MPS

Spring 2009

Society for Mucopolysaccharide Diseases

www.mpssociety.co.uk

What's Inside…

INTERNATIONAL DATES for your diary!

MPS Awareness Day 2009

NEWS on Birmingham Children's Hospital LSD Service

… plus the LATEST in research, treatment, news and views

MPS National Weekend Conference

Early Bird Discount extended to **14 April 2009**

See inside for further details…

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

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

Front cover photo:
Jamie Topkul at the Northern Ireland Get Together (page 14)

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

Caroline Anderson
Advocacy Assistant
c.anderson@mpssociety.co.uk

Miriam Blowers
Volunteer & Event Co-ordinator
m.blowers@mpssociety.co.uk

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

Sarah Irvine
Advocacy Support Officer
s.irvine@mpssociety.co.uk

Andrew Jones
Human Resources Officer
a.jones@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

Magazine Deadlines

Summer	1 Jun 2009	Autumn	1 Sep 2009
Winter	1 Dec 2009	Spring	1 Mar 2010

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 2009

MPS National Weekend Conference

26-28 June 2009, Hilton Northampton

Early Bird Discount extended to 14 April 2009.

The full and informative programme includes:

- Thinking Ahead – from paediatrics to independent living
- MPS Neurological Diseases
- Living with Fabry Disease
- Research – Looking to the Future
- Bereaved Programme
- Gala Dinner
- Full children's childcare programme

A family of four consisting of 2 Adults/2 Children can attend the full weekend for £230. Please contact us now on 0845 389 9901 if you wish to discuss the cost for your family's requirements and make your booking. **Book now as there are only limited rooms available.**

We look forward to seeing you there.

A MESSAGE FROM THE CHAIRMAN

The Credit Crunch: Where is MPS?

Dear Members, Friends of MPS and supporters

In the Winter 2008 MPS Magazine our Chief Executive, Christine Lavery addressed the difficult financial times that so many of you may be dealing with or facing over the coming year and our thoughts are with you.

I am now writing to you in respect of the MPS Society's financial position. I am sure you will have heard through the media the loss of income some charities have suffered through the demise of the Icelandic Bank. Thankfully the MPS Society is not facing that challenge, however the MPS Society is experiencing a worryingly 'quiet' time when it comes to income from fundraising and donations.

Whilst this will not have any immediate impact on the level of support and activities provided to members in this financial year, if there isn't a substantial upturn in unrestricted income over the next seven months up until the next financial year we will face having to make substantial cuts that will inevitably affect the support and services offered to our members.

Facing such difficult times the Management Committee have already ensured that the MPS Society is running very lean with operational costs being cut to the bone whilst delivering a high level of support to our members.

I am pleased to announce that we have now appointed a full time Fundraising Officer, Kate Barker, who will start on 30 March. Kate's priority is to generate grant applications to underpin the support and advocacy service and provide encouragement and advice to our members, 'friends' and supporters in their fundraising efforts.

Regrettably, we can't beat the credit crunch and recession without your help. We rely on you to please tell your family, friends and acquaintances, indeed anyone about the invaluable work of the MPS Society and ask them to support us.

Thank you and 'Every Penny Counts'.

Barry Wilson

Chairman of Trustees of the MPS Society

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**.

Spring 2009

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 Mucopolysaccharde diseases, Mucolipidosis or Fabry disease. The truth is most of the families we support have 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.

Would you like to volunteer for MPS?

Volunteering is fun and rewarding. It could also help you learn new skills and gain valuable work experience. The MPS Society relies on volunteers for our events and conferences to assist in the care needed for children and young adults affected by MPS and Related Diseases.

All of our volunteers undertake training in moving and handling and are fully briefed prior to the event. Volunteers should be 16 years or over, will need to provide two references and undergo a Criminal Records Bureau check and attend a training day in Amersham. Those volunteering for our conferences will receive accommodation and all meals throughout the weekend.

Contact us now to register your interest and availability.

Highlights from the Management Committee

The Society's Board of Trustees meet regularly. Here is a summary of the main issues that were discussed and agreed at the Management Committee Meeting held on 30-31 January 2009.

PERSONNEL

Trustees were advised that following a second round of interviews, Andrew Jones was appointed part-time to the post of Human Resources Officer. In respect of the Fundraising Officer post, a round of interviews is being planned for two new candidates.

GOVERNANCE

Due to the demand on time at this meeting Trustees agreed that they would defer review of policies to the next Management Committee along with a major review of the Society's Risk Management Register and Business Continuity Plan.

TREASURER'S REPORT

The Treasurer reported on the Society's income and expenditure. It was confirmed that the Society was benefiting from falls in the Bank of England interest rate in respect of the Society's mortgage on MPS House. It was noted that the Society is encountering a considerable reduction of income and the CEO spoke of her priority area of securing funds for the next financial year to underpin the salaries of the current staff team.

GENERATING INCOME

It was agreed that the Society's fundraising consultant continue her one day a week until the fundraising officer post is filled. Trustees agreed that the MPS Members need to be made aware of the Society's continuing slow down in income from fundraising and donations. The Finance Officer informed Trustees that members are taking up fundraising opportunities advertised in the MPS Magazine and that one way forward in the current economic climate is the 'feel good and fun factor' of fundraising events.

SUPPORT TO MEMBERS

The Senior Advocacy Officer's report giving an overview of the range and scale of work currently being undertaken by the team was discussed. Feedback was given to Trustees on the planned events for 2009 including the Northern Ireland Get-Together, MPS Conference Weekend, Childhood Remembrance Day, Sibling Weekend, Scottish Respite Weekend and the Disney Conference for which restricted funding has been received.

MPS RESEARCH GRANTS

The Chief Executive confirmed that agreement has been reached that the £70,000 for research raised through the 'Ollie G Ball' be divided between the MPS Stem Cell Group at the University of Manchester and the MPS Blood Brain Barrier Group at Kings College, London for two important projects.

The Chief Executive spoke to the peer-reviewed application from Prof. Maurizio Scarpa for a novel study of MPS VI in zebra fish. Despite the merits of this research to be supported, Trustees agreed that due to the greatly reduced income from Jeans for Genes for MPS Research they could not offer a grant at this time.

MPS Annual General Meeting 2009

The 2009 Annual General Meeting of the Society for Mucopolysaccharide Diseases will be held at the **Hilton Hotel, Northampton at 9am on Sunday 28 June 2009.**

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

New faces at MPS...

My name is **Jolanta Turz** and I started with the MPS Society on 26 January 2009. The team is great and I have been made to feel very welcome from the very beginning.

I am a psychology graduate. At my previous job I used to work with vulnerable adults who suffer from epilepsy. In my spare time I like learning languages, going to the cinema or doing mountaineering.

I am looking forward to meeting the MPS members and working with you in the future.

Jolanta Turz
j.turz@mpssociety.co.uk

Hi! My name is **Andrew Jones,** commonly know as Andy, from the Heads of the Welsh Valleys, Ebbw Vale.

I have now been with the MPS Society since 12 January 2009. My role is as part time HR Officer. I would like to take this opportunity to thank all the staff at MPS House for making me feel very welcome and always having time not just for me but for each other which includes the MPS sufferers and their families. It's a credit to MPS and long may it continue.

With the absence of my previous counterpart I have had to knuckle down and learn new areas, one being the CRB. Again I have to say this is also achieved through the helpfulness of the staff, thank you. My next big challenge will be attending the Trustees Meeting on 27th and 28th March 2009. It is here that I will talk about the current Risk Register and Business Continuity Planning (BCP) and of course have the opportunity to meet all the Trustees in person.

I have spent the past 15 years in an HR role and I absolutely enjoy every minute, enjoying the day to day challenges in personnel along with the compliance with Employment Law Legislation. I have recently started an Employment Law Degree with the CIPD and I expect to qualify in approximately two years having completed all four modules.

Passionate about rugby, I have played the game and retired at the age of 35

having suffered a serious injury. I went on to coach and completed my level one coaching with the Welsh Rugby Union, coached at London Welsh and now I am currently coaching the U16's at High Wycombe Rugby Club.
Andrew Jones
a.jones@mpssociety.co.uk

My name is **Sarah Irvine** and I started with the MPS Society on 2nd February 2009. It is a great team here and I have been made to feel very welcome.

My background covers the police and prison service, and more recently support work with vulnerable adults in their own home. Much of my spare time is covered with Open University study, but I also enjoy films, music, watching football and photography.

I am looking forward to working with as many of you as I can in the future.

Sarah Irvine
s.irvine@mpssociety.co.uk

MPS Awareness Day
Friday 15 May 2009

Members' Announcements

New Members

Ms Elizabeth Silcock and Mr Thomas Gill have recently been in contact with the Society. Their son Bobby has been diagnosed with Sanfilippo Disease. Bobby is four years old. The family live in the South East.

Ms Terry Ann Cornwall has recently been in contact with the Society. Her daughter Chloe is 2 and half years old and has a diagnosis of Maroteaux Lamy Disease. The family live in the South East.

You are important to us, please keep in touch.
Please remember to let us know if you are moving and your let us have your new address and telephone number. In addition to helping keep our printing costs to a minimum, you will help us keep our database up to date so that we can provide the best possible support to you and your family.

Deaths

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

Hannah Chisling who died on 9 January 2009 aged 16 years. Hannah suffered from Sanfilippo Disease.

Stacey Cliff who died on 22 January 2009 aged 22 years. Stacey suffered from Sanfilippo Disease.

Sarah Cupitt who died on 27 January 2009 aged 12 years. Sarah suffered from Sanfilippo Disease.

Aasiya Allana who died on 1 February 2009 aged 13 years. Aasiya suffered from Hurler Scheie Disease.

Lisa Stewart who died on 5 February 2009 aged 13 years. Lisa suffered from Sanfilippo Disease.

Ramzam Begum who died on 26 February 2009 aged 13 years. Ramzam suffered from Morquio Disease.

Natalie Pidden who died on 10 March 2009 aged 28 years. Natalie suffered from Sanfilippo Disease.

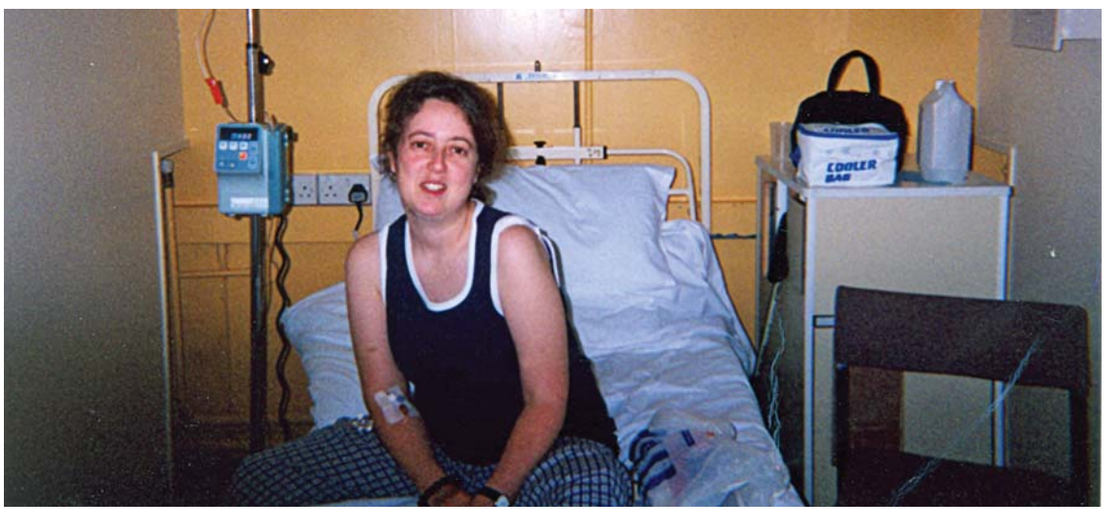
Claire Rowland

30 May 1970 – 12 December 2008

As a child Claire was full of life dragging me off to dancing lessons, gymnastics, riding lessons, I could go on. Life began to change for her in her teens and eventually she was diagnosed with Fabry disease in her late twenties. The Enzyme Replacement Therapy was a blessing and Claire continued with her life, working and playing hard.

Claire never moaned or complained; she just got on with her life. She always had a smile for everyone. The last almost three years were very difficult and Claire died very suddenly of a heart attack while in hospital.

Linda Rowland (Claire's Mum)



In memory - Lisa Louise Stewart

It is with great sadness that we write to inform you that our precious Lisa Louise passed away on February 5th.

She had been in hospital for four weeks with breathing problems and everyone was very optimistic that she would recover. However, on Tuesday 3rd she was given a GA for the pulmonary specialist to put a scope down and see what the problem was. Unfortunately her airway was "compromised" and collapsing in on itself when she inhaled and she would be unable to breathe without a ventilator. We made the heartwrenching decision that this would be no quality of life for our darling girl and with the help of Brisbane Children's Hospital ICU and her consultant, Jim McGill, we were able to bring her home on Thursday morning, where we all cuddled her in her own bed as she peacefully passed away. Her struggle is over and she doesn't have to fight any more as in the final weeks.

In no way are we religious people, but we desperately hope that there is a heaven, because if anyone deserves to be there, chuckling and prancing and dancing around, it is our precious little angel.

We would like to thank you for all your help during the years we lived in England. We really wouldn't have coped without you.

Here are a couple of photos of Lisa, one with Christine (Lavery) in Brisbane. Ja**ckie, Dick and DJ Stewart**

Birthday Congratulations

We wish to send our best wishes and congratulations to the following individuals who recently celebrated birthdays:

40th

Daryll Westland celebrated his 40th birthday on 19 January 2009. Daryll has Sanfilippo Disease.

30th

Helen Skidmore celebrated her 30th birthday on 14 March 2009. Helen has Hurler Disease and underwent a Bone Marrow Transplant.

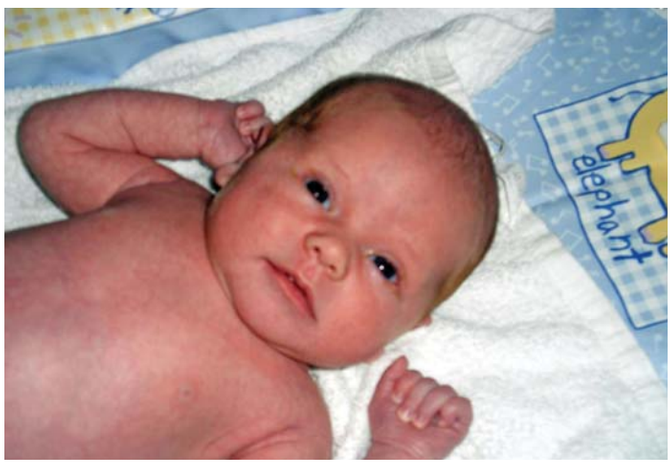
21st

Matthew Davenport celebrated his 21st birthday on 10 January 2009. Matthew has Fabry Disease.

Jo Farrow celebrated her 21st birthday on 16 January 2009. Jo has Scheie Disease.

We are delighted to announce that Jack Andrew Robertson was born on 18 November 2008. He weighed in at 7lbs 11oz. In June 2007 our daughter Jess, who suffered from Hurler Disease, passed away, but Jack is Hurler free. Hope you are all well at MPS House.

Steven Robertson and Emma Morrice



WHAT'S ON 2009!

MPS Awareness Day
Friday 15 May 2009

MPS CLINICS

1 May	Manchester BMT clinic (over 6 yrs)
14 May	Northern Ireland MPS Clinic
19 May	Bristol MPS Clinic
21 May	GOSH MPS III Clinic
22 May	Manchester BMT clinic (under 6 yrs)
12 June	Birmingham MPS Clinic
17 July	Manchester BMT Clinic (under 6 yrs)
24 July	BMT Clinic (over 6 yrs)
25 August	Bristol MPS Clinic
16 October	Manchester BMT Clinic (under 6 yrs)
23 October	Manchester BMT Clinic (over 6 yrs)
10 November	Bristol MPS Clinic
19 November	Northern Ireland MPS Clinic
20 November	Birmingham MPS Clinic
TBC	Cardiff MPS Clinic

CONFERENCE EVENTS

26 - 28 June	MPS National Weekend Conference
17 - 21 Dec	MPS Disney Conference Orlando

REGIONAL EVENTS

15 May	MPS Awareness Day
27 June	Childhood Wood Remembrance Day
14 - 17 August	Sibling Weekend
2 October	Jeans for Genes Day
23 October	Childhood Wood Planting

MPS Disney Conference, Orlando

17 - 21 December 2009

Check out the flyer for more information

Date for your Diary!

Mark your calendars and hop down for the

11th International Symposium on Mucopolysaccharide and Related Diseases

Adelaide, South Australia, 23 – 27 June 2010

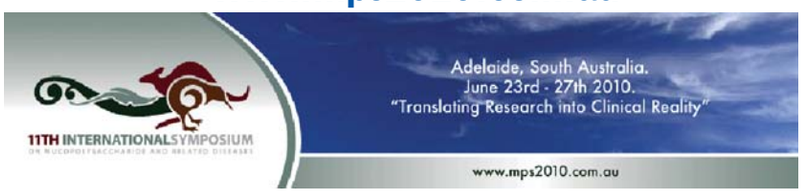
Mucopolysaccharide and Related Diseases Society Aust. Ltd., Lysosomal Diseases Australia and Lysosomal Diseases New Zealand warmly invite you to join them in Adelaide, 23 – 27 June 2010 for the 11th International Symposium on MPS and Related Diseases.

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.

www.mps2010.com.au



Coping with a three day weekend

Or how to fit five days of education into four days at school.

My son, Ben Cooper, has Hunter Disease and has been on the enzyme replacement therapy since April 2007. After the first three months of weekly trips to Royal Manchester Children's Hospital, we were able to have the treatment at home and quickly settled into the weekly routine of a Monday visit by the nurse for Ben's infusion. Mondays are seen differently by each of us.

Initially Ben was apprehensive about the process, but surprisingly quickly he has adapted to being 'hooked up' for hours to the drip. My wife Jackie sees Mondays as the most stressful part of the week, whilst I am the lucky one, usually at work, usually otherwise involved and able to simply view Mondays as a day of hope, the day that my son gets the treatment that will give him a healthier and longer life than he otherwise might have expected.

The enzyme replacement therapy has meant a number of changes to our life. At one extreme there is the tuppaware box in the refridgerator containing a small fortune of treatment vials of enzyme and the frequent checking of the digital thermometer to make sure that they are kept at just the right temperature. There is the ritual of the getting out the treatment box on a Sunday night and tidying the dining table ready for the Nurse to use on Monday. At the other extreme there is the reality of 'Ben not doing Mondays' at school.

Ben is fortunate in being a pupil at Longcroft School in Beverley and Longcroft are fortunate in having Ben as a pupil, as both school and pupil seem to get along very well.

When Ben moved to Longcroft nearly three years ago we had all of the usual fears about fitting in and getting on and then there were the added pressures of taking a day out each week for treatment and the impact this would have on Ben's education. In spite of our fears though, Ben's education is going very well and this is in a large part due to Ben's determination and the school's flexibility.

Ben is part of the Pathfinder group at his school. This is a new programme that seeks to 'fast track' pupils through key GCSE subjects by compressing the three pre GCSE years into only two, starting GCSE's in Science, Maths and English a year early. Although Ben is only just 14 he has been doing GCSE's for nearly a year now and getting good grades.

Ben's contribution to this success is his willingness to work hard and his determination to do his best and not use Monday's as an excuse for being middle of the road. The school's contribution is to try and produce a timetable that minimises absences from key subjects and to allow Ben to miss games and PE classes and use that time for self study. The school has an active programme of Clubs and Ben misses no opportunity to participate in a lunchtime activity whether it is Latin Club, Warhammer Club or Theatre Workshop. The clubs, teachers and general community of the school have helped Ben to become more confident and mature over the last few years, something that my wife and I are very thankful for. Ben's hard work and determination was recognised earlier this year at Hull University when he was awarded the school's Peter Johnson Award for his determination and hard work.

Weekends, particularly Sundays, are a time for further study and revision for Ben, sometimes with my help and sometimes on his own. I had thought that Ben would spend much of Monday studying, particularly after things had become more routine. But, this was not to be as our usual nurse, Suzanne, and Ben just love to chat, watch Car Booty and Homes under the Hammer. When the two of them are together no-one else can get a word in. Occasionally, when Suzanne hasn't been able to come, one of her colleagues fills in and each, in their own special way, are able to fill the Monday with something interesting and entertaining.

While we don't know what the future holds we can but hope that it continues as it has with Ben and school working well together and Mondays full of card games, enzymes and car booty!

Editor's Note: We were very sorry to hear that, since this article was written, Suzanne Gowler, Ben's nurse, sadly passed away on 9 March 2009.

The photo below is of Ben receiving a trophy from Gervaise Phinn. Ben got his award at school presentation evening for 'Determination'. It was a very proud moment for us. **Lee and Jackie Cooper**

Gervaise Phinn, well known Yorkshire author and public speaker, presents Ben with his award at Hull University.

Life with Sophie

I am Sally, married to Tim with children Will and Sophie who has Sanfilippo disease. I wrote an article about two years ago when Sophie had a gastrostomy. When I wrote that piece I wanted to talk about how apprehensive I was about her having the gastrostomy but what a positive experience it has turned out to be and how Sophie had benefited from having the operation.

Now, further down the line, I know just how important it was for Sophie to have the operation when she did. She has been nil by mouth for a year and a half now and her hair and skin look great, and she is much healthier generally. Looking back she must have been quite dehydrated when she was still eating and drinking as the difference in her now is quite amazing and was obvious soon after the gastrostomy was performed.

Sadly, Sophie's condition has worsened and last year her mobility took a real turn for the worse. Last January she could walk with help from her bedroom to the lounge (both on the ground floor) and although it wasn't a brisk walk it certainly wasn't difficult for her. Sadly, now she is unable to walk and can barely weight bear. It has happened very quickly and it is very hard to accept and get used to.

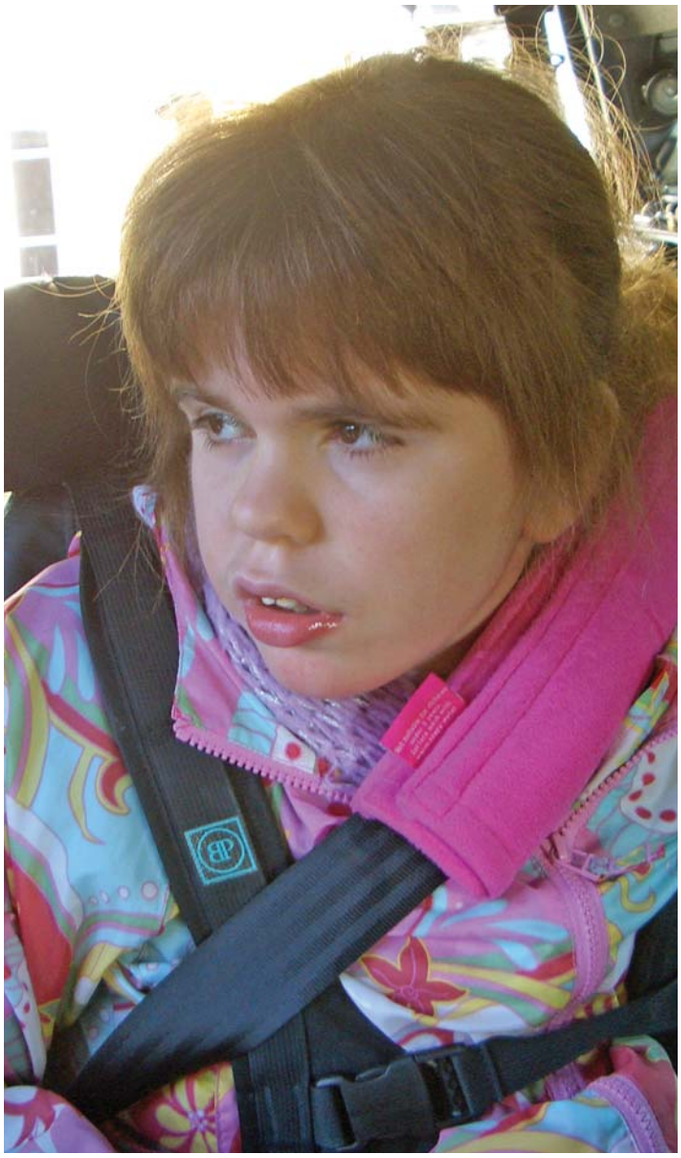
Sophie's lack of mobility has meant big changes for all of us. We have had to buy an adapted vehicle which was something we had wanted to avoid. For Will especially it has been hard having an adapted car on the drive and dealing with questions from his school friends (luckily so far, all remarks have been very positive and he is very pleased with our 'Truck' as it is known!). For us we have experienced another major change to our lives and have had to get used to getting Sophie in her wheelchair and in and out of the "truck". I found it a massive challenge to learn to drive it as I had never driven anything much bigger than a Fiesta! However, it was not as scary as I thought it would be and I now enjoy driving it!

One of the very positive aspects of the 'Truck' is that Sophie can sit in her wheelchair and look out of the window and see everything. It is so much better for her and she loves it! We have almost perfected the art of loading and unloading and like everything, it soon becomes the norm.

Although it means having to accept yet another stage of this cruel disease, having an adapted vehicle does have benefits. It becomes easier to transport your child and for me, not having to lift Sophie in and out of the car which was much lower, has eased the strain on my back.

I miss Sophie's walking ability dreadfully and not being able to run around and dance with her makes me so sad. However, looking at Sophie I truly believe she is a very happy girl and we have to enjoy all the things she can do. She goes horse riding every Saturday, swimming every week and does rebound therapy (trampolining!) which she loves.

Here are two photos of Sophie. The photo on the left was taken when she was four and very active and the one on this page is a recent one in her more restful state. Thankfully she looks very happy in both of them.
Sally Summerton



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

Northern Ireland Family Get-Together and Presentation of Ollie G Wishes

Fun was had by all who attended the Northern Ireland Family Get-Together which was held on Saturday 7th February at the Hilton Templepatrick hotel.

The MPS Society organised this event so that families from throughout Northern Ireland had a chance to meet up with each other, and also to hold a presentation of Ollie G Wishes. During 2008 an Ollie G Ball fundraising event was held and MPS Society families were invited to apply for Wishes on behalf of their children. Six children from Northern Ireland who applied for an Ollie G Ball Wish had their Wish granted from the funds raised, so this Get-Together was a lovely opportunity for those children to receive their Wishes.

MPS staff Christine Lavery, Sophie Thomas and Sue Cotterell flew into Belfast the evening before, from a very snowy Heathrow. We were worried that the heavy snow might mean the event would have to be cancelled, so we were very glad that the flight was able to go ahead after some de-icing of the plane.

In Belfast the weather was not quite so bad, so luckily none of the families had trouble travelling to the hotel on the Saturday. At midday the families started to arrive, and soon there were 14 families including 18 children. Dr Fiona Stewart and her 2 children were also there, along with her colleague Dr Joanne Hughes and her daughters.

Christine Lavery opened the event with a few words of welcome, and then everyone enjoyed a buffet lunch. Towards the end of the meal Captain Franko, the children's entertainer, made his entrance. He went around the tables making balloon animals, flowers, hats and even swords! The children (and some grown-ups too) were delighted with their balloon models.

After the meal, Sophie Thomas gave a short talk about the support which the Advocacy team offers to all MPS families throughout the UK, including all our member families across Northern Ireland. Dr Fiona Stewart then spoke about the work of the MPS Society in Northern Ireland from the early 1980's up to the present day, and about the Wishes which had been purchased by the MPS Society with funds raised at the Ollie G Ball last summer.

Then Dr Stewart presented each child's Wish. Hannah Shannon, Brooke Harvey, Clare McDonagh and Jade McAfee each received a box full of wonderful sensory toys for them to enjoy. Aisling Hawkins received a special communication board. A Wish had been purchased for Santana McDonagh but she could not be present on the day as she was unable to travel. Her Wish, of a playmat, will be delivered to her separately.

Jade, Aisling, Clare, Brooke and Hannah were soon enjoying trying out their new toys and equipment – with some help from their brothers and sisters, and Mums and Dads!

Then Captain Franko started his magic show. This included tricks with his "pet dog", magic linking hoops, and a very silly magic wand which just wouldn't do things right in the hands of the children who assisted him. The finale was pouring a jug full of milk into the ear of one young assistant, then setting the cone of milk alight, magically making it disappear!

After the show it was time to start heading home. Fortunately there had been no more snow during the day so everyone could get home safely.

It was lovely to see so many of our Northern Ireland families at this Get-Together, and there was a great atmosphere as everyone had a chance to chat and have fun together. Families met up with old friends and made some new ones.

We look forward to future events in Northern Ireland, and also hope that many Northern Irish families will make the journey across to Northampton for the National MPS Conference in June this year. **Sue Cotterell**

News on the Ollie G Wishes 2008

Here are some of the lucky recipients of the Ollie G Wishes 2008.
Each of the families would like to convey their appreciation and gratitude to those who organised the Ollie G Ball 2008 and all the guests on the tables who pledged their support. A big thankyou!

Zara Watson was thrilled with her new television which will take pride of place in her bedroom.

I would like to say thank you to the Ollie G Ball for making my wish come true. I am on my laptop every day talking to lots of people I know on Facebook. Thank you again.
Amy Pain (MPS I)

Dominic Nowland Wall loves playing on his Wii and was thrilled with his new games.

I am writing to express our sincere gratitude for the items we received at Christmas from the Ollie G Fund.

The playmats are a must for outdoor play in the summer as Isaac is so boisterous now. I never thought I would be glad to say that about a child! He loves the motorbike, red just like his Dad's, and has become a bit of stunt man on it. Isaac has just clicked with swings and I can see him on his new one all summer.

The toys are all thoughtful and safe so appreciated. Thank you all for adding to a great Christmas at home.

Joe, Oni, Lee, Tom and Isaac Keighley

Hannah's Wish

We went to the Northern Ireland MPS family get together in February. We had to, because the MPS Society were giving our daughter Hannah her wish from the Ollie G Ball.

We never want to go to MPS events. We seem to spend so much of our time only just staying afloat on the sea of emotions that comes with having a little girl like Hannah with Sanfilippo, that to go to one of these events feels like stirring it all up. Because you forget, on good days, you forget all about the disability and the prognosis and all the problems that lie ahead, and the things you've already come through. On good days she's just your chuckling, smiling little girl. Sure, she can't walk, she can't talk, she can't feed or clothe herself. Toilet? What's that? Sometimes all that goes away when you're raspberrying her tummy and she's giggling uncontrollably, and you just smile yourself.

Then there are other times, when you're knocked so low by the weight and sheer exhaustion of it all, that when just one more thing looms up in front of you, you have just no clue how to carry on.

MPS clinics and conferences and get togethers sometimes bring you back down to earth, sometimes they lift you up off the floor. We always dread them before we go, but by the time we leave we are always so pleased to have gone, happy for the time with other families, for the camaraderie and solidarity and the happy positive spirit that permeates these events. We always leave fortified, refreshed to go on a bit further.

So we went to the Hilton, excited and scared as usual, and this time Hannah was to receive a present from the Ollie G Ball. We, of course, had to make Hannah's wish for her, and she was presented with a box full of sensory toys. We thought it was great, but knowing Hannah like I do, I know her first thought was, 'Yes Daddy, that's all well and good, but can I chew it? Can I chew it?' Well, yes Hannah, you can. Because those good people from the Ollie G Ball and the MPS Society know all about little girls and boys like Hannah with Sanfilippo and they know that they like nothing better that a good chewy toy, something that you can get your teeth into.

So the first thing out of the box was a good strong, safe rubberised ring that Hannah can chew on and grip. She munched on it for the rest of the function, watching the funny clown entertainer. There were lots of sturdy bits and pieces that made noises and had lights that grabbed Hannah's attention when we got home, and a crazy ball thing that jumped around on the floor, lights flashing and singing Olé! Olé! Olé! Hannah thought it was hilarious. And other things for when she was more reflective and still, chill out music and sounds for quieter times. All good stuff for Hannah. It is a constant search finding things that are suitable for a child with Sanfilippo, thanks MPS Society. Thanks Ollie G Ball. **Steven Shannon**

News from
Birmingham Children's Hospital LSD Service

Philippa Turner
Business Manager, Clinical IMD Service

I joined the metabolic team at Birmingham Children's Hospital in May 2007 as Business Manager. The Trust had just received NCG designation for the paediatric Lysosomal Storage Disorders (LSD) service which was at the early stages of development and the first patients were being accepted for treatment. From the very beginning, I became involved in the development of the service: securing resources and setting up recording and monitoring mechanisms.

I act as the commissioning interface between NCG and the Trust, notifying them of new patients accepted for treatment and changes in drug dosages and providing them with monthly summaries. I manage the budgets for the service and I am responsible for drawing up business cases to secure additional investment and funding to enable the service to develop further.

Over the last 2 years we have made major progress in developing and implementing a joint adult IMD service in conjunction with a neighbouring acute hospital trust so that adolescents and adults in the West Midlands with inherited metabolic disorders (IMD) now have a service in an appropriate care environment to move on to, which was not available in the past. In addition the metabolic service will shortly be relocating to a new-build unit which will enable all members of the multi-disciplinary IMD team to work more effectively as a team from a centralised base. I consider myself fortunate to be in a unique, dedicated role to support a rapidly developing specialised service and to have contributed to major service improvements for the benefit of IMD patients.

Psychological Care in MPS

Patients with MPS should soon benefit from improved psychological care at Birmingham Children's Hospital. So says Dr Anupama Iyer, Consultant in Adolescent Developmental Neuropsychiatry.

'The specific behaviours associated with certain MPS subtypes (behaviour phenotypes) remains an under-researched area. We hope to be collating more information about these from the young people and their parents in our effort to develop informed intervention strategies. We will also look towards supporting the team in enhancing communication with the young people and their families.'

'I am looking towards developing a psychiatric consultation liaison service to meet the needs of young persons with MPS. We will be assessing strengths and resilience of young persons and their families, especially in times of stress, with a view to enhancing proactive coping strategies. That way, we can address issues of treatment compliance and overt psychiatric illness as they may arise.'

Birmingham Children's Hospital IMD Team

Back row, (L-R): Jenny Hutchinson, Emma Scobie, Saikat Santra, Chris Hendriksz, Anupam Chakrapani, Suresh Vijay, Satnam Chahal, Liz Wright, Paul Gissen.
Front row: Phillippa Turner, Victoria Riches, Catherine Little, Rosie Jones

Dental Care in MPS

Alison Hutton is the Specialist Registrar in Paediatric Dentistry at Birmingham Children's Hospital, and knows the needs of children with MPS only too well:

'Oral health is key to an individual's quality of life, affecting the level of nutrition and contributing to social well being. Different mucopolysaccharide (MPS) diseases can affect the developing dentition in various ways. The MPS child is therefore not only at risk (like everyone else) of tooth decay and gum disease but can also have other complications of oral health depending on the type of MPS disease they have.

'Working with the MPS team is extremely challenging and rewarding. It enables me to review and examine the children, identifying any oral health problems early, reinforcing oral hygiene measures and providing information about preventing decay specific to individual families. Where necessary we can organise or provide dental treatment. The MPS team are also carrying out a research project into the specific oral health problems MPS children and adolescents can suffer from. This will enhance our knowledge and enable us to provide a better oral heath service in the future.'

Special Diet, Special Person: Rosie Jones

The special dietary needs of children with lysosomal storage disease are well catered for at Birmingham Children's Hospital. Rosie Jones is the Specialist Paediatric Dietitian for LSD Disorders at the Hospital:

'I am delighted to be in the unique position of having the post, working within such an experienced multi-disciplinary team. My role enables me to work closely with families to provide advice and support on various nutritional issues such as optimising intake, dietary supplementation, texture modification and home enteral feeding.

'I liaise with community specialist nurses, Health Visitors, Speech and Language therapists, schools, and the respite centre. I am looking forward to developing the service to further meet the needs of the children and their families. For example, we

have a newly appointed Speech and Language Therapist, Emma Scobie, and we are looking at ways of combining our expertise to manage the feeding difficulties some children experience. Emma's job is to work only with LSD patients, so we have a unique position for our patients by having the joint expertise.'

Birmingham IMD Team Win World Conference

The Inherited Metabolic Diseases (IMD) Team at Birmingham Children's Hospital (BCH) are hosting a major conference including the latest research on MPS disorders. The 'SSIEM' conference was founded in 1963 to get those involved in rare metabolic disease to work together. This has grown to include members from all over the world and is now annually the biggest meeting of metabolic experts anywhere: and in 2012 it's in Birmingham.

About 1500 delegates are expected. Dr Chris Hendriksz leads the BCH team:

'This gives us an opportunity to showcase some of our work and firmly establish our position in the metabolic world as one of the main contenders. We have been planning to host this meeting for three years! Since winning the bid we have already had contact from groups that we have not dealt with before.'

'We are now becoming established as one of the best metabolic centres in the world and our patients in the West Midlands deserve this. It is likely to attract more research and collaboration with our unit. This means more benefit to patients.'

News from

Birmingham Children's Hospital LSD Service

Smart Chip Will Ease Parents' Wait

Dr Paul Gissen, Senior Lecturer and Consultant in Inherited Metabolic Diseases

A new 'biochip' technology will soon be helping scores of families find out if their child has lysosomal storage disease in a fraction of the time more conventional methods take, and it was developed by the researchers at Birmingham Children's Hospital, working as part of a multi-centre team. The development of the "Storage Disease Genechip" uses new microarray technology to allow detection of mutations in the genes causing a number of lysosomal storage disorders. The aim of the researchers is to provide accurate genetic information to the patients and families many times quicker and cheaper than the current techniques can.

This is not only the latest example of the BCH IMD research unit's very close working relationship with the University of Birmingham Department of Medical Genetics and West Midlands Regional Genetics laboratory at the Birmingham Women's Hospital. These links have already led to important discoveries of genetic causes of a number of inherited childhood disorders.

The Unit is developing an active research programme in the field of lysosomal diseases. Recent opening of the Wellcome Trust Clinical Research Facility at BCH provides an excellent environment for the clinical studies on hospital premises. The West Midlands have one of the largest groups of patients with MPS IV (Morquio syndrome) in the world and Birmingham is leading the clinical study into the natural history of the disease with a follow-on trial of a new enzyme replacement therapy.

MPS Regional Specialist Clinics

Cardiff Clinic

9 January 2009

The year started off with a visit to the Cardiff Clinic. The clinic was a very busy one and it was lovely to see old and new faces again. The clinic also gave an opportunity for families to meet up as for some it had been a long time since they last saw each other.

Despite being very busy, the clinic managed to finish on time!

A special mention must go to Gavin Hyde who passed his driving test since the last time I saw him. Gavin said how much he is enjoying driving and the new independence it has given him.

Linda Warner
Roald Dahl Advocacy Officer for Progressive Neurological MPS Diseases

Providing practical support for children with brain, blood and literacy problems

Manchester BMT Clinic

16 January and 23 January 2009

Our first Bone Marrow Transplant (BMT) clinic of 2009, was the under 6 year olds on 16 January. It was lovely to meet all the children and their families, as they were all new to me but made me feel very welcome.

I always enjoy listening to the children as they talk about nursery and school and what they like best whilst there! Painting and playing are still firm favourites!

As the clinic ended the only evidence that children had been there was the empty cartons and snack packets!

The next BMT clinic on 23 January was the over 6 year olds, and this was both very busy and lively! It was lovely to spend time with the children, their siblings and parents and I was made to feel very welcome. As this was an over 6 year old clinic we even had a couple of teenagers and it was very interesting for me to hear about looking towards 6th form or college choices.

As the day progressed more families arrived and it was a great opportunity for them to get together as for some they had not seen each other for so a long time.

Linda Warner *Roald Dahl Advocacy Officer*

Photos this page:
Clockwise from left - Thomas Mett (MPS I), Callum Pollock (MPS I) and Sarah McKnight (MPS I) at the BMT Clinic

Bristol MPS Clinic

24 February 2009

On 24 February 2009 I attended the Bristol Clinic with my colleague Linda Warner. The staff at The Bristol Royal Hospital for Children made me very welcome on my first visit there, particularly as I must have looked slightly alarming carrying random carrier bags filled with all the essential items for our day. I think they must have taken pity on me as they found us a room to hide them all away in. The clinic wasn't a busy one but it was lovely to meet the families that did attend. Just before lunch the hospital fire alarm went off and we had to evacuate to the street. Linda and I would like to thank Dr. Jardine for letting us take refuge in his office in the building opposite while we waited for the all clear. He also makes a great cup of coffee.

On behalf of the Society, Linda and I would like to thank Dr. Jardine, Dr. Wraith, Sally Melson and once again the staff at The Bristol Royal Hospital for Children for all their hard work in making this clinic successful.
Jolanta Turz *Advocacy Support Officer*

Birmingham MPS Clinic

27 February 2009

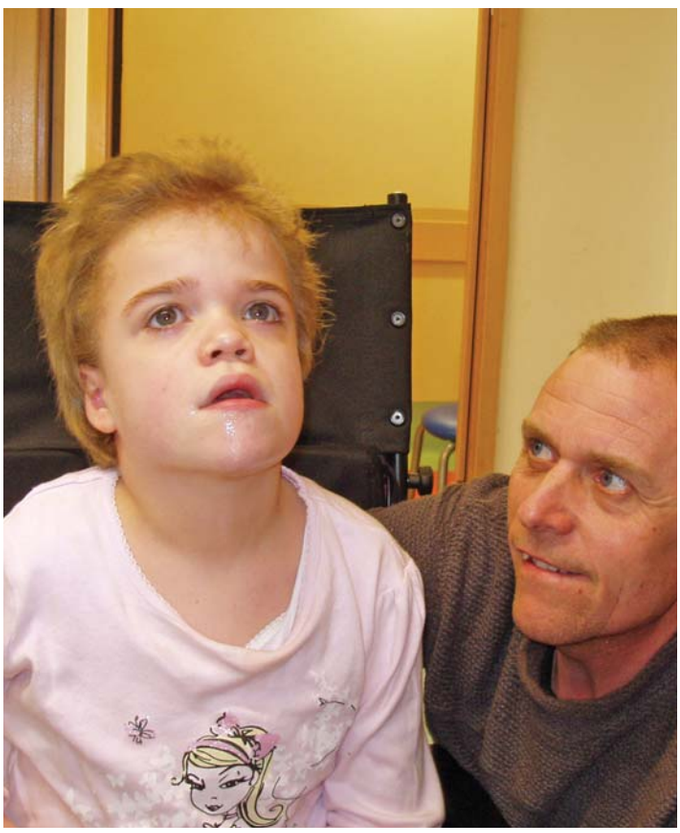
On 27 February I was very excited because I was going for my first clinic to the Birmingham Children's Hospital. Sophie and I were showed the room where we could meet with the families and then it started.

Catherine and Satnam were very busy and they were directing some of you to our room. All families I met were very welcoming and all the children were very lively despite getting up earlier to get to the clinic. Some of you just came to say "Hello!", some of you were collecting their Ollie G wishes.

It a was busy morning clinic but I was able to meet with most of you. I am already looking forward to seeing you again. Thank you for making my first clinic such a lovely experience! **Jolanta Turz** *Advocacy Support Officer*

Do you need help or advice
from the MPS Advocacy Team?
Please phone us on 0845 389 9901
or email
advocacy@mpssociety.co.uk.

Photos this page: Left -Alice Coombes (MPS III) at the Bristol Clinic; Lisa Marie Goodwin (MPS I) at Birmingham MPS Clinic



MPS Awareness Day

Friday 15 May 2009

Each year the Society celebrates MPS Awareness Day on 15 May. This is a day devoted to raising awareness for 23 rare, genetic diseases known as Mucopolysaccharide (MPS) and Related Diseases.

How do I get involved?

Help us spread the word about MPS and raise funds to provide vital services.

Spreading the word...

Use MPS Awareness Day to tell everyone you know about MPS. If you suffer from MPS why not tell your story, talk about your job, or school, your family life, local issues or campaigns that you may be involved in, pursuits you enjoy or sports you are passionate about.

If you can't support MPS yourself, maybe you can approach your place of work. The Society is eager to explore more ways of working with our supporters in the corporate sector.

Raising funds...

Does your workplace, friends, relatives or neighbours support a chosen charity? Why not tell them about MPS? Or are you organising a fundraising event?

Email **fundraising@mpssociety.co.uk** to tell us what you are doing and to order your fundraising pack. Tell your supporters where the money is going and what it will support.

Try to get some coverage in your local media by giving them a call or writing them a letter. Local press like to feature inspirational stories so let them know about your event. Make use of your local amenities, for example, pubs, restaurants and shops as they are great places for promoting awareness. Remember to check whether you need permission from anyone to use their venue.

Ask small companies to donate gifts as they will benefit from the publicity and supporting worthwhile causes.

Do you have your own place in an event and haven't told us or are you still looking for a charity to support? Set up your own fundraising web page. For collection boxes, stickers, balloons and other support materials please contact us. You can also call us for tips on organising an event or advice on how to publicise it locally.

For all activities remember to check the legal insurance requirements and health and safety regulations.

Photo courtesy of The Buckinghamshire Examiner
www.buckinghamshireexaminer.co.uk

MPS Awareness Luncheon at Hampden House

You can help support MPS Awareness Day by ordering some of our promotional goods:

MPS Piggy Bank

MPS Awareness Ribbon

MPS Dog Tags

MPS Engraved Keyring

MPS Bag For Life

MPS Awareness Wrist Band

MPS Trolley Key Ring

To order your MPS Awareness Day promotional goods please complete the reply slip below and return it to the MPS Society.

To make a Donation
I wish to make a general donation of £ .....

Gift Aid
Contributions to charities are eligible for tax relief. This means that if you pay income tax or capital gains tax, and you make a donation during the year, the Society can claim tax on this donation. To make this possible just tick the box and complete this section.

I wish for all contributions I may make to the Society for Mucopolysaccharide Diseases to be treated as Gift Aid donations.

Signature .....

Date ..... Postcode .....

If your circumstances change, please let us know.

To Order Goods

MPS Bag For Life	£4.50	Qty .....
MPS Piggy Bank	£3	Qty .....
MPS Engraved Keyring	£3	Qty .....
MPS Awareness Ribbon	49p	Qty .....
MPS Trolley Key Ring	£1	Qty .....
MPS Awareness Wrist Band	£2	Qty .....
MPS Dog Tags	£2	Qty .....
	TOTAL	**£**

Please send me a free MPS Fundraising Pack YES/NO

Address and payment details

Mr/Mrs/Miss/Other (please delete as appropriate)

Name .....

Address .....

..... Postcode .....

☐ I enclose a cheque/postal order made payable to the MPS Society (UK Sterling only)

☐ I wish to pay/donate by (please delete as appropriate) Mastercard/Visa/Visa Electron/Maestro/Solo

Card No .....

Last three digits on signature strip

Valid From ..... / ..... Expiry Date ..... / .....

Issue No ..... Name on Card .....

Numbers in address .....

Numbers in postcode .....

Manchester MPS Stem Cell Laboratory

The goal of the MPS Stem Cell Research Laboratory in Manchester is to use a multidisciplinary approach to investigate stem cell and gene therapies to improve bone marrow transplantation in MPS and related diseases.

Haematopoietic stem cell therapy (HSCT) or bone marrow transplantation can ameliorate most of the clinical features of MPS IH including neurological symptoms. However, neuronal correction is not observed in MPS IIIA. Neuronal correction relies on bone marrow-derived microglial cells crossing into the brain to produce enzyme as it is difficult for an enzyme administered directly to the blood to cross the blood-brain barrier. In MPS IIIA there may be abnormal stem cell engraftment or microglial trafficking to the brain, or insufficient enzyme dose in the brain. Dr Fiona Wilkinson (post doctoral research scientist) is comparing the level of correction of the disease and cell trafficking in transplanted normal, MPS I and IIIA mouse models using tagged bone marrow donor cells. She is also examining the potential of other bone marrow stem cell subsets, such as mesenchymal stem cells to repopulate other tissues that are difficult to treat in MPS IH such as bones and joints.

Alex Smith (PhD student) is using a MPS IIIA mouse model to investigate a combined stem cell and gene therapy approach to increase the amount of enzyme that is deficient in MPS IIIA. This involves modifying stem cells in the bone marrow to produce a large quantity of enzyme. The modified stem cells are transplanted and will repopulate the immune system. Some cells will traffic to the brain and produce enzyme which can be taken up by other cells which may improve neuronal correction. In a short-term study, Alex has shown that the level of enzyme activity is increased in the liver, spleen and blood of MPS IIIA mice transplanted with gene therapy treated stem cells over levels achieved by transplant of no rmal cells. Long-term experiments are now being set up to determine the level of enzyme activity and storage reduction in the brain.

In MPS IH, bone marrow transplants are not always successful first time. Angharad O'Leary (PhD student) is investigating the mechanisms of cell engraftment in MPS and how to improve stem cell homing and engraftment. In particular she is examining the role of the storage material, heparan sulphate (HS), on migration, homing and engraftment of transplanted bone marrow in a MPS I mouse model. There is no defect in long term engraftment in MPS I, however, the bone marrow microenvironment in MPS I is altered and is affecting transplanted cells. She has found that HS molecules are not only more abundant on the surface of MPS I cells but are structurally altered compared to normal cells. This finding is important because cells communicate through molecules on their surfaces, so alterations mean that signals can be confused, and can have widespread implications throughout the entire body. HS signalling is known to be involved in growth, development and immunity, all of which have implications for the pathology of MPS I. She is currently developing a model to determine the way in which transplanted bone marrow stem cells leave the circulatory system to enter the patient bone marrow by passing through vessel walls.

Kia Langford (research technician and part-time PhD student), is working towards reducing the severity of conditioning required for successful bone marrow transplantation. The risks of bone marrow transplantation are high due to the immune responses between the donor cells and patient cells. Therefore, before receiving a bone marrow transplant a patient will receive a severe treatment called myeloablation, which clears a space for the new donor bone marrow cells, and the immune system is suppressed to prevent rejection of the transplant. The aim here is to find conditioning regimens that are less severe by combining immune blocking antibodies with chemotherapy drugs. Kia is also investigating biomarkers to monitor disease progression and outcome of treatment in MPS patients. Recent work has shown that heparin cofactor II-thrombin complex is a useful biomarker to distinguish MPS I and II patients from unaffected individuals, and that levels of this biomarker decrease with treatment.

Marcelina Malinowska, a visiting PhD student from the group's collaborators in Poland, is investigating the use of a GAG or storage material reducing agent called genistein to improve the symptoms of MPS III subtypes in a mouse model of MPS IIIB. Genistein is a small molecule that blocks the synthesis of the stored material. Around 10% of genistein is able to cross the blood–brain barrier and therefore it has the potential to reduce storage in the brain, the main organ affected in MPS IIIB. So far she has established the most effective and non-toxic dose of genistein which can significantly reduce storage in visceral organs in a MPS IIIB mouse model in a short-term study. She is half way through a long-term study using the optimum dose of genistein, and is measuring the reduction in storage and behavioural changes in treated and non-treated MPS IIIB mice. The behavioural tests allow her to estimate the hind-limb strength, which may be weaker in the MPS IIIB mice as a result of disease progression, as well as differences in activity and motor co-ordination due to accumulation of storage material in the brain.

Characteristics of patients in the MPS I Registry

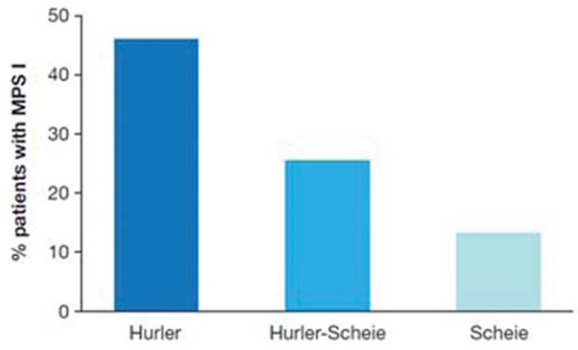
Emma James DPhil (Oxon) – UK Registry Co-ordinator, Genzyme

Mucopolysaccharidosis type I (MPS I) is a life-threatening, inherited disorder, occurring in about 1 in 100,000 births (1, 2). However, MPS I is now treatable, and therapy usually consists of enzyme replacement therapy (ERT) or haematopoietic stem cell transplantation (HSCT) (2).

This article summarises the first published results from the Genzyme-supported MPS I Registry: an ongoing global disease registry for monitoring patients with MPS I (3). Guidelines for treatment are available in the UK (4): these may differ between countries and may be reflected in Registry data. All participating patients have been diagnosed with MPS I (of any severity), they give their consent to participate (via their treating physician), and any information they give is confidential and anonymous. Participating doctors can request analyses of patients within the Registry. All such analyses are reviewed and assessed by a group of specialist physicians who oversee the Registry.

The MPS I Registry programme started to recruit patients in October 2003. Results have been published from the first 302 patients from 24 countries (most live in North America or Europe; 75 are from the UK), and 84 treating physicians from the following specialities: paediatrics (57%), genetics (43%) and/or neurology (40%) (3). The proportion of males (52%) and females (48%) are similar, and the majority (68%) are Caucasian. The current age of patients ranges from under 1 year to 65 years with a median value of 9 years. (A median value is sometimes used instead of an average, and this just means the middle value.) About one-quarter of patients are between 1 and 5 years of age. Most (47%) have Hurler syndrome, 25% have Hurler-Scheie and 13% have Scheie syndromes (Figure 1a). This figure also shows that the median age of patients for each syndrome is lowest for the most severe syndrome (Hurler) and greatest for the least severe syndrome (Scheie) (Figure 1b).

a.



b.

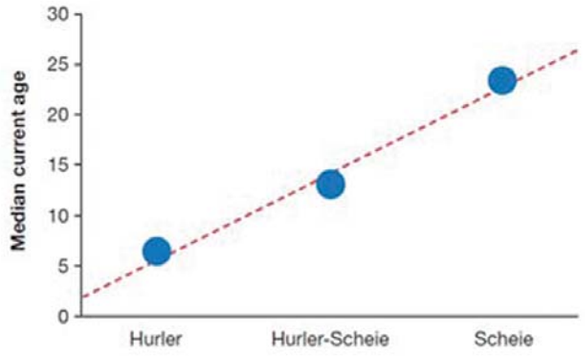


Figure 1. Percentage of individuals in the MPS I Registry with Hurler, Hurler-Scheie, or Scheie (a); median current age for each group (b).

Another important fact to arise from the Registry is that patients with the two milder syndromes (Hurler-Scheie and Scheie) are much more likely to experience a longer delay between the appearance of their first symptoms and a correct diagnosis of MPS I than those with Hurler syndrome (Table 1) (3). This suggests that it may be easier for doctors to diagnose Hurler than Scheie and Hurler-Scheie syndromes and highlights the need for better characterisation of these disorders.

Table 1

MPS I syndrome	Number of patients	Median age at diagnosis (years)	Time from first symptom to diagnosis (years)
Hurler	115	0.8	0.3
Hurler-Scheie	51	3.9	0.9
Scheie	24	9.3	1.5

The MPS I Registry collects results from 29 symptoms of MPS I, and these data have been analysed by patients' age at the onset of MPS I. The following were most common symptoms, occurring in over 70% of patients in all age group (3):
• heart valve abnormalities
• joint contractures (stiff joints, reducing the normal range of joint movement)
• corneal clouding (cloudiness of the usually clear front of the eye, reducing sight)
• hernia.

In contrast, some symptoms are three- to five-times more common in younger age groups, including cognitive impairment (reduced memory, judgement, comprehension, etc.), gibbus/kyphosis (abnormal curve in the spine) and pneumonia.

The Registry has also been useful for recording and analysing the treatment of patients with MPS I. More than half (57%) have been given ERT. The next most frequent treatment is HSCT, which has been used in 16% of patients in the Registry, and involves the transplantation of blood stem cells (e.g. from bone marrow or umbilical cord) (4). This treatment is usually an option for patients with severe MPS I (5). About 1 in 5 patients (21%) are untreated, and the remaining 6% received ERT/HSCT combination therapy. Treatment with ERT occurs across all age groups, but is somewhat more widespread in patients over 5 years of age (61%) than those under 5 years (47%) (3).

This Registry is an ongoing project collecting long-term information on MPS I in both treated and untreated individuals, and the participation of patients is critical to its success. The higher the number of participants – who often have numerous other challenges and commitments on their time – the more comprehensive and useful the information provided by the Registry. As such, the importance of patients to the Registry programme cannot be underestimated. Their contribution, in terms of both time and information, is pivotal to increasing the available evidence, developing medical knowledge, and ultimately improving the diagnosis and treatment of individuals with MPS I.

References

1. 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.
2. Muenzer J, Wraith JE, Clarke LA. Mucopolysaccharidosis I: management and treatment guidelines. Pediatrics 2009;123(1):19-29.
3. 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.
4. Wraith JE, Vellodi A, Cleary MA, Ramaswami U, Lavery C, Jessop M. Guidelines for the investigation and management of Mucopolysaccharidosis type I. http://www.ncg.nhs.uk/documents/lsd_guidelines_for_mucopolysaccharidosis_type_I-010805.pdf Accessed 21 January 2009.
5. Malm G, Gustafsson B, Berglund G et al. Outcome in six children with mucopolysaccharidosis type IH, Hurler syndrome, after haematopoietic stem cell transplantation (HSCT). Acta Paediatr 2008;97(8):1108-12.

Declaration of interest:
The author is employed by Genzyme Therapeutics, Oxford, UK. Date of preparation: February 2009
Code: ALD-UK-2/09-1286

Clinical trials for Enzyme Replacement Therapy for MPS IVA, Morquio Type A

The MorCAP Multinational Clinical Assessment Study (no treatment) of MPS IVA, Morquio Disease Type A, started in October 2008 and is ongoing. MorCAP is open to all UK MPS IVA patients with no age limitations. Through three selected sites in the UK, Great Ormond Street Children's Hospital, London; Birmingham Children's Hospital and the Royal Manchester Children's Hospital, patients are now being enrolled.

The Phase 1/2 Enzyme Replacement Therapy Clinical Trial will begin sometime in the first half of 2009. The endpoints in the Phase 1/2 trial will likely include changes in plasma and urine, pulmonary function and effort based measures.

If you are living in the United Kingdom and you or your child have a diagnosis of MPS IVA, you should talk to your MPS specialist or contact the MPS Society.

Further information about MorCAP is available at www.morquiobmrn.com and www.clinicaltrials.gov/ctz/show/NCT00787995?terrm=morquio&rank=2

Early enzyme treatment best *in Fabry disease*

Clinical cardiology - September 17, 2008
Lisa Nainggolan

Munich, Germany – A new study has found that early enzyme replacement therapy brings the most benefit in patients with Fabry's disease. Dr Frank Weidemann (University Hospital Wurzburg, Germany) presented the results of his prospective study at the European Society of Cardiology Congress 2008.

Specifically, Weidemann and his colleagues found that only patients with no myocardial fibrosis at baseline improved in cardiac morphology, function and exercise capacity following enzyme replacement therapy. Those with mild or severe fibrosis stayed stable but saw no improvement in these parameters with the treatment, he noted.

'The results indicate that the extent of the fibrosis at baseline is crucial,' Weidemann told heartwire. 'No fibrosis equals better outcome. Early treatment is desirable in order to achieve long-term cardiac involvement.' He stressed, however, that this does not mean that those with fibrosis should not be treated. 'They do need treatment because at least you will stabilise them, otherwise they will die, but you cannot bring them 'back' if they already have fibrosis, it's too late.'

Prospective, three year study

Fabry disease, an inherited fat-storage disorder, is caused by an x-linked deficiency of the enzyme alpha-galactosidase A, which is involved in the biodegradation of lipids. It results in the lipid globotriaosylceramide accumulating in the vascular epithelium, heart, kidneys, cornea and other tissues, causing a range of related problems and increasing the risk of early heart attacks and stroke.

Men with the classic form of the disease typically die in early middle age, while those with the cardiac variant of Fabry's, in which residual galactosidase A exists in the system, but at levels less than 10% that of the normal people, frequently experience myocardial dysfunction during middle age. Seven years ago came the first glimmer of hope that supplementing this deficient enzyme in people stricken with this progressive disease might have some impact.

In their Fabry's centre at the University of Wurzburg, Weidemann explained to heartwire: 'We had observed that sometimes people responded to enzyme replacement therapy and sometimes they didn't. It was also known that sometimes people had myocardial fibrosis (around 50% of Fabry's patients have some fibrosis) and sometimes they didn't, but noone knew if this had any impact for the patient. We hypothesised that patients at an earlier stage of the disease, without any fibrosis, would have a better outcome compared with patients at a later stage of the disease.'

They prospectively followed 32 Fabry patients on enzyme replacement therapy, 1.0 mg/kg recombinant agalsidase beta (Fabrazyme, Genzyme) every two weeks for three years, grouping them according to the amount of fibrosis at baseline, detected by cardiac MRI using the late enhancement technique.

Weidemann explained that it is very easy to detect fibrosis in Fabry's patients as 'it is always found in one particular segment of the left ventricle, the posterolateral segment, which is unique to Fabry's disease.' The patients were grouped into those with no fibrosis at baseline (n=12), those with mild fibrosis (n=11) and those with severe fibrosis (n=9).

In those with no fibrosis at baseline, the treatment resulted in a significant reduction in left ventricular mass (238g at baseline vs 202g at three years; $p<0.001$). In those with mild or severe fibrosis there was only a minor reduction in LV hypertrophy at three years.

And in the 'much more important' area of myocardial function, as assessed by strain-rate imaging, 'only those with no fibrosis at baseline improved,' Weidemann said. The same was true of the bicycle stress test, improvement was seen only in those with no fibrosis at baseline, 'while the other two groups stayed stable,' he noted.

Message is to treat early

Weidemann said the message for clinicians treating these patients is that myocardial fibrosis should be assessed at baseline in Fabry disease patients, using MRI with late enhancement, because early treatment is superior to late treatment for long-term improvement of cardiac morphology, function and exercise capacity.

Weidemann said patients with Fabry disease present at different ages. Some come later in life, because they are discovered, by screening or by accident, to already have a problem with some organs, whereas others are younger, discovered via family screening following a diagnosis of Fabry's in another family member. Currently, treatment with enzyme replacement therapy begins whenever a patient begins to show signs of organ damage whether in the heart or other organs, such as the kidney, he explained.

Article appears courtesy of FSIG.
Fabry Support and Information Group
www.fabry.org/FSIG

Update on Amicus Research and Clinical Programs

Fabry Disease

In August, Amicus announced that it had successfully completed an End of Phase 2 meeting for AT1001, a treatment being developed for Fabry Disease. Amicus, along with its partner Shire Human Genetic Therapies, is engaged in ongoing discussions with the FDA and the European Medicines Agency (EMEA) regarding plans for a global Phase 3 clinical development program for AT1001. In line with previous guidance, Amicus expects to complete these interactions in the second half of 2008. Subject to the outcome of the discussions, the Company plans to initiate Phase 3 development of Amigal in the first half of 2009.

New Research Facility

In September Amicus announced that it leased laboratory space for a focused, small-scale research facility in San Diego, CA. The facility will be used to support ongoing research into new applications of the company's platform technology.

'With proof of concept for pharmacological chaperones now established, research at the San Diego facility will support Amicus' efforts to expand our early stage pipeline,' said David Lockhart, PhD., Amicus' chief scientific officer. 'Locating the new laboratory in San Diego allows us to complement and expand our existing research capabilities in Cranbury, NJ, by tapping into the significant resources and expertise of the San Diego science and biotechnology community.'

'We believe pharmacological chaperones have significant potential to treat a broad range of genetic diseases,' added John Crowley, president and CEO of Amicus. 'The focus of our research efforts in San Diego will be to assess new chaperone applications in diseases with high unmet medical needs and larger patient populations particularly in the areas of neurodegenerative and metabolic disorders. This small-scale facility will complement our core research and development activities in New Jersey.'

Article courtesy of Amicus Connection, Winter 2008, Vol 2, Issue 2
www.amicustherapeutics.com

BioMarin appoints new Senior Vice President and Chief Medical Officer

BioMarin Pharmaceutical Inc. have announced the appointment of Dr Henry J Fuchs as Senior Vice President and Chief Medical Officer (CMO). Dr Fuchs replaces longtime BioMarin CMO, Dr Emil Kakkis. The MPS Society has enjoyed working closely with Dr Kakkis over many years and wishes him well in his new venture. On his appointment Dr Fuchs is quoted as saying 'I am excited to join a company founded on great science, with a proven track record abnd a pipeline of important new opportunities.'

We wish Dr Fuchs every success in his appointment as CMO at BioMarin.
Christine Lavery
Chief Executive

Statement from Vivendy Therapeutics Ltd

Following the message that you received from Dr Arndt Rolfs, may I take this opportunity of confirming that Dr Rolfs was relieved of his duties as Medical Director of Vivendy at the end of November, 2008 and as of that time, Dr Rolfs has not been part of any deliberations or discussions within Vivendy. Furthermore, in the event that he may have done so, Dr Rolfs has not been requested to make, should not have made nor was he authorised to make any representations for or on behalf of Vivendy. I regret that Dr Rolfs may have appeared to imply that there is a lack of motivation on the part of Vivendy with regard to developing E6-GALNS, Dr Tomatsu's enzyme replacement treatment for Morquio.

As a result of a strategic review of the development plans for Vivendy's E6-GALNS enzyme, the Company concluded that on the basis of the design and time course of the Natural History Programme (NHP), its continuation was not sufficiently justified. The Company will continue with manufacturing activities, including the development of a primary seed lot, and these are anticipated to be concluded in early 2009. All efforts of the Company are now directed to preserving and protecting the E6-GALNS asset in order to optimise the opportunity for potential further development activities. Management has been mandated by the Board of Directors to seek and pursue those strategic initiatives focused on third party participation in, or assumption of, further development activities.

We remain encouraged by the data that has been generated by Dr Tomatsu and his team at St Louis University, however our recent decisions are based on what we believe to be a realistic assessment of today's competitive, regulatory and intellectual property environment of the E6-GALNS enzyme. As we move forward we remain committed to trying to further an ERT treatment for Morquio patients while keeping the interests of patients and shareholders/investors aligned.

Gosse Bruinsma MD
Vivendy Therapeutics Ltd

WORLD Symposium 2009

The WORLD Symposium, 18 - 20 February 2009, in San Diego, California, was co-presented by the Lysosomal Disease Network and the National Institute of Neurological Disorders and Stroke. WORLD's primary aim is to raise awareness of Lysosomal Storage Diseases, promote the funding of research into Lysosomal Storage Diseases and disseminate research findings through an annual Symposium.

The keynote address was given by Prof Elizabeth Neufeld best known as an authority on human genetic diseases. She received her PhD in comparative biochemistry and studied cell division in sea urchins. Later, as a junior research biochemist she studied the biosynthesis of plant cell wall polymers – which would prove significant when Prof Neufeld began studying the Mucopolysaccharide diseases. Prof Neufeld's research opened the way for prenatal diagnosis of Hurler disease that translated as a benefit for the other Lysosomal Storage Diseases.

As well as the Symposium being well attended and stimulating, the mealtimes were taken up with a number of sub meetings that included an update on BioMarin's MorCap Natural History Study for MPS IVA and meeting of American and Worldwide LSD patient Organisation representatives. **Christine Lavery** Chief Executive

Announcing Expression of Hope II: Inspiration through Art

In collaboration with patient organisations around the world, Genzyme is proud to bring you Expression of Hope II: Inspiration through Art, a global program featuring works of art by the community touched by lysosomal storage disorders (LSDs). People living with LSDs and their caregivers are invited to submit artwork which will be shared with the worldwide community via the Web and a travelling art show. Our vision is that those who experience this art will be inspired and moved by the powerful expressions of the human spirit which the pieces will reveal.

Free! The Expression of Hope Book
Everyone who submits a photo of their art will receive a beautiful book with selected images from the travelling art show.

It's easy to take part:
1) Create your art
2) Photograph it
3) Submit the photo

Visit **expressionofhope.com** to learn more.

With support from patient organisations around the world, **Genzyme** is proud to sponsor this program.

On behalf of the Board of Directors and the members of the National MPS Society, I want to thank you and your Trustees for sponsoring my trip to the New Zealand consensus meeting on Biophosphonate Therapy in Oligosaccharides. The presentations were most interesting and sparked spirited discussions. I came away from those two days with new appreciation of the complexity of this treatment.

I will be posting the consensus statement on our website after it is published in the Journal of Inherited Metabolic Disease (JIMB) and sharing it with families who have children with an oligosaccharide disease.

I commend the MPS Society for helping to sponsor meetings such as this. Bringing together professionals to discuss the efficacy of treatments provides a direct and very important service to our member families.

Barbara Wedehase Executive Director

National MPS Society
www.mpssociety.org

MPS Spain, the Spanish MPS Society, has recently announced that it is now providing **support to patients with Fabry disease.**

Here at the UK MPS Society we believe this is very good news and wish MPS Spain every success. **www.mpsesp.org**

BRAINS for BRAIN

European Task Force on Brain and Neurodegenerative Lysosomal Storage Diseases

Third European Workshop, Frankfurt, Germany
March 6th-8th 2009

This meeting of 80 invited scientists, clinicians and patient organisation representatives throughout Europe met for two and a half days to discuss the challenges and developments to getting therapeutic enzyme into the brain for the Lysosomal Storage Diseases (LSDs). The lysosomal enzymes are ubiquitous molecules and their deficiency has important effects in all organs, in particular the central nervous system (CNS), liver, spleen, heart and bones.

With the advent of recombinant DNA technology, the identification and cloning of all the known lysosomal enzymes has been recently achieved, and therefore, expression and purification of recombinant proteins is now possible and enzyme replacement therapy (ERT) is now available for a growing number of storage disorders.

However, although ERT has proven to be valuable to possibly change the clinical history of the disease it has been evident that the recombinant proteins do not have any effect on the CNS, as they are unable to cross the blood brain barrier. Furthermore, the mechanisms and etiology of CNS pathology in LSDs are still poorly understood.

We still do not know whether storage and accumulation of mucopolysaccharide is really the "primum movens" of the metabolic disaster or whether other processes might be more important (inflammation, alteration of ion channel activity, lack of chaperone molecules etc.). The understanding of these basic aspects might be extremely valuable to unravel why most of the LSDs have an attenuated and a severe form without and with CNS involvement, respectively, depending on whether there is a total enzymatic deficiency or not.

The Brains for Brain Task Force

The task force takes advantage of the expertise of the most distinguished European scientists, leaders in basic and applied neurotechnology and neurology grouped together to create a co-ordinated effort toward the comprehension of the pathophysiological processes of the neurological disorders, the implementation of knowledge on the blood brain barrier and the development of new molecular and/or biochemical strategies to overcome the blood brain barrier and treat neurological disorders.

Brains For Brain (B4B) was formally founded in March 2007 as a research group formed by international specialists and leaders on clinical and basic research in the field of neuro-paediatrics and neuroscience. The group has attracted interest from major biotech companies working on the development of new therapeutical strategies for lysosomal diseases, and furthermore has a strong interaction with international patients' associations involved in taking care of the needs of lysosomal patients, and has stimulated collaborations toward coordinate actions to disseminate knowledge about the diseases.

B4B is also collaborating with International Scientific Associations, such as the European Study Group for Lysosomal Diseases (ESGLD) and the International Blood Brain Barriers Society (IBBS) and it is a member of the European Brain Council.

B4B members have participated to the submission for scientific proposals to the European Union in the FP7–Health 2008 programme and have presented projects related to the Understanding of the Blood Brain Barrier and in the context of the Rare Neurological Diseases. We are all waiting with fingers and toes crossed for the outcome of the B4B 7th Framework grant application made by B4B last December.

The Brains for Brain Foundation

The BRAINS FOR BRAIN FOUNDATION is a no-profit international organisation addressed to disabled children who are affected (or healthy carriers) by rare neurological diseases.

The purposes of the FOUNDATION are :
- scientific research
- dissemination of knowledge
- social and socio-medical assistance
- health assistance

Aims of the Workshop

The aims of this workshop were:
1) to discuss research achievements at clinical and basic science level in the field of neurodegenerative lysosomal storage disorders and Blood Brain Barrier;
2) to discuss how B4B might collaborate with the European Union to stimulate interest in the research on LSDs and BBB. For this reason representatives from EU Commission were invited;
3) to discuss with international patient associations ways to co-operate and collaborate to increase knowledge about lysosomal storage diseases and research projects.

This year, B4B opened the Workshop to young scientists and I am delighted that the MPS Society and Gaucher Association were able to support a young scientist from Dr Fran Platt's Research Laboratory in Oxford.

Despite some of the science of the presentations going over my head, I gained a terrific amount of knowledge from the weekend. As well as the amazing opportunity to network, I was able to meet with two MPS Grant holders, Dr David Begley and Dr Brian Bigger; discuss specific clinical questions with European experts and feedback to families; and discuss with fellow patient organisations strategies for working with LSD Groups throughout Europe.
Christine Lavery Chief Executive

Family Experience of Pamidronate

By Sally Motomura, mother of Tetsuya Motomura (ML III)
Presented in Christchurch, New Zealand, Nov 2008

We are an international family and never suspected the possibility of an inherited disease in our children. The first two boys were and are healthy, each dealing in his own way with his bicultural background and frequent international moves.

We expected that our third son, Tetsuya would follow suit. He was born in Paris, in 1984, one month early. He arrived before our household goods which had been delayed en route from Sri Lanka, and spent the first few weeks sleeping in a plastic drawer.

A little behind in his milestones, he surprised the peadiatrician when his growth slowed towards his first birthday. Kyphosis had also developed and his knee and hip joints were stiffening. Store-bought baby clothes didn't seem to quite fit. We heard some strange theories, treatments and advice, including a suspected link between my pupil dilation rate, my scoliosis, a suggestion we could 'fix' his knees with surgery and therapy involving little glass 'ampules' of base minerals that had to be snapped open.

Struggling with interpreters, I had a frantic run-around to various Paris hospitals and specialists, and we ended up with Dr Pierre Maroteaux in his offices in the Lamy Medical Research Building at the Paris Hospital for Sick Children. It took six months until the diagnosis of Mucolipidosis (MLIII) was finally confirmed, when Tetsuya was 18 months old. My husband Yuki, had made enquiries in Japan. Professor Orii of Gifu University had mentioned the British MPS Society, and Christine Lavery's name came up. By weird coincidence, Christine and I already knew one another, but that's a long story.

It was wonderful at last to have someone to offer her experience in dealing with the diagnosis. Just to be sure, we had the tests run again in London at Great Ormond Street Children's Hospital by Dr Rosemary Stephens, who confirmed the ML III diagnosis. Ironically, when we returned to Japan soon afterwards, Professor Orii insisted on running his own tests, as he felt Tetsuya wasn't typical of ML III. It has since been clarified to me by Dr Sara Cathey, that his type was a subgroup of ML III alpha/beta, thought of as long survivors of ML II (Mucolipidosis II).

From the time of diagnosis onward for all his 23 years, Tetsuya's height remained almost static, below 90 cm. One of his orthopedic specialists remarked that there was almost nothing normal about his bones. However, his kyphoscoliosis did not progress into any of the spinal complications often seen in ML III, and he was lucky enough not to need any major surgeries.

Despite his extreme short stature, ongoing chest infections and physical difficulties, Tetsuya thrived at nursery school in Tokyo, elementary schools in New York and Vienna. He always needed help getting around, with personal care and with manual tasks. When 10 years old, Tetsuya underwent bilateral myringotomy, which greatly improved his communication skills and general health.

Austria was his happiest period. He attended a small international school in Vienna, was in regular grade 2 and 3 classrooms with his cousin as a full-time aide. He was included with a gang of boisterous children and took part in the school play. However, he was hospitalised a few times with pneumonia, after which air travel involved special arrangements for oxygen in case of need.

At age 14, Tetsuya returned with us to Tokyo. His cardiopulmonary symptoms worsened, and he was hospitalised a few more times with pneumonia. He started taking diuretics (lasix) to ease the strain on his heart, and soon needed a beta blocker as well (seroken, then metoprolol). His stamina was poor and he had to pace himself with day-time naps and reduced academic expectations. He was enrolled at a tiny international school for children with special needs on the outskirts of Tokyo. It involved a long bus ride and he missed the comraderie of normal school life. Though he always maintained a cheerful outlook, it seemed that everything in his life was becoming a struggle.

On the positive side, Tetsuya's reading ability finally took off and he derived much pleasure there. He also started writing his own stories. A great boost to his development came from the computer, with which he became quite proficient over the years. He gained skills for private study, and it opened up the world for him. He also acquired a motorised wheelchair, and proved to be an excellent driver.

Our next posting was to Manhattan, when Tetsuya was 17. This time no international schools would admit him, but he was welcomed into the New York City Schools system and offered all the advantages of their special services, for which we were extremely grateful. However, with his low stamina, he had a scaled back timetable and was missing a lot of school due to chest infections. During this time, in addition to MPS meets, we took him to several Little People of America conventions, which he loved. He also obtained his amateur radio license.

His cardiologist suggested a sleep study, which revealed severe sleep apnea. At first we were reluctant to accept that he should sleep with CPAP, but once he got used to it, the change in his stamina and overall health was amazing, dramatic. In a short time he regained his vigor, his daytime breathing problems lessened, and he was able to take on more at school. It was like a 4-year jump back to his health in Vienna days, though his joint stiffness continued to progress. A major drawback for CPAP and oxygen users is the question of its use during long distance air travel. Economy class travel becomes impossible, though for first and business class passengers, permission is sometimes reluctantly granted.

Our return to Japan when he was 20 coincided with the advent of ISMRD and the online penguin cafe, in which Tetsuya really enjoyed participating. He continued his high school studies based on his computer at home, and graduated a year later. Through ISMRD and also a Korean doctor who lectured at a Japanese MPS Society meeting in Tokyo, we heard of Pamidronate therapy.

Tetsuya had never really complained of joint pain, although when asked, he said he had a headache every day. He was very keen to try the therapy, after reading about how it had changed the lives of other ML III sufferers. Our positive experiences with myringotomy and CPAP having boosted his general health, we decided it was worth a try. At this point, Yuki was in remission after major cancer surgery and we were expecting another international move, but we decided to go ahead and at least start the pamidronate treatment to see how things went.

Tetsuya's primary physician was fairly easily persuaded, although he warned that no-one he knew of in Japan had tried the treatment on ML patients. He agreed that we could start with a low dose, to be gradually increased. Tetsuya was willing to be a guinea pig, despite knowing of the initial side effects. We gathered as much information as we could, thanks mainly to Jenny Noble and awaited the first infusion date, Feb 2005.

When we were about to proceed, the doctor in charge of the treatment told me that OF COURSE HE'D BE USING THE FULL DOSE. He had no information about the recommended gradual increase from a small dose (only the full dose was described in the paper he'd researched on the internet 'Bisphosphonate Therapy in Mucolipidoses, Advantages and Disadvantages', by David Sillence MD FRACP 2003/4). He did change his plan when I made a song-and-dance about it, after checking with the primary physician. One third of the full dose was used for the first, two thirds for the second infusions, which I know is still high. It became clear after the first two infusions that Tetsuya's breathing was more laboured and his heart had enlarged, so we cancelled further treatment. Thankfully, his condition bounced back to pre-treatment levels soon after the infusions were stopped. When I asked a few months later whether the doctors had reported our case, I was told that the process was stopped before enough data could be gathered, and therefore it didn't become a proper study.

It is not clear whether the negative outcome of our pamidronate experience was caused by the protocols glitch. The impression is that Tetsuya's basic heart condition was probably already too far advanced for the pamidronate to have a beneficial effect. He had already been using CPAP (when sleeping) for almost four years. His condition did deteriorate gradually and he died from heart failure two years after the first infusion, though this was not apparently related to the effects of the pamidronate treatment. Since his autopsy the medical team has been researching various aspects of Tetsuya's condition, which I hope will be shared in due course with other researchers.

One of Tetsuya's dreams was to write fantasy novels. He also wished he could dance with his brother, Kazuma, who has since choreographed "Destiny in the Wind" in his honour, based on Tetsuya's ideas while listening to a favourite piece of music.

Something For You

Way far
in a land that 'time'
is a forgotten word,
was a green forest
now overgrown with age.

Sun shines through the small stream
as if it was
a stained glass window
of a church long abandoned.

A busy beaver
with never-a-time-to-spare
was making his dam with his family,
winter coming fast
every moment counted.

Their sleek bodies
gliding in the water with triangle lines,
then vanished without a splash.

In a hanging willow tree
it's branches making lines in the water,
a king fisher eyeing the stream
for a sign of movement.

A fish splashed
the bird dived.
it was over in the instant
like a dream always on replay.

Tetsuya Motomura

Introduction to Children's Hospices UK

Children's Hospices UK
Together we make a lifetime of difference

Children's hospices exist to provide specialist care and support for children with life-limiting conditions – and their families – from the moment of diagnosis onwards. They provide a range of care and services within their teams, under one roof, in a friendly home from home environment. All services are free.

In a purpose built hospice, or in your own home, highly trained staff will be able to help your child and the rest of the family with the medical and emotional challenges that having a life-limiting condition can bring over many months and years.

Children's hospices provide specialist respite care: regular short stays for the child or for the whole family together. They also provide emergency and end of life care; specialist advice and expertise; practical help and information; and 24 hour telephone support.

Above all, they are positive places, helping children and families to make the most of life. Children's hospices are committed to working with families from all faiths, cultures and ethnic backgrounds and fully respect the importance of religious customs and cultural needs that are essential to the daily lives of each family.

Children's Hospices UK recently published a guide written for parents who have children with life-limiting conditions.

Children with a wide range of conditions or illnesses could use a children's hospice. Usually these are children defined as having life-limiting or life-threatening conditions or potentially life-threatening disabilities.

How can a children's hospice help a child with a life-limiting condition?
Specialist staff can help a child with the medical, emotional and physical impact their condition can cause, supporting and helping them to make the most of life. Children's hospices put the child and family at the centre of everything they do. All care provided is based around meeting a child and family's needs and trying to follow and maintain a daily home routine.

What help is there for families?
Families caring for a child with a life-limiting condition – often 24 hours a day, over many years – can feel extremely isolated and under huge emotional, physical and financial strain. Children's hospices are there to provide care, help, support and advice for all family members, for as long as it is needed.

Children's hospices help to give families a rest from the demands of caring by welcoming the child or the whole family for regular stays. Parents will often use this free time to do things together, or with brothers and sisters of the child, that they would not normally be able to do.

For how long can a children's hospice offer support?
Children's hospices do not just provide care for a child when they are in the last stages of their life, but offer care and support for a child and their family for as long as it is needed. Sometimes this can mean from diagnosis onwards and sometimes children and their families have contact with their local children's hospice service over many years. During this time, they form friendships and relationships with staff, volunteers and other families, which can help during difficult times. Many children, parents, brothers and sisters may be facing similar experiences and the children's hospice provides an opportunity to meet with other people who truly understand what you are going through.

Are there many children's hospices?
The first children's hospice in the world, Helen House, opened in Oxford in 1982 and since that time the children's hospice movement has grown throughout the UK and internationally.

There are now 41 children's hospice services open across the UK, providing friendship, care and medical expertise for the whole family, with a further five at project stage. You can find these listed at www.childhospice.org.uk. All of these children's hospice services are members of the Children's Hospices UK.

Will we have to pay to use a children's hospice?
Children's hospice services are provided free to children and families. Each service is a charity that relies on donations and fundraising by the general public, organisations and companies to continue providing care.

MPS Awareness Day
Friday 15 May 2009

How do we find out if we can use a children's hospice?
Speak to your GP, community nurse, health visitor, social worker or hospital consultant/nursing staff about whether your child can be helped.

How do we get referred?
A children's hospice service may receive a request to help a child from a wide range of people – from parents, family members, friends, or from experts such as doctors, nurses or health visitors – in fact from anyone who knows the child. Children's hospice staff acknowledge the expertise that parents have as the main carers of their child and will work in partnership with them. The process of asking for and then gaining support varies from area to area.

Families should contact their local children's hospice if they are unsure of what to do. Most children's hospices now have a catchment area with children normally using their nearest service.

What is Children's Hospices UK?
Children's Hospices UK is the national voice for children's hospice services and the children and families they care for. We strive to ensure that the estimated 20,000 life-limited children in the UK, and their families, have access to the unique range of services children's hospices provide. Our work includes public and professional awareness raising, fundraising on behalf of all children's hospices and service development through education, training, research and lobbying. We are a registered charity which relies on public support to fund our work.

Children's Hospices UK
Second Floor, Canningford House,
38 Victoria Street,
Bristol
BS1 6BY
Tel: 0117 9897820
info@childhospice.org.uk
www.childhospice.org.uk

Article appears courtesy of Children's Hospices UK and is adapted from their Children's Hospice Services: A Guide for Parents.

Do you need help or advice
from the MPS Advocacy Team?
Please phone us on **0845 389 9901**
or email **advocacy@mpssociety.co.uk**

Family Fund
Helping Disabled Children

Age Limit increased
Thanks to new money from the four UK governments (including £8.4m in England), the Family Fund has extended its age limit to help severely disabled young people aged under 18 in England and Wales and those under 17 in Northern Ireland and Scotland. Before, we could only afford to help those under 16.

We want to give 16- and 17- year-olds applying for the first time the things that make the most difference to them. So while families can apply for the same things as in the past, such as washing machines, holidays and driving lessons, we would welcome other requests that have particular meaning for this age group.

Family Fund *extra* launched
A new online buying club called Family Fund *extra* means that all families with a disabled child can get discounts of up to 25% from leading online retailers including Comet, Argos, BSM, Haven Holidays and Stone Computers. *extra* is free to join and will help families make their money go further.

Family Fund *extra* is part of the Family Fund, which provides grants to nearly 50,000 low income families with severely disabled children a year to meet their additional needs. All monies raised by Family Fund *extra* is gifted to the Family Fund to enable them to extend their reach to more families.

For more information about grants for the severely disabled 16- and 17- year olds, please call us on 01904 550037 or visit www.familyfund.org.uk. For more information about Family Fund *extra*, please visit www.familyfundextra.org.uk.

WANTED!

Adventurous people throughout the UK to do something like this...

for

FREE!

Minimum sponsorship required

Help us raise funds by making an exhilarating 10,000 feet freefall parachute jump. No experience is required and if you raise the minimum amount of sponsorship you will get to jump for FREE!

Phone **0845 389 9901**
or email **fundraising@mpssociety.co.uk**