finished college but was unable to work as his health was deteriorating rapidly and he made a life for himself at home. About this time while out in his car, somebody crashed into the rear of him as he was stationary at a pedestrian crossing. This, combined with the fact that Mark had lost the sight in one eye and had had a shunt fitted, frightened him and he never drove again.

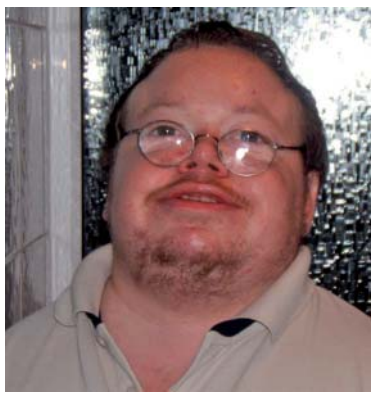
Mark soon developed Obsessive Compulsive Disorder (O.C.D.) and began to wash his hands over and over again, sometimes lasting four hours at a time, every time he visited the bathroom. His hands and arms were red raw most of the time; we had taken him to see a psychiatrist and therapists many times, but to no avail.

Mark had become nocturnal, up at night until five in the morning and asleep most of the day. He loved his TV programmes, his music, and surfing the net for information about sport, especially about his beloved Arsenal football club and was a great fan of Sir Arsene Wenger, as he called him. He was obsessive about his TV programmes, taping series after series of Australian and American programmes, such as Blue Healers, Friends, Frasier, Monk, The Mentalist and also loved his cockney characters like Minder, Only Fools and Horses and Lovejoy to name but a few. He loved his music, downloading and copying to CD, he had shelves full of videos, DVDs and CDs.

Mark loved his extended family and always loved family gatherings and enjoyed a beer or two. Mark never really enjoyed traveling abroad, although we had taken him on holidays to Malta, Spain, Greece, and the USA a couple of times and in the last five to six years we left the boys at home while we holidayed, to fend for themselves. They coped brilliantly as long as we filled the fridge and freezer up before we left. His brother fetched fresh milk and the odd beer when needed.

Mark was very grateful to get on the trial for the new drug; although we guessed he was on the placebo at first, he eventually got the drug. It helped him enormously, but in the end the Hunter syndrome got him. He was going through a good time in his life; he was much happier in himself and had started to lose some weight. He felt he was conquering his O.C.D. habit. Mark always had a problem chewing his food, so he always just ate simple food so that he didn't need to chew for long.

Mark seemed much happier with life and had become much closer to his brother, always laughing and joking together, or arguing with me - his dad about footie. He always finished the banter with me, telling me "Not to embarrass myself". He always called his mum "Sugar," his brother was "Micky Dicky" and me, I was the "Saddo Spurs Supporter."



Mark Robert Fitzgerald - MY BROTHER

I have so many memories of Mark, each from different eras and stages from our lives. I'll start with the earliest.

Mark was three and half years older than me and my big brother, he would always know about everything before me and understand it, he would always get the better opportunities than me because he was older and it was natural. From my early memories of being aged 5-9 years, before we moved out of London, Mark was my idol whether it be for football or his friends or if he would go to the highest rung on the climbing frame. He would have the latest stereo system - a ghetto blaster or an Amstrad CP464, and understand exactly how it worked and how to play the games. He would record things on the VHS player while I had no idea how to, he would have friends and gangs in the playground and I was always the annoying younger brother. Mark was nice to me always, yes we fell out and he'd make me cry, but ten minutes later I would be in awe again.

My dad took Mark to see Rocky 4 at the cinema and it was a late showing and I was so jealous because I was the little boy left at home. They had a good time and I was allowed to look at the program. My brother and I used to have play fights in the front room. He got me into WWE wrestling and we would pick our heroes out of the magazine. It was our passion and we would compete and watch our wrestlers whenever we could. That was a passion that Mark gifted me with and I still now follow it hardcore at 28 years of age. Mark was a great big brother and we traveled through life together.

We then moved to Luton, away from family and friends, to a new start with mum dad and our dog Ned. Mark would continue in secondary school and I would continue in junior school. I was a big baby. I cried for mum daily but Mark just got on with things and as always, took anything in his stride - often telling me what a baby I was. Mark would continue to take life in his stride taking all hurdles in his way. I was too young to know his body was failing him. From the ages of 11 to 18 Mark and I weren't close. We would just get on with taking our new adventures in life, him in high school and 6th form, me just following behind. Mark would always be the one to get me into things. He would recommend and I would usually follow. Mark introduced me into the world of comedy, we would follow Bottom, exchanging quotes and cracking up with mum and dad not knowing what we were on about. That continued until Mark's last days but of course by then I thought I was better and had a better mind for better comedy.

Mark changed from being my older brother to just being my brother with problems, although I would never admit them because I felt it would be admitting defeat. Mark had struggles and he just took them in his stride but he always looked out for me and supported me whenever I needed it. We weren't very close and argued a lot because of frustration, but if I needed him he was there and it wasn't until I was older that I would return support in any way possible, little things, but things I could do.

Mark grew older and his body failed him and his OCD would take over. He would be in the bathroom a lot cleaning and wouldn't interact as much as he used to. I feel in some ways I turned into the older brother. I would run around after him when asked to, make sure he was ok before I went out and do what he wanted when it was just us two on our own - when if truth be told he looked after me just as much if I had been out on the beer.

Mark joined the infusion program. I never took much notice but I could see that his personality returned, we would wind each other up during the course of the day STILL quoting comedy scenes. We tried to make each other laugh but not one of us would give the other the satisfaction of knowing they were successful - it was all banter, but OUR banter.

Mark was Arsenal through and through and I was Liverpool for some reason. He would take great delight when Arsenal did well and Liverpool didn't and even when Tottenham failed it was a quip for dad - Mark hated any other premiership team with a passion. Mark was a very intelligent man. He did well in school and his thirst for knowledge was amazing.

From the day I was born until the day Mark passed away, he never stopped being a brother to me. Yes, the characteristics changed, we got on so well in the later years and really knew each other's personalities. Humour was what brought us together, he was a keen drinker and could put me to shame every time.

I was lucky enough to go out for a meal for his birthday as a family. It was me and him drinking Stella and it was pure sarcastic comments all night at any situation. My dad was always the funny one but he didn't realise that me and Mark followed suit and we were just as funny - Mark was a very funny so and so.

I was shocked the day Mark passed away and still am, but in his short time on this earth in his own way he was happy, on his terms he dealt with everything and never once complained and would often offer me advice. Mark and I went through many stages and never once did I stop loving him or ever will and he loved me, I know that because we told each other quite recently.

Mark got a poor deal with Hunter's but he dealt with it. He had a great start to life, a patchy middle when things went down and a happy end. Mark touched our lives so much and it's only now you realise it.

We miss you Mark. Come back soon.
Micky Dicky, Sugar, Dad

"You're embarrassing yourself!"
1977 – 2009

My Story by Darrin Minett

It had been an average sort of summer's day and I had just finished work. Being a postman means I could enjoy the rest of the day in the garden when the weather is fine. There was a lot of mail as usual, which does take it out on you, but I was feeling more tired than normal. This tiredness, I thought, was down to the heat of the day along with the exertions of my three and half hour round (or workout as I call it). Cycling, running and walking are all part of my daily work routine. As I cycled home, I could feel my mobile vibrating in my pocket. As I ride along a very congested busy road, I decided to ignore it and check who had called when I got home.

On arriving home I was greeted by the wife, a cup of tea and a plate of bacon sandwiches. Marvellous. I decided to take my refreshments onto the patio on the back garden. Once seated, I remembered that I needed to inform my delivery office that I had successfully delivered both the special deliveries that morning. When I took out my phone, I noticed the missed call. I couldn't believe I had forgotten about the call almost immediately. Another thing I put down to getting older.

On checking, I realised my brother Chris had rung, so I rang him back straight away. I had to, as he worked on afternoons for a local brewery and I needed to catch him before he set off for work. He never answered any calls whilst at work. When he answered, he told me that he had something to tell me, but he wouldn't do it over the phone, so he arranged to pop round the next day for a chat. I instantly thought he'd won the lottery. He was lucky that way. He always seemed to be getting four balls or more, but never the magical six numbers. Maybe this time eh!

Anyway, he turned up the next day, (I'd forgotten he was coming incidentally). He instantly reminded me about the illness that he had suffered from back in February, earlier that year. I knew he had been diagnosed with pneumonia at the time, but we'd never spoken about it.

He then proceeded to tell me about a doctor at the local hospital who had been dealing with him. He had been kept in hospital several nights to monitor the illness and during his time there, a doctor noticed several symptoms that seemed strange to him and he stated that they needed further investigation. He informed my brother that he believed he had underlying health problems that may or may not be connected to a rare disease that he had some knowledge about. He was then sent to have tests in a Manchester hospital.

Unbeknown to me, Chris had been to Manchester two or three weeks earlier and yet again the previous day. He then told me he had been diagnosed with Fabry disease: I just listened. Dr Waldek had taken several blood and urine samples which confirmed the disorder, but he told Chris that he was already sure he had it because he had visible signs of accompanying problems.

This is when my brother proceeded to tell me about his own medical history. It was probably the first time in twenty years that he had told me anything personal. We are both very private about our lives. What he told me however had no resemblance to my own medical history other than the pains in the hands and feet we both experienced when we were kids. Then he told me that he had (probably) inherited it from our mum and that the chances that I had it were very high. I was advised to inform my doctor and to ask to be referred to Dr Waldek to have the simple tests done, just to be safe. He also told me not to read about it on the internet, as Dr Waldek had told him not to read too much into it.

Anyway as soon as he had gone, I ignored his advice and read into the illness on the internet. You see, I'm just too nosy and I needed to know. I found a medical page and read some of the symptoms. Then things began to register as I started to recall my own medical history.

When I read about Acroparasthesia, I finally knew I had found a possible answer to the pains in my hands and feet when I was younger. This often occurred when I did anything sporty, but it never stopped me. I played football daily, I ran for the school in the cross country team and everything I did as a child involved sports-like activities, so basically, I ignored the pains and became almost ignorant of them. I actually believed they were 'growing pains', as my doctor once told me they were those mythical ailments we suffer from as children.

These pains also often occurred just before the onset of damp weather. "It'll rain in an hour" I used to say to people. Then in my late teens they disappeared, well almost. I no longer had any problems when doing physical activities. Maybe I had become so accustomed to them, I ignored them totally. However, the pains reappeared whenever I had any illnesses. They seemed to make every illness follow the same pattern. It felt like my hands and feet were on fire and it would spread up my arms and legs and I would be in agony. This is when I established that I may have been having 'Fabry crises' for over twenty five years. The evidence was beginning to stack up.

I haven't eaten or drank any dairy products for over twenty years, as these caused upset stomachs and migraines in my late teens. So I removed them from my diet and the migraines stopped straight away and I've never had one since. However, I still had minor digestive problems, which although I'd never admit it, they were getting worse. In my late twenties, I had tests done to deal with this problem, but the doctors found nothing. Another problem arose during the tests when I had to have a barium drink, so I could have my intestines and bowels x-rayed. This drink sat in my stomach for just

under four hours. The doctor wasn't impressed, he couldn't understand why it sat there for that length of time and actually shouted at me asking 'what's wrong with me?'. 'You tell me pal, that's why I'm here', I recall thinking. To make things worse, I had the pleasure of being left in a main corridor of the hospital, laying on a trolley, with every Tom, Dick and Harry walking by and staring at me continuously. Great!

The only other time I had been properly ill, was thankfully, the last one and it was back in 1998, when I contracted Chicken Pox. I caught it off my kids. I'd never had it as a child and while my kids were both back to full health in two to three days, I was very ill for just under five weeks. Since then, I have been of very good health for about the last six years.

Then in 2005, I was made redundant from my company Kodak, who were relocating to China. As the company was a very high profile one, we had lots of other companies i.e. Warburtons, Cadburys and the like, come and offer work for our employees. One company based on Merseyside wanted fifteen gas engineers. The offer was to train them from scratch and give them positions after training. I jumped at this and was successfully set on.

Sadly, doing work with vibrating tools had a massive effect on me. During the night, I had very poor blood circulation in fingers, hands and arms. It got so bad that I could not lift myself up to try and let the blood flow back down my arms. Eventually, due to sleep disruption and being unable to feel anything in my hands, I was medically finished. I was diagnosed with Reynaud's Syndrome, which is extremely annoying, especially in my current job. My hands and feet react to the weather conditions, even when I wear waterproof, windproof, double insulated skiing gloves. I have to put my hands into very warm water when I get in, to help them get warm and help the blood flow.

Although I was still very fit, (I attend a gym four days a week, as well as doing my job) I had noticed a slight deterioration in my energy levels, but I put that down to my diet and my age (42). On a couple of occasions I noticed I had palpitations, which did not go unnoticed. The thing with palpitations was that I knew most people have irregular heartbeats. These made me slow down however and I stopped doing my job at one hundred miles per hour and took it steadier in the gym. I had no need to rush about like I did, but I knew I had to keep an eye on it. Also, my digestive problems seemed to be getting worse, but like I say, I tolerated it.

So the information on the Fabry medical page helped me piece together the jigsaw of minor problems that I had questioned in my own mind. Now to find out if Fabry was the answer?

My own doctor, thankfully, referred me to Manchester and the rest is history. I had no visible symptoms like my brother, but I do have minor heart problems, (i.e LV Hypertrophy, Bradycardia), Tinnitus, and poor blood circulation in my hands and feet. Since having my infusions at home, (by the wife, I must add), my digestive problems have almost ceased and there has been welcoming improvements in the conditions I have mentioned. My MR scan on my brain showed minor activity (yes, something is occurring inside, but not much, just the odd tumble-weed rolling by) but hopefully nothing too serious.

Since diagnosis, my sister has been tested all clear, which is great news. My daughter however, has the condition, but as yet she has no ailments. Fingers crossed there then. A search into the family history found that I could have ended up like my great grandma, who died aged forty, my grandmother, also died aged forty and my uncle dying aged 54 from heart and kidney problems. Though Fabry was not diagnosed on their deaths, the problems they had does indicate possible Fabry disease. My mum as yet, has not been tested. At 65, I don't know whether it would be of any benefit to her or if she is interested about finding out. It is bad enough for her to think she has been responsible for our illnesses, even though WE do not see it that way.

The Enzyme Replacement Therapy (ERT) has given me cause for optimism. My results show a reversal in some symptoms, so hopefully this will continue. I have switched from Fabrazyme to Replagal, like most UK patients, and I am still not sure which I like best. My initial ERT with Fabrazyme gave me random side effects, which obviously diminished over time. The switch to Replagal was comfortable with only minor side effects. Time will tell. Luckily, Royal Mail have been fantastic about my illness, especially as it is covered by the Disability Discrimination Act.

Overall, I consider myself very lucky, especially compared to some of the other Fabry patients I recently met at the German Morbus Fabry Patients Conference held in the beautiful city of Vienna at the end of November 2009. This condition does affect everybody differently, though I am not going to get complacent about my health, as every little shot of pain or palpitation is noted.

So I'd like to wish everyone who has Fabry all the best of luck in the world and I'd like to thank the MPS Society for giving me the opportunity to tell you all this.

Darrin Minett

Living with Fabry

How do I start to write this article? At the moment I am sitting in my reclining chair writing this and recovering from a stroke! Let me tell you what happened…

Last time I wrote for the MPS Magazine I told you about the recurring infection in my leg. I never really felt that I had got over it. Then on the evening of Friday 4th December I had been talking, as I do often, with my friend Leslie Hilliard, who also has Fabry. I finished on the phone and was ready to start eating my evening meal. At the same time I was also downloading something on the computer and the spinning curser started moving very fast, but not as fast as the room was.

The next thing I was aware of was slipping onto the floor and being unable to get up or move my legs. The paramedics were called and strapped me up to an ECG machine. Of course it was abnormal and this concerned them but it was up to me if I wanted to go to hospital. We explained that the ECG was normal but the symptoms really corresponded with a TIA, a kind of stroke.

I went off to A&E (where I felt like I was issued with a 12 month season ticket). I got a bed overnight and was sent home on Saturday afternoon after being told I would be called back to the cardiology department because (you guessed it) I have a heart problem.

The rest of that day and Sunday I did not feel good. On Sunday night I went to bed as normal with my usual glass of pop at the side of the bed just in case I woke in the night. I was indeed woken by a horrible pain in my nose. It was really severe so I went to get up and sat on the edge of the bed and took a good swig from the glass. But, it wouldn't go down my throat, it came back up just as fast as I tipped it into my mouth. I then tried to get cleaned up but as I stood up I promptly fell over. Again we called 999 and I was taken back to A&E. However, after a couple of hours of being quite poorly in A&E, a bed was found for me, as it happened, on the stroke ward.

Apparently it is not easy to get on that ward but I guess I was in the right place at the right time. I was under Dr Meara who was lovely. He had a very gentle manner and every day he wore a bow tie which reminded me of the Carry on Doctor film!

Initially there was some debate as to what had actually happened to me but after 36 hours a neurologist looked at my scan and detected a rare type of stroke in the cerebellum. This caused all sorts of problems as you can imagine. I could not swallow so I was on intravenous treatment, I had now got pneumonia as well so loads of antibiotics and pain killers were being pumped into me and I had an naso-gastric tube for feeding. I was also being treated for hiccups which I had constantly for 10 days. Although they wore me out, they gave my visitors a good laugh!

One of my friends, Alan, runs a small coach firm but he also works as an undertaker. Judging by the way he was dressed when he came to visit me in hospital, Alan must have done a funeral that day. We were sitting talking when suddenly he pulls a tape measure out of his pocket and starts to measure me! It made everybody laugh and I asked if he had any brochures or cards I could leave on the ward for him.

The following weeks came and went with a mixture of physio sessions, treatment and scans. It is very hard to learn to walk again but I'm on the case. The problem is not so much paralysis of the limbs but is instead to do with the balance centre and my control panel which have malfunctioned.

I am very fortunate really as it could have been a lot worse. However, at the moment I have problems swallowing so I keep aspirating and because of that I get chest infections and walking is hard. But, hey guys, I don't have to walk round shops anymore, I always knew there would be a silver lining!

I now have a long recovery ahead but I'm sure things will continue to improve and in the meantime I am trying to live as normal life as possible.

I think it is only natural to ask if the stroke could have been prevented. I don't really know the answer to this, but maybe medication might have helped. What I do know is that the stroke consultant said "the stroke was a direct result of the Fabry disease."

Stroke is in our family pathology and not all with Fabry will have a stroke. Prior to my stroke I had a constant headache for about six weeks and, as it got closer to the event, I started to have problems with my vision. My advice would be that if you are concerned at any stage you must get it checked out. Don't be afraid of saying you think there is a problem. Nobody knows you as well as you do and if you feel things are not right then you must seek advice.

This article has been a challenge to write as my brain gets tired very quickly but I hope my experience can benefit you all. Keeping a PMA (positive mental attitude) is hard but in all honesty it does get easier the more we try and look for the good side of things. **Ian Hedgecock**

Editor's Note: If you would like to contact Ian, please get in touch with the MPS office

The Fabrazyme Shortage - Update

The shortage of Fabrazyme has presented a major challenge to the Fabry community. The initial requests from Genzyme in July 2009 were to reduce Fabrazyme requirements for a period of a few months. Unfortunately, it was clear by August 2009 that much more substantial reductions would have to take effect.

These requests called for concerted action by patients, carers, the Lysosomal Storage Disease teams at the various centres across the UK and the Department of Health. A series of telephone conferences were held and a strategy was developed. Initially as it appeared that the shortage would be short-lived, we all agreed a dose holiday of one or two infusions over several months. By September it became obvious that the problem of shortage of Fabrazyme was much more serious and unlikely to be resolved into 2010. At this point, many Fabry sufferers, who relied on Fabrazyme as their Enzyme Replacement Therapy (ERT), were being transferred to Replagal, an alternative ERT for Fabry disease. As the true extent of the Fabrazyme shortage only became known in a drip drip fashion, it has been incredibly hard for doctors wanting to make decisions in the best interests of their patients and the patients themselves.

The Fabrazyme shortage continues and it now looks as if normal supplies of Fabrazyme will be available towards the end of 2010. This is positive news for patients that have continued on Fabrazyme throughout the shortage and for those on alternative products who wish to transfer back to Fabrazyme. We do not underestimate the upheaval patients have felt. Many of you we know have transferred to Replagal (Shire HGT) and some of our members have enquired recently as to whether they have to change back to Fabrazyme. If you, or you acting for your child, do not wish to transfer back to Fabrazyme you should be having this discussion with your Fabry doctor or child's paediatrician. If there is no over-riding clinical reason given for transferring back to Fabrazyme, the decision as to which product you are on should be yours. It is always a good idea to ask your Fabry doctor to explain in lay terms his or her reasons for prescribing one ERT over the other and the clinical benefits to you.

Getting involved with MPS....

Can you help with our media campaign?

Do you ever wish that more people knew what you meant when you started talking about MPS or Fabry?

One way to help change this is to spread awareness through the media - newspapers, magazines, TV and radio for example. We are looking to promote MPS Awareness Day through the media and need your help. Would you be willing to provide a case study, a story about your or your family's experience of living with MPS or Fabry? Perhaps you have an unusual story about receiving the diagnosis, have achieved something amazing with MPS or Fabry, are thinking of doing some fundraising, or simply want to tell your story.

To find out more please email us at
fundraising@mpssociety.co.uk

Fundraising resources available from MPS

Fundraising packs
Fundraising fact sheets
Sample press release
Sponsorship forms
Become a Friend of MPS
Promotional Goods Order Form
Publication Order Form
T-shirst, posters, balloons, collection boxes...

Email us at fundraising@mpssociety.co.uk,
visit www.mpssociety.co.uk or phone 0845 389 9901

Find us on Facebook

We have set up the MPS Society Facebook page as a means of providing information to our MPS Members and friends quickly and efficiently. We hope to feature some of our events and activities and recognise those that contribute to the Society and the work that we do.

You can find us by entering *MPS Society* into the facebook search engine.

As well as aiming to provide you with news from our fundraising activities and MPS events to coincide with our quarterly MPS Magazine we are also hoping to encourage greater awareness of the MPS Society.

If you have any ideas or suggestions for our facebook page please email
facebook@mpssociety.co.uk

Do you have a story to share?
Please email
newsletter@mpssociety.co.uk
or phone **0845 389 9901**

The Childhood Wood

History of the Childhood Wood

In 1992 the Society was asked to propose an idea to commemorate children who had died from an MPS or related disease. Out of many ideas, the Childhood Wood was born.

The following year the MPS Society was given under licence an area of Sherwood Forest to create a Wood of saplings which were cloned from the Great Oak.

In February 1993, 150 saplings from ancient Sherwood Oaks were planted by MPS families, supported by Sir Andrew Buchanan, Lord Lieutenant of Nottinghamshire along with local MPs and representatives from Nottinghamshire County Council.

The first sapling to be planted was in memory of Simon Lavery who died from Hunter Disease MPS Type II, in 1982, by the Rt. Hon. Michael Howard, then Secretary of State for the Environment.

Since then each of the oak trees planted annually celebrate the life of a child whose childhood was destroyed by these cruel, degenerative diseases. These trees which were planted in the early years are now branching out and forming a canopy.

Over the years further saplings have been planted by families and friends of children who have lost their lives to MPS or Related Diseases.

Annual Tree Planting in October

The annual Tree Planting Day takes place during October and families are invited to attend and plant a tree in memory of their loved one. The day starts with lunch at a nearby hotel with all the bereaved families and they are joined by local dignitaries including the Lord Lieutenant and Councillors from Nottinghamshire. After lunch we gather in the Childhood Wood, where the names are read of those children who have lost their lives to MPS or related diseases and we listen to a reading of the beautiful poem 'Remember'. This is followed by the planting of a sapling and the release of balloons by those who have gathered to remember their children.

Childhood Wood Remembrance Day

Each July the MPS Society invites families who have lost sons or daughters to MPS or related diseases for our annual Remembrance Day. Families gather in the tranquillity of the beautiful Childhood Wood to attend the Remembrance for those who have lost their lives, this is then followed by a balloon release in their memory.

Over the years and working in Collaboration with the Forestry Commission, the MPS Society has put in place memory boards and new pathways, all of which are accessible to visitors to the Childhood Wood.

Walking through the pathways, there are a number of wooden animals and a picnic area which are dotted throughout the area offering a peaceful setting for those who wish to reflect on their thoughts before leaving the Childhood Wood. This later development and enhancement of the facilities was made possible by two generous grants, in 2007 and 2009, from the Geoff & Fiona Squire Foundation.

Directions to Childhood Wood

The Childhood Wood is situated in Sherwood Pines. Nottinghamshire and is part of the Clipstone Forest off the B6030 near Clipstone Park. Park in the car park (which costs £3.00), walk down to the picnic area and then follow either the Blue or White Path. The White Path is suitable for wheelchair access.

The dates for this year's events:

Childhood Wood Remembrance Day	11 July 2010
Childhood Wood Tree Planting Day	22 October 2010

Linda Warner l.warner@mpssociety.co.uk

During our National Conference in August 2009 we included a presentation which was dedicated to bereavement and the support provided by the MPS Society, in particular the plans for the work of the Advocacy team.

Some of this work includes:
- The Childhood Wood
- To recognise and understand the grieving process
- Useful and practical information for bereaved families
- Information booklet for Grandparents
- Directory of Bereavement Support Services throughout the UK

In the first of our specialised articles we look at the Childhood Wood, its history and development over the years and information regarding the annual Remembrance Day in July and Tree Planting Day in October.

In the next MPS magazine, we will look at how to recognise and understand the grieving process.

For further information regarding the Childhood Wood please contact the MPS Society on 0845 389 9901.

Editor's Note:
Linda Warner is the *Roald Dahl Advocacy Officer for Progressive Neurological MPS Diseases*

Providing practical support for children with serious brain and blood related problems

Help us care for today and give hope for tomorrow
Leave a gift in your Will

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 achieve its long term objectives is to include the Society when drawing up your Will.

For more information please contact us for our Leaving a Legacy fact sheet which is also available to download from our website.
www.mpssociety.co.uk

Grandparents Fact Sheet

Here at the MPS Society we realise the importance of the extended family when it comes to caring for a child with an MPS or related disease.

With this in mind we are developing a Fact Sheet for Grandparents to address the significant role they play and the emotional hurdles and issues that they face when a grandchild is diagnosed.

Our first edition of this will be available to download from our website from May. Those who do not have access to the internet can request a hard copy to be posted to them for a small fee to cover printing, postage and packaging.

We hope that this new information resource will not only provide useful advice and information on how Grandparents can help both their children and their grandchildren, but also encouraging grandparents to come to terms with, and identify, their emotions.

We are encouraging Grandparents from all walks of life to come forward and share their stories with us in the hope that their experiences will help others. You could make a contribution to our Grandparents Fact Sheet by writing a case study, a poem or simply providing us with a few 'helpful hints' on how you have dealt with your situation from a grandparent's perspective.

Get in touch with us either via email:
newsletter@mpssociety.co.uk
or by calling us on: 0845 389 9901

MPS REGIONAL CLINICS

Bone Marrow Transplant Clinics

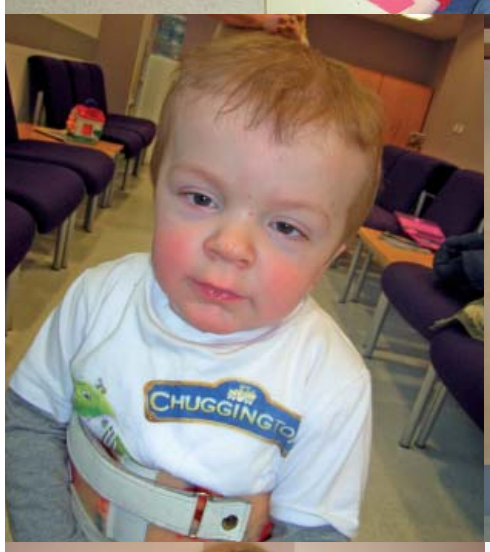
Royal Manchester Children's Hospital

Under 6's BMT Clinic, photo top row: Shantelle Reilly with her siblings, photos middle row: Ethan Greening, Luke Blignaut and Lyla Heppleston. Over 6's BMT Clinic, photo middle row, far right: Callum Pollock and Jordan Mount, photos bottom row: Rubina Jalani, Thomas Mett and his dad, Leighton Barker.

The first Bone Marrow Transplant clinic of the New Year took place on 15th January after another big fall of snow. By the time I had arrived in Manchester most of the snow had melted, however it was still very icy. This clinic was for the under six year olds. I had met the families before but I was wondering how the children had changed since I last saw them. I wasn't disappointed! All the children were definitely bigger than last year and they didn't lose any of the enthusiasm they showed previously (all the toys the children played with would agree with me!).

On 22 January I was on a train to Manchester again to meet with the families of the over six year olds. The clinic was a busy one and it was lovely to see old faces again and some new ones too! The clinic also was an opportunity for families to meet up as for some it had been a long time since they last saw each other. As usual, the clinic was very lively and it ran smoothly.

The turn up at both clinics was excellent. Adults were exchanging stories about the extreme weather conditions and children were happy that because of the snow they didn't have to go to school. It was great to meet with everybody and I'm looking forward to seeing you again when it's warmer! **Jolanta Turz** j.turz@mpssociety.co.uk



Bristol MPS Clinic

On 2 February Bristol held its first MPS clinic of the year. Professor Ed Wraith joined Dr Jardine, Dr Pierre and the metabolic team to see patients from in and around the South West area. The MPS Society was also there to support individuals and their families and it was nice to catch up with so many of you.

At the end of last year the Society wrote to all patients seen at Bristol to explain that the organisation of this clinic was transferring to Bristol. This was following the appointment of Dr Germaine Pierre who is the new paediatric Metabolic Consultant.

We would like to assure you that clinics in Bristol are still continuing and that Professor Ed Wraith and the Society will still be attending as we have always done. The only real change is that appointments will be sent from Bristol, so if anyone specifically wants an appointment or needs to know when the next clinic is being held, they need to contact the Bristol team directly.

Lastly I would like to thank all the team at Bristol and Prof. Wraith for a successful clinic and wish Sally Melson bon voyage as she has taken up a new position within the hospital. **Sophie Thomas** s.thomas@mpssociety.co.uk

Birmingham opens their new consultation room at the MPS clinic

In 2007 the Society, as part of its 25th Anniversary celebrations, wanted to recognise MPS Specialist Centres who have strived over the years to make a real difference in the lives of those suffering from MPS, Fabry and related diseases.

With this in mind we invited each centre to submit a proposal detailing how they would spend a small grant to benefit those working in the field of Lysosomal Storage Diseases and their patients.

Birmingham Children's Hospital proposed to spend the money on adapting a consultation room, to make it a safer area for children, particularly those with challenging or destructive behaviour. To do this it would require padding for the walls and activities to try to occupy or distract them.

On Friday 12 February 2010 I attended the Birmingham MPS Clinic and was also invited to open their new consultation room along with Nathan Oakley and Adikah Shah. We are pictured below right.

Nathan Oakley had also gone to great lengths to help raise money for the room and his family wanted to pass on their thanks to Pat Tite, for her continual hard work and dedication raising funds at Langley Ward Conservative Club in the West Midlands and presenting the Metabolic department at Birmingham Children's Hospital the proceeds which went towards creating a much needed secure room for children when visiting clinic. "It is the hard work people like Pat do to help children like our beautiful son and brother Nathan to give them the best quality and care life can give and often these people go unappreciated but today we are happy and proud to say you have impacted on our lives, reminding us of how beautiful and selfless people can be. Our thanks also extend to the Committee and its members for playing their part in raising these much needed funds. God bless, our love and thanks now and always." **Jo, Dave, Fran and Nathan Oakley**

It was a privilege to be invited to open the room along with Nathan and Adikah who has attended the hospital for approximately the past 15 years. I sincerely hope that this room offers families a better experience when attending appointments.
Sophie Thomas s.thomas@mpssociety.co.uk

Party at Birmingham Children's Hospital

On 13 February Birmingham Children's Hospital invited me to join them at their Valentines party. It was held at a local conference centre and on arrival I was greeted by an abundance of heart shaped balloons and decorations. They had a great entertainer who was a jack of all trades as he was able to do balloon modelling, circus tricks, act as a magician and lastly was a DJ getting everyone up dancing, including parents and staff. In addition to this there was a raffle, the parents were offered a back, shoulder and head massage and the kids had a goody bag to take home.

Catherine Stewart wrote the following explaining the purpose and the background to why these types of events are important.

'Birmingham Children's Hospital decided to hold a party for children with mucopolysaccharide disorders to enable them to meet together socially on an informal basis away from the hospital environment in Birmingham. The party was intended to provide a fun social activity for the children whilst also providing the opportunity for parents, families and siblings to meet together informally and share some quality time. In addition to providing entertainment and fun activities for children, we felt it important to provide a social forum for families to enable them to get to know each other, share experiences and pool knowledge and advice. We also felt that it was useful for the children to see some of their health professionals away from the hospital setting to help them feel more comfortable with their carers when they come into hospital for appointments and to demonstrate our commitment to their social needs in addition to their physical / medical needs as part of our commitment to a holistic 'whole child' approach to their care.' **Sophie Thomas** s.thomas@mpssociety.co.uk

Help us celebrate MPS Awareness Day on 15 May 2010

Each year the Society celebrates MPS Awareness Day on 15 May. This is a day devoted to raising awareness of MPS and Related Diseases and many of the International MPS Societies participate in this special day.

In conjunction with this, Birmingham Children's Hospital are delighted to announce a National Lysosomal Storage Disorders Awareness Study Day.

It will take place on Friday 14th May 2010 at The Studio in central Birmingham. Registration is free of charge.

The meeting is aimed at all healthcare professionals with limited experience of Lysosomal Storage Disorders.

To view the programme and to register for the meeting, go to www.cfsevents.co.uk and click on National Lysosomal Storage Disorders. A map and directions to the venue can also be downloaded from here.

If you would like further information, please do not hesitate to contact us on 01438 751519.

Further information about MPS Awareness Day can be found at **www.mpssociety.co.uk**

Introducing Victoria Crook

My name is Victoria Crook and I am a Clinical Nurse Specialist (CNS) for Lysosomal Storage Disorders at Great Ormond Street Hospital. I joined the team in July 2009 and it's been non-stop ever since.

Prior to this, I worked in various areas of nursing over the course of my career. I have covered medicine and surgery for children under three years old, child and family nursing in the community, childrens A&E, and worked as a specialist nurse in the Magnetic Resonance Imaging department. Most recently I was working in clinical research which led to my current job.

My role is both exciting and challenging as it involves co-ordinating a service for children and families where I am a key member of a team looking after their needs. Together with the other CNS – Niamh Finnegan, we co-ordinate care, support families, and liaise with other professionals and groups involved with the care of these children both within GOSH and in the community. This also includes working closely with support groups and ensuring families are well informed of all services available to them.

I look forward to meeting all the families and working together with all of the families that attend Great Ormond Street Hospital.

The MPS Society would like
to congratulate Catherine Little
on her marriage to Chris Stewart
on 23rd December 2009.
Catherine is the Clinical Nurse Specialist
and MPS IVA Study Co-ordinator at
Birmingham Children's Hospital

Thank you to all our supporters!

The MPS Society is very grateful to our fundraisers and supporters for all their hard work in raising money through organised fundraising events, sponsored events and other activities, big or small.

We are so appreciative of your support and for thinking of the MPS Society. We get a number of requests each year to attend cheque presentations or give talks on our work. We always like to do these when possible but to minimise the costs to our charity, try to coincide these with other visits in the local area or en route to other meetings or events. Thank you to all our fundraisers for their continued and very vital support. We need you!

For more information or to request a fundraising pack, please phone us on 0845 389 9901 or email us at fundraising@mpssociety.co.uk.

Clinical Trials Update

MPS I, MPS II and MPS VI

A clinical trial of human growth hormone (HGH)
This clinical trial is being conducted at the University of Minnesota Children's Hospital. Children with MPS I, II and VI with short stature are invited to participate. HGH is a US Food and Drug Administration-approved treatment for short stature, however there is no data at this time on using this treatment specifically in children with MPS. The goal of this clinical trial is to determine what, if any, effect HGH has on growth velocity, bones and cognitive functioning of children with MPS I, II and VI. For additional information contact Linda Polgreen MD, Polgr001@umn.edu

Phase II Study of Aldurazyme Enzyme Replacement Therapy (ERT) with Haematopoietic Stem Cell Transplantation (HSCT) for Hurler Disease
This study is currently recruiting participants.
The purpose of this study is to prove the hypothesis that weekly infusions of Aldurazyme ERT for 10-12 weeks prior to transplant and 8 weeks following transplant will result in a reduction of glycosaminoglycans (GAGs) burden that is associated with Hurler and transplant related pulmonary complications following transplant.

Clinical Trial Site: University of Minnesota, Minneapolis, orchaoo1@umn.edu

MPS I

Extension Study of Intrathecal Enzyme Replacement Therapy for MPS I
This study is currently recruiting participants.
This is a one-year extension study of the use of laronidase into the spinal fluid to treat spinal cord compression in mucopolysaccharidosis I. Spinal cord compression occurs in this condition due to accumulation of material called glycosaminoglycans (GAG). Laronidase is the manufactured form of the enzyme alpha-l-iduronidase that is deficient in mucopolysaccharidosis I patients. The aim of this study is to determine whether laronidase is safe and effective when given into the spinal fluid as a potential non-surgical treatment. This study is a Non-Randomized, Open Label, Uncontrolled, Single Group Assignment, Safety/Efficacy Study for spinal cord compression due to mucopolysaccharidosis I. If successful, intrathecal delivery could represent a practical, straightforward method of treating central nervous system disease due to lysosomal storage.

Clinical Trial Sites: Los Angeles Biomedical Research Institute at Harbor-UCLA, USA and Helsinki University Central Hospital, Finland and University of Minnesota, Minneapolis, USA

For additional information contact Patricia I Dickson, MD pdickson@ucla.edu

A Multi-Centre, Multinational, Open-Label Study of the Effect of Aldurazyme Treatment on Lactation in Female Patients with MPS I and their Breastfed Infants
This study is currently recruiting participants.
This study is for lactating women receiving Aldurazyme, and their breastfed infants, or women who are pregnant, receiving Aldurazyme and planning to receive Aldurazyme whilst breastfeeding. The purpose of this study is to determine if Aldurazyme is present in the breast milk of post-partum women and the effects of Aldurazyme on the growth, development and immunologic response of their breastfed infants.

For further information discuss with your clinician, medinfo@genzyme.com.

MPS II

A Multi-Centre Observational Study Evaluating Anti-Idursulfase Serum Antibody Response in Hunter Syndrome Patients Enrolled in the Hunter Outcome Survey (HOS) Receiving Idursulfase Enzyme Replacement Therapy
This study is currently recruiting participants.
The objective of this study is to evaluate the effect of anti-idursulfase IgG, IgM & IgE antibodies on idursulfase safety (measured by infusion related adverse events) between patients who develop anti-idursulfase antibodies and patients who do not after long-term idursulfase enzyme replacement therapy. This is a multi centre, observational study in a prospective cohort of affected patients.

Research Sites: Children's Hospital & Research Centre Oakland, California, USA; Hospital de Clinicas de Porto Alegre, Servico de Genetica Medica, Porta Alegre, Brazil; Birmingham Children's Hospital, UK

A Safety and Dose Ranging Study of Idursulfase (Intrathecal) Administration Via an Intrathecal Drug Delivery Device in Paediatric Patients With Hunter Syndrome Who Have Central Nervous System Involvement and Are Receiving Treatment With Elaprase®
This study is currently recruiting participants.
This Phase I/II study is designed to obtain necessary safety and exposure data, as well as secondary and exploratory outcome measures, to be interpreted and used in the design of subsequent clinical trials. This is a Phase I/II Randomized Safety and Ascending Dose Ranging Study of Idursulfase (Intrathecal) Administration Via an Intrathecal Drug Delivery Device (IDDD) in Paediatric Patients With Hunter Syndrome Who Demonstrate Evidence of Central Nervous System Involvement and Who Are Receiving Treatment With Elaprase.

MPS IIIA

A 12 month Longitudinal Prospective Natural History Study of Patients with Sanfilippo Disease Type IIIA (MPS IIIA).

This study is currently recruiting participants.

The purpose of this study is to identify potential surrogate endpoints that may be utilised in future ERT trials of MPS IIIA via defined assessments including standardised clinical, biochemical, neurocognitive, development and imaging measures. The primary outcome is to evaluate the course of disease progression in patients with MPS IIIA who are untreated with any investigational products to inform possible future treatment studies.

Clinical Trial Sites: Royal Manchester Children's Hospital, UK and University of Minnesota, USA.

Surrogate Endpoint Trial (SET) for individuals with MPS IIIA (Sponsored by Shire Human Genetic Therapies)

SET is a one-year, multi-centre study designed to study the natural progression of Sanfilippo A Syndrome, or MPS IIIA, in approximately 20 patients.

Over a period of 12 months, participants in the study will be evaluated to assess the severity and progression of MPS IIIA, as measured by developmental age and milestones, central nervous system (CNS) function (including cognition, speech and motor skills) and biochemical markers or the condition (levels of heparin sulphate and its breakdown products in blood, urine and cerebrospinal fluid (CSF).

Additional information can be found on www.clinicaltrials.gov (identifier NCT01047306)

MPS IVA

Enzyme Replacement Therapy phase I/II Study

This open label within-patient dose escalation trial in 18 patients at three UK sites is nearing completion and will be followed by a treatment continuation phase. An announcement is expected soon regarding the Phase III/IV clinical trial.

Fabry

Stroke in Young Fabry Patients (sifap1): Frequency of Fabry Disease in Young Stroke Patients

This study is currently recruiting participants.

More than one million people in Europe suffer from a stroke every day. Normally older people have a stroke, but also a significant number of younger people between 18 and 55 years. Usually, these cannot be explained by the classical risk factors such as diabetes, overweight and high blood pressure. New studies indicate that in about 1 - 2 % of the younger stroke patients the cause could have been an undiagnosed genetic disease, the so called Fabry disease. The purpose of this study is to determine in a large number of young stroke patients, how many strokes were caused by Fabry Disease.

For additional information contact Arndt Rolfs, Prof., MD
arndt.rolfs@med.uni-rostock.de

Multi-centre, Open-label Study of the Safety and Efficiency of Control of Proteinuria with ACE Inhibitors and ARBs in patients with Fabry Disease who are Receiving Fabrazyme.

This study is currently recruiting participants.

The primary hypothesis of this study is to show that titrations of ACE inhibitors and Angiotension Receptor Blockers (ARBs) reduce urine protein excretion to <500mg per day in Fabry patients receiving agalsidase beta therapy at 1mg/kg every two weeks slowing the progression rate of decline of glomerular filtration rate (GFR) compared to case controls drawn from the Genzyme-sponsored Phase III extension study or the Phase IV study.

Clinical Trial Sites: Five throughout the USA; University of Wurzburg, Germany; General Hospital Slovenj Gradec, Slovenia

For other clinical trials currently recruiting participants, full details can be found on www.clinicaltrials.gov

Fetal Umbilical Cord (UCB) Transplant for Lysosomal Storage Diseases.

Safety and Efficacy Study of Several Replagal Dosing Regimes on Cardiac Function in Adults with Fabry Disease.

Safety and Clinical Outcomes in Hunter Disease Patients of 5 Years of Age and Younger Receiving Idursulfase Therapy.

Evaluation of Efficacy and Safety of Agalsidase Beta in Heterozygous Females in Fabry Disease.

Anderson-Fabry Disease in Chronic Kidney Disease Patients No on Renal Replacement Therapy.

A Study of the Effects of Fabrazyme on Mother's Lactation and on the Growth, Development and Immunologic Response of their Infants.

Study of the Effects of Oral AT1001 (Miglustat Hydrochloride) in Patients with Fabry Disease.

The genetic basis of MPS I: data from the MPS I Registry

Emma James DPhil (Oxon) – Senior Project Manager, Global Registry programme, Genzyme

Mucopolysaccharidosis type I (MPS I) is caused by deficiency of an enzyme known as alpha-L-iduronidase (IDUA), which results from changes, called mutations, in the normal genetic sequence of the IDUA gene [1,2]. (See Box 1 for definitions of terms used in this article.) People with MPS I inherit two disease-causing mutations – one from each parent – and the combination of two mutations is referred to as their genotype. More than 100 different mutations have been identified to date, and the type and location of a given mutation influences how deficient or abnormal the IDUA enzyme will be. Therefore, the two mutations in a person's genotype determine, to a large extent, how severe that person's MPS I disease will be [3,4]. The term 'phenotype' refers to the disease severity associated with a genotype, and MPS I is usually categorised into three general phenotypes of increasing severity: Scheie (less severe), Hurler-Scheie (intermediate severity) and Hurler (most severe) [1,2]. The phenotype is known for some, but not all genotypes. In general, the severity of a mutation is determined by the type and the location of the mutation, both of which influence how functional or non functional the abnormal enzyme will be.

The MPS I Registry is an ongoing global programme to collect data from people with MPS I [5]. At present, the Registry includes data from over 800 people with MPS I from over 30 countries, and is the largest observational database of its kind in the world. Thus, it is an unparalleled resource for clinicians and scientists, who can use the database for analyses that contribute to the medical understanding of MPS I disease and improve the quality of care for people with MPS I worldwide. One important analysis currently underway involves examining the association between genotype and phenotype among MPS I Registry participants. Such information may ultimately help doctors to make clearer decisions about when and how to treat MPS I, especially if newborn screening programmes are implemented.

In this article we summarise the published data to date on genotype–phenotype associations in the MPS I Registry and initial progress in predicting which mutations have the most severe consequences. Of the total Registry population at the time of analysis (over 700 people), genotype was reported for 293 individuals [6]. (Not everyone has genotype information, as it is not necessary for diagnosis of MPS I.) Approximately equal proportions of people with the three phenotypes had genotype data [6]:

- 45% of 182 Hurler patients;
- 45% of 73 Hurler-Scheie patients;
- 41% of 31 Scheie patients.

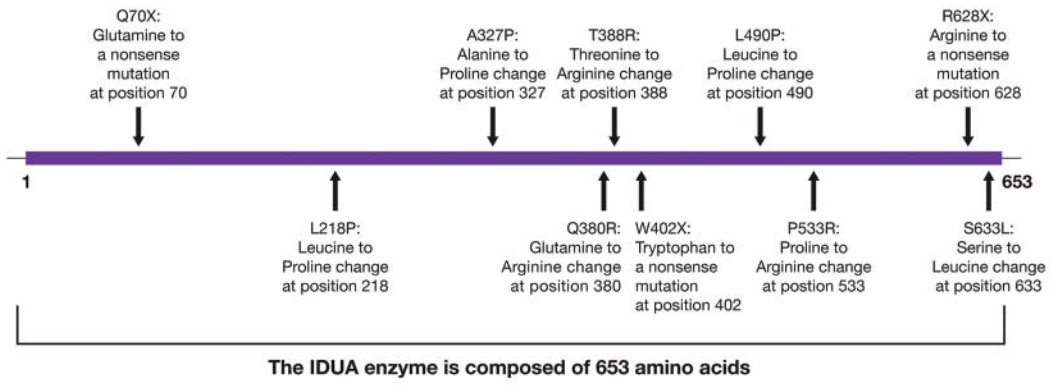
A total of 109 different mutations were reported for the 293 individuals with genotype information. Many of the mutations were reported in only one or two individuals, but 10 of the mutations were reported frequently and accounted for 74% of all mutations. A mutation can be described by using a letter-number-letter, which is shorthand for the location of the amino acid change that occurs in the abnormal enzyme (see Figure 1). The 10 most common were [6]:

- W402X, which was the most common mutation, accounting for 39%;
- Q70X, which accounted for 13% of mutations;
- P533R, which accounted for 7% of mutations;
- L490P, A327P, L218P, Q380R, T388R, R628X, and S633L, which each represented 1–5% of mutations.

It is possible for a person to inherit the same mutation from each parent, a condition known as homozygous, or to inherit a different mutation from each parent, known as heterozygous. The shorthand to describe a person's genotype shows the two mutations separated by a slash ('/'). When genotypes were looked at in relation to phenotype in the Registry, the following observations were made:

- Of 182 people with Hurler syndrome, the most common genotypes were: W402X/W402X (25%), W402X/Q70X (15%), and Q70X/Q70X (4%).
- All people with two 'nonsense' mutations, which produce little or no functional enzyme and are shown by 'X', had a Hurler phenotype.
- Of 73 people with Hurler-Scheie, the most common genotypes were: L490P/L490P (18%) and P533R/P533R (7%).
- Of 31 people with Scheie syndrome, 3 had D445del/W402X.

FIGURE 1



In addition, by looking at the phenotype that occurred when mutations were homozygous or when an uncharacterised mutation occurred in combination with a known severe mutation, the investigators were able to assess the likely severity of a number of previously uncharacterised mutations.

One of the benefits of having a global registry is that data from people around the world can be compared to see if there are differences in the disease between different geographic or ethnic groups. For example, the MPS I Registry has been used to explore phenotype distribution in patients from Latin America and in patients of Asian origin [7, 8]. In both of these groups, there are proportionally more patients with Hurler-Scheie syndrome than with Hurler syndrome, which is the opposite of what is seen among Registry participants from the United States and Europe, where the majority of people enrolled are reported to have Hurler syndrome. Thus, generalisations that hold for patients in one part of the world may not be true in other parts of the world, and these differences help inform clinical practice. As more data are collected, it will become possible to compare analyses on different sub-populations; for example, these data from Asian people could be compared with Caucasian people to see whether the disease progresses differently or if there are differences in responses to treatment between the two groups.

In conclusion, the genetic analysis of MPS I Registry data represents the largest study of MPS I genotype–phenotype relationships performed to date [6]. Continuing work in this area is helping to predict the severity of a given mutation by assessing the phenotypes of MPS I patients in whom the mutation is present. This includes assessing any differences in genotype and phenotype distributions in different geographic and ethnic populations. The ability to predict the severity of a given MPS I mutation may help determine if an individual is likely to develop symptoms, their severity, the likely clinical course of disease, and, in future, the most appropriate management approaches. This is ground-breaking genetic research, which will potentially transform the basis for diagnosing and treating people with the different types of MPS I disease. The value of the continued support of people with MPS I, their families, and their physicians in providing both genetic and clinical data to the MPS I Registry cannot be underestimated in achieving this goal.

Box 1. Genetics terminology.
What is an MPS I phenotype? MPS I is usually categorised into three syndromes of increasing severity: Scheie (mild), Hurler-Scheie (intermediate) and Hurler (severe). These syndromes are sometimes called phenotypes. The term phenotype just means an observable or physical property of a patient.

What is a mutation? This is a change in the normal genetic sequence, and can sometimes cause a disease such as MPS I. A range of different mutations in the gene responsible for MPS I causes the range of clinical severities (phenotypes) of MPS I.

What is a genotype? This term refers to a patient's genetic type (genetic composition). Humans have two copies of each gene, one of which might contain a mutation and the other a normal sequence (or both normal, or both containing a mutation). A patient's genotype encompasses both copies.

What are amino acids? The IDUA enzyme is a complex three-dimensional protein, which is made up of building blocks called amino acids. Mutations in the gene may change an amino acid, which, in turn may alter the structure and function of the enzyme.

References

1. Scott HS, Ashton LJ, Eyret HJ et al. Chromosomal localization of the human a-L-iduronidase gene (IDUA) to 4p16.3. Am J Hum Genet 1990;47:802-807.
2. Moore D, Connock MJ, Wraith E et al. The prevalence of and survival in Mucopolysaccharidosis I: Hurler, Hurler-Scheie and Scheie syndromes in the UK. Orphanet J Rare Dis 2008;3:24.
3. Muenzer J, Wraith JE, Clarke LA. Mucopolysaccharidosis I: management and treatment guidelines. Pediatrics 2009;123(1):19-29.
4. Terlato NJ, Cox GF. Can mucopolysaccharidosis type I disease severity be predicted based on a patient's genotype? A comprehensive review of the literature. Genet Med 2003;5:286-94.
5. Pastores GM, Arn P, Beck M et al. The MPS I registry: design, methodology, and early findings of a global disease registry for monitoring patients with mucopolysaccharidosis Type I. Mol Genet Metab 2007;91(1):37-47.
6. Cox GF, Wraith JE, Whitley CB et al. Genotype frequencies in the MPS I Registry. Mol Genet Metab 2009;96:S19 (abstract 31).
7. Rojas MVM, Norato DYJ. The MPS I Registry: do patients in Brazil, Latin America and Worldwide share the same phenotype distribution? 10th International Symposium on MPS and Related Diseases, 26–29 June 2008. Vancouver, Canada.
8. Jin D-K, Wraith JE. Patients identified as "Asian" in the MPS I Registry: toward a global collaborative effort to understand MPS I phenotypic expression in Asia. Fourth Symposium on Lysosomal Storage Disorders; 29–31 March 2007. Vienna, Austria.

Declaration of interest: The author is employed by Genzyme Corporation.

6th Annual WORLD Symposium

We're Organising Research for Lysosomal Diseases

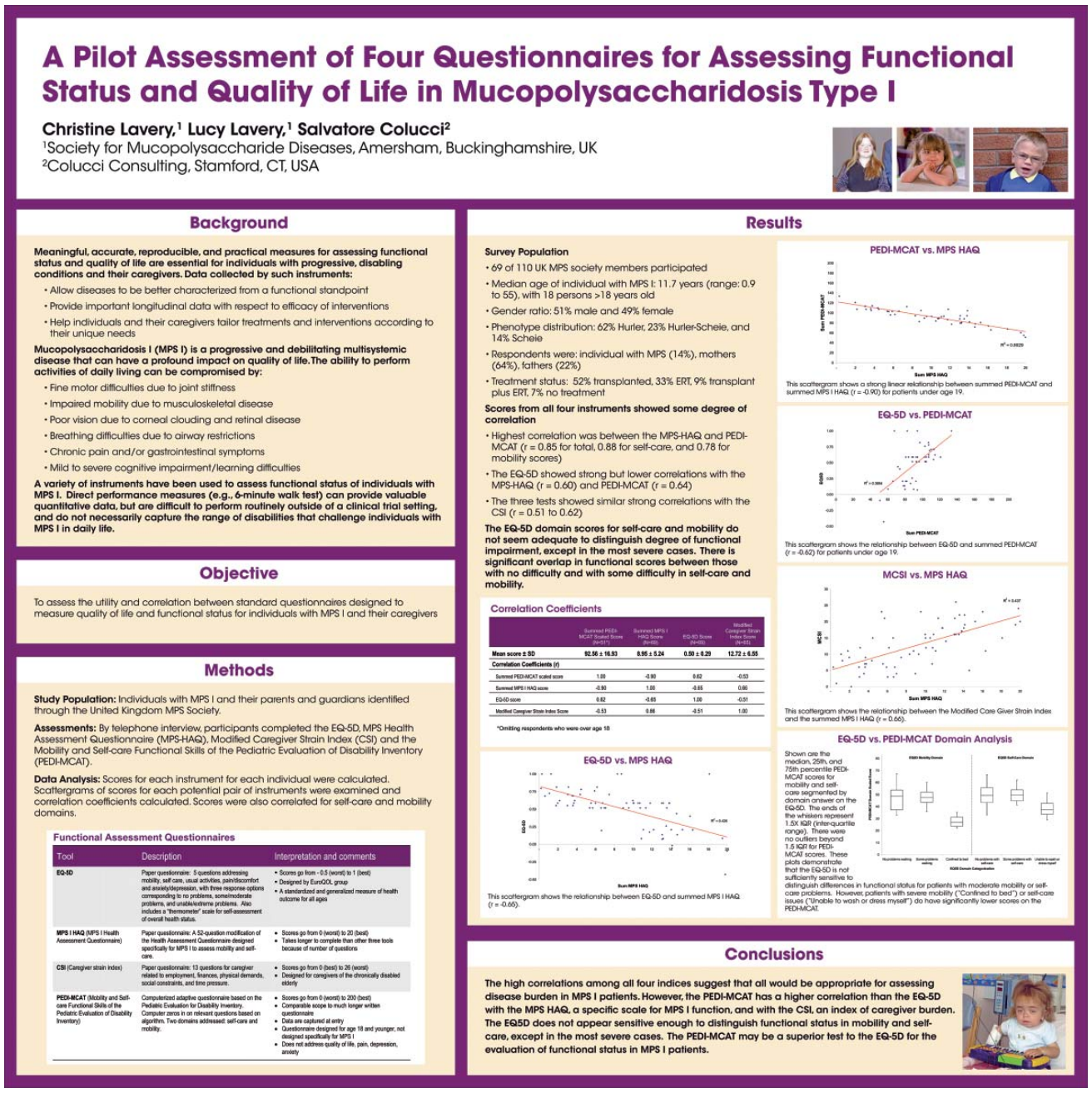
The Lysosomal Diseases Network (LDN) arose from competition for the National Institutes of Health (NIH) Rare Diseases Clinical Research Network (RDCRN) grants. Following on from three separate applications in 2003, a unified group emerged reorganised under the name Lysosomal Disease Network. This group has met annually since 2004 and the WORLD Symposium has become the major unifying activity of the LDN. It has evolved into a highly interactive international research activity involving scientists, academics, clinicians and patient organisations.

The first day of the WORLD Conference in Miami, Florida, 10-12 February 2010, focussed on basic research, the second day on translational research and the third on clinical research and outcomes. Ample time was allowed for poster sessions and for the first time the UK MPS Society had a poster accepted. The MPS poster 'A Pilot Assessment of Four Questionnaires for Assessing Functional Status and Quality of Life in Mucopolysaccharidoses Type I' is the result of the MPS I survey that so many of our MPS I members and their families participated in last Summer.

During the Symposium William Sly, the Alice A. Doisy Professor and Chairman of Biochemistry and Molecular Biology, St Louis University, was presented with the award for Innovation and Accomplishment in the field of lysosomal disease research and therapy.

Outside of the Symposium there was plenty to take in with over 150 posters to read, as well as meetings with the pharmaceutical industry. The LDN also organised a lunch meeting to bring all the patient organisations together for their common good.

Christine Lavery c.lavery@mpssociety.co.uk



A Pilot Assessment of Four Questionnaires for Assessing Functional Status and Quality of Life in Mucopolysaccharidosis Type I

Christine Lavery,[1] Lucy Lavery,[1] Salvatore Colucci[2]
[1]Society for Mucopolysaccharide Diseases, Amersham, Buckinghamshire, UK
[2]Colucci Consulting, Stamford, CT, USA

Background

Meaningful, accurate, reproducible, and practical measures for assessing functional status and quality of life are essential for individuals with progressive, disabling conditions and their caregivers. Data collected by such instruments:

- Allow diseases to be better characterized from a functional standpoint
- Provide important longitudinal data with respect to efficacy of interventions
- Help individuals and their caregivers tailor treatments and interventions according to their unique needs

Mucopolysaccharidosis I (MPS I) is a progressive and debilitating multisystemic disease that can have a profound impact on quality of life.The ability to perform activities of daily living can be compromised by:

- Fine motor difficulties due to joint stiffness
- Impaired mobility due to musculoskeletal disease
- Poor vision due to corneal clouding and retinal disease
- Breathing difficulties due to airway restrictions
- Chronic pain and/or gastrointestinal symptoms
- Mild to severe cognitive impairment/learning difficulties

A variety of instruments have been used to assess functional status of individuals with MPS I. Direct performance measures (e.g., 6-minute walk test) can provide valuable quantitative data, but are difficult to perform routinely outside of a clinical trial setting, and do not necessarily capture the range of disabilities that challenge individuals with MPS I in daily life.

Objective

To assess the utility and correlation between standard questionnaires designed to measure quality of life and functional status for individuals with MPS I and their caregivers

Methods

Study Population: Individuals with MPS I and their parents and guardians identified through the United Kingdom MPS Society.

Assessments: By telephone interview, participants completed the EQ-5D, MPS Health Assessment Questionnaire (MPS-HAQ), Modified Caregiver Strain Index (CSI) and the Mobility and Self-care Functional Skills of the Pediatric Evaluation of Disability Inventory (PEDI-MCAT).

Data Analysis: Scores for each instrument for each individual were calculated. Scattergrams of scores for each potential pair of instruments were examined and correlation coefficients calculated. Scores were also correlated for self-care and mobility domains.

Functional Assessment Questionnaires

Tool	Description	Interpretation and comments
EQ-5D	Paper questionnaire: 5 questions addressing mobility, self-care, usual activities, pain/discomfort and anxiety/depression, with three response options corresponding to no problems, some/moderate problems, and unable/extreme problems. Also includes a "thermometer" scale for self-assessment of overall health status.	• Scores go from - 0.5 (worst) to 1 (best) • Designed by EuroQOL group • A standardized and generalized measure of health outcome for all ages
MPS I HAQ (MPS I Health Assessment Questionnaire)	Paper questionnaire: A 52-question modification of the Health Assessment Questionnaire designed specifically for MPS I to assess mobility and self-care.	• Scores go from 0 (worst) to 20 (best) • Takes longer to complete than other three tools because of number of questions
CSI (Caregiver strain index)	Paper questionnaire: 13 questions for caregiver related to employment, finances, physical demands, social constraints, and time pressure.	• Scores go from 0 (best) to 26 (worst) • Designed for caregivers of the chronically disabled elderly
PEDI-MCAT (Mobility and Self-care Functional Skills of the Pediatric Evaluation of Disability Inventory)	Computerized adaptive questionnaire based on the Pediatric Evaluation for Disability Inventory. Computer zeros in on relevant questions based on algorithm. Two domains addressed: self-care and mobility.	• Scores go from 0 (worst) to 200 (best) • Comparable scope to much longer written questionnaire • Data are captured at entry • Questionnaire designed for age 18 and younger, not designed specifically for MPS I • Does not address quality of life, pain, depression, anxiety

Results

Survey Population

- 69 of 110 UK MPS society members participated
- Median age of individual with MPS I: 11.7 years (range: 0.9 to 55), with 18 persons >18 years old
- Gender ratio: 51% male and 49% female
- Phenotype distribution: 62% Hurler, 23% Hurler-Scheie, and 14% Scheie
- Respondents were: individual with MPS (14%), mothers (64%), fathers (22%)
- Treatment status: 52% transplanted, 33% ERT, 9% transplant plus ERT, 7% no treatment

Scores from all four instruments showed some degree of correlation

- Highest correlation was between the MPS-HAQ and PEDI-MCAT (r = 0.85 for total, 0.88 for self-care, and 0.78 for mobility scores)
- The EQ-5D showed strong but lower correlations with the MPS-HAQ (r = 0.60) and PEDI-MCAT (r = 0.64)
- The three tests showed similar strong correlations with the CSI (r = 0.51 to 0.62)

The EQ-5D domain scores for self-care and mobility do not seem adequate to distinguish degree of functional impairment, except in the most severe cases. There is significant overlap in functional scores between those with no difficulty and with some difficulty in self-care and mobility.

Correlation Coefficients

	Summed PEDI-MCAT Scaled Score (N=51*)	Summed MPS I HAQ Score (N=69)	EQ-5D Score (N=69)	Modified Caregiver Strain Index Score (N=65)
Mean score ± SD	**92.56 ± 16.93**	**8.95 ± 5.24**	**0.50 ± 0.29**	**12.72 ± 6.05**
Correlation Coefficients (r)				
Summed PEDI-MCAT scaled score	1.00	-0.90	0.62	-0.53
Summed MPS I HAQ score	-0.90	1.00	-0.65	0.66
EQ-5D score	0.62	-0.65	1.00	-0.51
Modified Caregiver Strain Index Score	-0.53	0.66	-0.51	1.00

*Omitting respondents who were over age 18

EQ-5D vs. MPS HAQ

This scattergram shows the relationship between EQ-5D and summed MPS I HAQ ($r = -0.65$).

PEDI-MCAT vs. MPS HAQ

This scattergram shows a strong linear relationship between summed PEDI-MCAT and summed MPS I HAQ ($r = -0.90$) for patients under age 19.

EQ-5D vs. PEDI-MCAT

This scattergram shows the relationship between EQ-5D and summed PEDI-MCAT ($r = -0.62$) for patients under age 19.

MCSI vs. MPS HAQ

This scattergram shows the relationship between the Modified Care Giver Strain Index and the summed MPS I HAQ ($r = 0.66$).

EQ-5D vs. PEDI-MCAT Domain Analysis

Shown are the median, 25th, and 75th percentile PEDI-MCAT scores for mobility and self-care segmented by domain answer on the EQ-5D. The ends of the whiskers represent 1.5X IQR (inter-quartile range). There were no outliers beyond 1.5 IQR for PEDI-MCAT scores. These plots demonstrate that the EQ-5D is not sufficiently sensitive to distinguish differences in functional status for patients with moderate mobility or self-care problems. However, patients with severe mobility ("Confined to bed") or self-care issues ("Unable to wash or dress myself") do have significantly lower scores on the PEDI-MCAT.

Conclusions

The high correlations among all four indices suggest that all would be appropriate for assessing disease burden in MPS I patients. However, the PEDI-MCAT has a higher correlation than the EQ-5D with the MPS HAQ, a specific scale for MPS I function, and with the CSI, an index of caregiver burden. The EQ5D does not appear sensitive enough to distinguish functional status in mobility and self-care, except in the most severe cases. The PEDI-MCAT may be a superior test to the EQ-5D for the evaluation of functional status in MPS I patients.

European Science Foundation Brains for Brain Exploratory Workshop on Treating Paediatric Neurodegenerative Diseases: from Laboratory Bench to Bedside

The European Science Foundation (ESF) is an association of 79 Member Organisations devoted to scientific research in 30 European countries. The mission of ESF is to provide a common platform in order to advance European research and to explore new directions at a European level. The exploratory workshop scheme aims to allow academics, scientists, clinicians and patient representatives to explore an emerging and/or innovative field of research.

The aim of the Brains for Brain Exploratory Workshop in Frankfurt, Germany, 3-5 March 2010, was to open up new directions in research related to its efforts towards the treatment of neurodegeneration associated with Lysosomal Storage Diseases (LSDs) and uniting the separate disciplines existing within Europe that are required to develop therapeutic treatment strategies for LSDs.

This two-day programme was convened by Prof Maurizio Scarpa of the University of Padova and Dr David Begley of Kings College London. It addressed the scientific basis and expected breakthroughs in neurodegenerative diseases for LSDs. It aimed to reach a consensus and produce a document discussing the need of supporting research for:

The early diagnosis of neurodegenerative disorders caused by storage of macromolecules and their pathophysiology

The study of the physiology of the blood brain barrier by creation of in vitro models, and analysis of in vivo models

The development of new strategies to cross the blood brain barrier as a major goal to achieve brain therapy and positive modification of the natural history of these lethal progressive disorders.

The workshop report will be published on the website of the group: www.brains4brain.eu
Christine Lavery c.lavery@mpssociety.co.uk

European Task Force on Brain & Neurodegenerative Lysosomal Storage Diseases

This meeting in Frankfurt, Germany, 5 - 7 March 2010, was opened with a plenary lecture on Lysosomal Storage Diseases: Beyond the State of the Art, given by Prof Timothy Cox of Addenbrookes Hospital, Cambridge, and focused on the clinical manifestations and future treatment options for Tay-Sachs disease.

The first two scientific sessions addressed the pathophysiology of the blood brain barrier in the Lysosomal Storage Diseases (LSDs) and the challenge of transporting lysosomal enzymes into the brain.

The third session dealt with the challenge of crossing the blood brain barrier and therapeutic options, and this was followed by presentations from Glaxo, Genzyme, BioMarin, Shire and ToBBB who presented on their work and biotech collaborations with Brains for Brain.

Unfortunately, I had to leave the meeting before it ended, so the patient organisation presentation was given by Tanya Collin-Histed representing the Gaucher Association. Tanya spoke of the very positive UK collaboration of LSD patient organisations and their preliminary plans to work together to raise vital funds for research that addresses paediatric neurodegenerative brain disease.
Christine Lavery c.lavery@mpssociety.co.uk

Expert Meeting on Sanfilippo Disease, MPS III

27 - 28 August 2010
Northampton Hilton

The MPS III Expert Meeting programme and booking form is available as a download from the MPS Society's website **www.mpssociety.co.uk.**

A Story of Love and Hope

by Julie Chou

Love is the expression of hope. To live is to love and to hope. Challenges of life come in different shapes and sizes. We deal with them by running away or embracing them. Facing reality, we get rid of the fundamental problems, leaving behind an empty heart. We then begin the search for miracles. The hunt is over when we send out our love.

It all hit me that day at the bus stop. I saw a mother holding hands with her child. She took him to the bus stop to go to school like everyone else. The bus came and she went up with him. They went to school together. He is eighteen and mentally challenged. I "can't" only imagine what she has been through, because our family underwent the exact same scene.

My elder brother David was diagnosed with a rare disease called MPS. He wasn't welcomed like any grandson should be because he was different. At first, it was hard for my parents to accept the fact. The compassionate anger my mom reveals whenever my brother did something wrong; the odd and uncomfortable stares people shot at us just during a walk outside in the park; and the twisted feeling my parents have for him were all like a tug-of-war in their hearts. Nevertheless, he is still a family member, and my parents loved him just the same. They were always ready for anything, except giving up. They went to see the best doctors in the country; they've been to temples and hope to seek an answer through religion; they've also gone overseas to search for doctors. Their love was endless, and they were willing to light a match in a rain storm just to find a clue of hope.

Unfortunately, my brother passed away and there still isn't a cure up to this day. They were shattered by the death of my brother. They didn't only lose a son, but they've lost their hope. My age helped me avoid the harsh fact, but thinking back now, the fear of loss still creeps and attacks me. Moreover, my parents had to live the fact. After my brother was sent away, there was this dead silence in the house. My parents felt something must be done.

I could feel their hearts were in need of a miracle. The tug-of-war competition was over. The rope has worn out. They started thinking and worrying about all the other parents whose children might be diagnosed with MPS. After years of hard work, they've established the "Taiwan MPS Society" and gathered up to 128 families in Taiwan. The Taiwan MPS Society has been founded for thirteen years up to now. The first ten years consisted of both my parents' perseverance. It's been three years since the sudden death of my father. This loss was even more devastating. Our whole family collapsed. Fear of death haunted me and followed me like a shadow. Nothing seemed meaningful or substantial anymore. There came a moment that I finally realised it was time, time to let go; time to realise there was still so much to cherish in life even if we suffered great losses. Bravery is not the absence of fear, but rather a soul of courage pursuing freedom and tranquility. And for my mother to pull herself together was even more so. My parents have truly inspired me, and taught me a memorable lesson. Care and love today, means hope and miracles tomorrow.

So that day at the bus stop, I decided to help the mother out. I found them a seat on the bus and gave her a warm smile. She smiled back, and I realised, miracles are right here in our hearts. We must learn to give before we can receive.

We can never fill up a glass that is already full. We must pour out what is in there to fill it with new things. Our hearts are filled with love, believe it or not. By expressing and sharing our love, we can bring hope to others, and more importantly, to ourselves. The challenges we face in life are a great chance to express love and plant a seed of hope for the future.

Photo left: Julie and Virginia Chou. Julie was a volunteer at the Taiwan MPS Conference in October 2009 and interpreted for her mother, Virginia.

News from Nobuhiro Kasa Vice President, Japanese MPS Society

I am happy to tell you about our news from the Japanese MPS Society. Mr Toyoshige Mikami, the President of the Japanese MPS Society and father of a boy with Hunter disease, is pleased to announce that the Society has diversified considerably to not only address the clinical problems but also the social matters experienced by our members.

Dr Tadao Orii, the Exeutive Head of the Japan Society for the Study of MPS and a special advisor of the Japanese MPS Society summarised at a recent meeting the therapies for MPS and explained the potential of chemical chaperone therapy as a new promising approach. Dr Akemi Tanaka of Osaka City University also presented the results of the post marketing surveillance study of the Enzyme Replacement Therapies (ERT) for MPS I, MPS II and MPS VI which have been in use over the past two or three years in Japan. At the meeting she pointed out that it has become clear that ERT has many advantages for improving quality of life for the patients. Dr Haruo Shintaku, also of Osaka City University, said "It is one of the most important achievements of the activities of the Japanese MPS Society that mucopolysaccharidoses were authorised as intractable diseases by the Japanese Government in 2001. The strategic and continuous lobbying activities of the Japanese Society are highly suggestive for the many other patient groups of paediatric chronic diseases".

Dr Yasuyuki Suzuki of Gifu University emphasised the importance of international co-operation and partnership for the acceleration of the development of new therapies. He finished by encouraging Japanese families to attend the International Symposium on MPS Diseases in Adelaide in June.

Finally, Ryu Ota (photo above right (who suffers from Hunter disease, won the prize of Genzyme's 'Expression of Hope' and was presented with his award at the meeting. A drawing by Sota Isomoto (photo bottom right), who suffers from Morquio disease, won a special prize of the 'Blue Sea Drawing Contest' and was presented with his prize by the Japan Coastguard. I am sure everyone would wish to join me in congratulating Ryu and Sota.

JEANS FOR GENES

Changing the world for children with genetic disorders

Friday 1st October 2010

Jeans for Genes is the national charity that holds Jeans for Genes Day. Jeans for Genes aim to provide funding for the care and support of children and their families affected by genetic disorders as well as funding research into the genes responsible and the development of effective treatments and cures.

Jeans for Genes Day takes place on the first Friday in October. This year's event takes place on Friday 1 October 2010. Wearing 'jeans' is a great reminder of the 'genes' that you're raising money for.

For more information on how you can help Jeans for Genes, support the campaign all year round and education and awareness opportunities visit **www.jeansforgenes.com**.

If you celebrate Jeans for Genes Day, please let us know how it goes!

INFORMATION EXCHANGE

New online benefits calculator

Contact a Family has launched Cash Counts, a new online service dedicated to ensuring families with disabled children are getting every penny.

Cash Counts includes:
- An online benefits calculator, allowing families to work out how much they are entitled to
- A top tips guide on the benefits available to families with disabled children written by the Contact a Family helpline
- A frequently asked questions page to help families navigate the complex benefits system.

Srabani Sen, Contact a Family Chief Executive, said: "Cash Counts gives families with disabled children the latest financial advice and information specific to them and all in one place."

"It costs three times more to raise a child with a disability, so families with disabled children are more likely to be living in poverty and be reliant on benefits. In these tough economic times, finances are tight for all families, but for those with a disabled child things are even harder. They need a helping hand to navigate the complex benefits system."

Families with disabled children face enormous financial challenges. They incur additional costs caring for their child – childcare, heating, transport, home adaptations and equipment. It can also be difficult for families to hold down a job due to the demands of caring. Despite this, many are not claiming the benefits they are entitled to.

Mum, Stephanie, said: "My husband recently stopped working because the strain of working and caring for Isaac was too much. It has meant we have had to tighten our purse strings. Isaac is doubly incontinent and this means clothes, bed linen and furniture need replacing far more frequently than is usual. And we have the washing machine on at least twice a day often doing a boil wash. This is not something we can reduce, so we have to incur the extra costs."

Srabani Sen added: "We would urge families with a disabled child to use Cash Counts or telephone the Contact a Family helpline to make sure they are getting every penny they are entitled to."
For more information get in touch with Contact A Family:

Free helpline 0808 808 3555
Email: helpline@cafamily.org.uk

FAMILY FUND UPDATE

From 1st April 2010, the Family Fund is changing how it looks at income. It has new income limits (split for different parts of the country) which are listed below and also has a new format for looking at the money coming into a house hold and whether the income meets their new income limits.

Income limits
- England £25,000
- Northern Ireland, Scotland and Wales £27,000

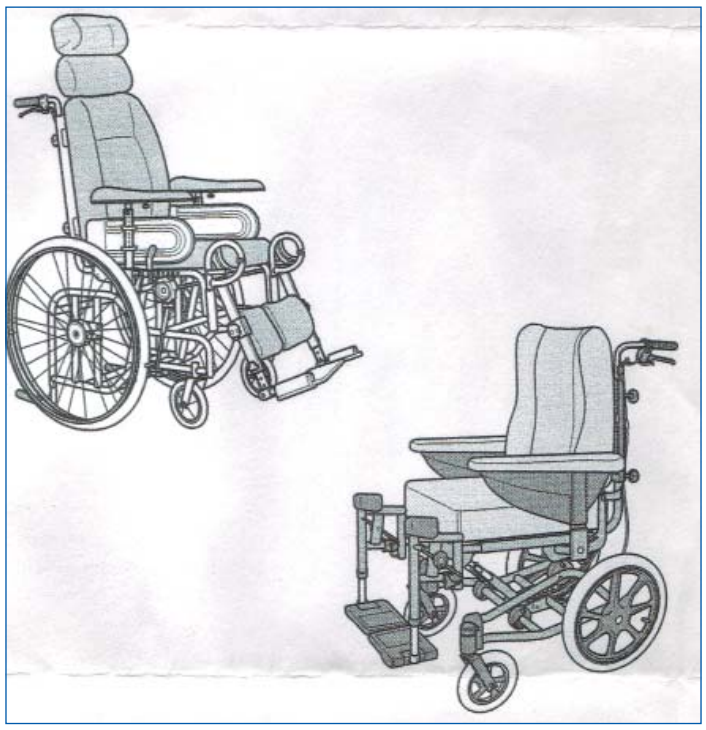
So what does this mean?
The Family Fund will now look at the total income coming into a household which will include; money from earnings, benefits, rental and maintenance income, occupational pensions, interest from savings and statutory payments. It will not include benefits such as Disability Living Allowance, Attendance Allowance or Child Benefit.

It is hoped that this new system will prevent working families being penalised and will ensure that the Family Fund is fair and accessible to all.

Second hand

Rea Comfort 97 Assist wheelchair

Available from a family in Kent
in return for a donation to the MPS Society.
Very good condition, two years old.
Please contact MPS for more information
0845 389 9901



Sleep Disorders in Children
Working party report available now!

The Royal College of Paediatrics and Child Health (RCPCH) have recently published a summary on sleep disorders in children summarising findings from the RCPCH Working Party. The full report containing recommendations (with evidence levels) is available to download electronically from the following website: http://www.rcpch.ac.uk/Research/CE/Guidelines-frontpage/Guideline-Appraisals-by-Organisation/RCPCH-Working-Party

There are also a number of patient leaflets dealing with different areas of the report available from this website.

The report makes recommendations in a number of important areas, including:

Obstructive sleep apnoea and adenotonsillectomy in children, including which high risk children should only have surgery in hospitals with paediatric intensive care facilities.

The ascertainment of sleep-related breathing problems in high risk children such as those with Down's syndrome, craniofacial abnormalities, neuromuscular diseases, Prader-Willi syndrome and mucopolysaccharidoses.

The assessment and management of infants with Apparent Life Threatening Events.

The assessment and management of narcolepsy and other causes of pathological sleepiness in children.

The organisation and quality control of services with some structures for peer review of services.

Hard copies of the summary can be obtained by emailing clinical.effectiveness@rcpch.ac.uk or calling 0207 092 6167.

Become a

Friend of MPS

Would you like to show your support by becoming a Friend of MPS? We would welcome relatives, friends, overseas MPS families, professionals or indeed anyone interested in the work of the Society or the field of MPS and Related Diseases.

This would encourage us, help us plan for the future and bring about more public awareness for this group of rare, genetic, life-limiting diseases. You can also keep up to date with the latest information, news and stories.

Visit **www.mpssociety.co.uk** to download the application or phone us now on **0845 389 9901**.

Challenging Behaviour Foundation
Newsletter available now!

'We all need champions', the Spring issue of 'Challenge' (newsletter of the Challenging Behaviour Foundation) is now available. This issue focuses on advocacy support for individuals with severe learning disabilities whose behaviour is described as challenging.

The newsletter highlights a new resource from the Challenging Behaviour Foundation designed specifically for advocates supporting individuals perceived as challenging. Other articles include 'Best interests decision making' by David Thompson (Practice Development Manager: IMCA, Social Care Institute for Excellence), and 'Comment' by Tony Osgood (University of Kent Tizard Centre).

Children and adults with learning disabilities and behaviour described as challenging are often a marginalised and significantly disadvantaged group of individuals. Independent advocacy is a very useful tool to empower and protect them. Ironically, it is those who are most in need of independent advocacy support who are least likely to access it.

Regular articles include 'your questions', 'what parents say' and an editorial by Vivien Cooper, Chair of Trustees and Founder of the Challenging Behaviour Foundation.

'Challenge', the newsletter of the Challenging Behaviour Foundation, is produced three times a year and is available free of charge by emailing: info@thecbf.org.uk or downloading from www.challengingbehaviour.org.uk

If you would like any further information then please do not hesitate to contact us:
The Challenging Behaviour Foundation
Email: info@thecbf.org.uk
www.challengingbehaviour.org.uk
General Enquiries: Tel. 01634 838739
The Challenging Behaviour Foundation is a registered charity (no. 1060714) supporting families caring for individuals with severe learning disabilities.

Get on in school, Get online at home

Home Access is a government drive that helps low-income families who currently lack access to a computer and/or internet to get online at home.

The programme is aimed at those that need it most. If you are a low-income family in receipt of certain benefits you could qualify for a grant to buy a computer and/or a minimum of one year's internet access.

If you think you are eligible for a Home Access Grant and would like to apply, please call the Home Access Grant Helpline to request an application form.

Home Access Grant Helpline: 0333 200 1004

E: enquiries@homeaccess.org.uk

RAISING AWARENESS

MPS Awareness Day

15 May 2010

One baby every eight days in the UK will be born with an MPS or related disease

Each year the Society celebrates International MPS Awareness Day on 15 May. This is a day devoted to raising awareness of MPS and Related Diseases

Help us celebrate International MPS Awareness Day on Saturday 15 May 2010

This year we're asking all our members, Friends and supporters to do something, big or small, to mark MPS Awareness Day

Download our Awareness Day flyer from **www.mpssociety.co.uk** or give us a call on **0845 389 9901** to find out more.

How your money helps...

More professional support for more MPS Families

MPS Advocacy Workers offer a whole range of services to help children and adults living with Fabry, Mucopolysaccharide and related diseases and support their families. We are there at the time of diagnosis and offer support for as long as we are needed. A donation of £2 per month could help us to offer so much more support in so many ways.

Access to expert clinical management & 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

More MPS advocacy workers

You'll be helping to fund more advocacy workers that are so crucial to empowering children and adults living with MPS and related diseases and their families through the information, advice and advocacy they provide.

More vital information

Your donation could help us to have more trained advisors running our MPS Helpline at the MPS Society's national resource centre. One child born every eight days in the United Kingdom will be diagnosed with an MPS or related disease.

More help to cope with the isolation of a rare disease

The chances are you have never heard of Mucopolysaccharide diseases, Mucolipidosis or Fabry disease. The truth is most of the families we support had never heard of these diseases either. That is why they need your help to enable MPS to provide national and regional family conferences, activity weekends for siblings, young adult weekends for those affected and run the MPS befriending scheme.

More noise to force through change

The MPS Society is already recognised for punching above its weight to achieve improved clinical care for all those affected, over half of whom will lose their lives in childhood. We campaign for change, we fight to eradicate discrimination and we aim to ensure that all affected children and adults get the health and social care whoever and wherever they are.

More help

Even if you don't know anyone living or dying with Fabry disease, a Mucopolysaccharide or a related lysosomal disease, your help is vital and enables us to help over 1200 affected families in the United Kingdom.

For more information, to seek support and advice from our advocacy team, or to help raise funds so we can continue our work, contact us now!

0845 389 9901 **mps@mpssociety.co.uk**